Gene	Diseases	Omim	Model Of Inheritance	Level4 disorder sub-category 100,000 Genomes Project	Level3 disorder sub-category	Level2 disorder category	version
AAAS	Achalasia-addisonianism-alacrimia syndrome, OMIM:231550;Triple-A syndrome, MONDO:0009279		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AARS	Developmental and epileptic encephalopathy 29, OMIM:616339;Developmental and epileptic encephalopathy, 29, MONDO:0014593		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AASS	Hyperlysinemia, OMIM:238700;Hyperlysinemia (disease), MONDO:0009388		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABAT	GABA-transaminase deficiency, 613163;developmental delay		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABCA2	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, OMIM:618808;Intellectual developmental disorder with poor growth and with or without seizures or ataxia, MONDO:0032930		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABCC9	Intellectual disability and myopathy syndrome, OMIM:619719;intellectual disability and myopathy syndrome, MONDO:0859224		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABCD1	Adrenoleukodystrophy, 300100; Adrenomyeloneuropathy, adult, 300100;ADRENOLEUKODYSTROPHY, X-LINKED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857;METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, CBLJ TYPE; MAHCJ		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABHD5	Chanarin-Dorfman syndrome, OMIM:275630		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACACA	Acetyl-CoA carboxylase deficiency, OMIM: 613933		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACAD9	ACAD9 deficiency, 611126;ACYL-COA DEHYDROGENASE FAMILY MEMBER TYPE 9 DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACADM	MEDIUM CHAIN ACYL-COENZYME A DEHYDROGENASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of 201470		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACBD6	Neurodevelopmental disorder with progressive movement abnormalities, OMIM:620785		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACO2	Infantile cerebellar-retinal degeneration, OMIM:614559;Infantile cerebellar-retinal degeneration, MONDO:0013802		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, OMIM:264470;Mitchell syndrome, OMIM:618960		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACSL4	Mental retardation, X-linked 63, OMIM:300387;Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome, MONDO:0010263		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACTB	Dystonia, juvenile-onset, 607371Baraitser-Winter syndrome 1, 243310;BARAITSER-WINTER SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACTG1	Deafness, autosomal dominant 20/26, 604717Baraitser-Winter syndrome 2, 614583;BARAITSER-WINTER SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACY1	Aminoacylase 1 deficiency, 609924;AMINOACYLASE-1 DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ADAR	Aicardi-Goutieres syndrome 6, OMIM:615010;Dyschromatosis symmetrica hereditaria, OMIM:127400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ADARB1	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ADD1	Global developmental delay;Intellectual disability;Seizures;Ventriculomegaly;Abnormality of the corpus callosum		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ADK	Hypermethioninemia due to adenosine kinase deficiency 614300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ADNP	MENTAL RETARDATION, AUTOSOMAL DOMINANT, 28; MRD28		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ADSL	ade(-)I bifunctional Adenylosuccinase deficiency, 103050;ADENYLOSUCCINASE DEFICIENCY (ADSL DEFICIENCY)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AFF2	Intellectual developmental disorder, X-linked 109, OMIM:309548;FRAXE intellectual disability, MONDO:0010659		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AFF3	KINSSHIP syndrome, OMIM:619297		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AGA	Aspartylglucosaminuria, 208400;ASPARTYLGLUCOSAMINURIA (AGU)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AGO1	Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, OMIM:620292		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AGO2	Intellectual disability;regression;seizures		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AHI1	Joubert syndrome 3 608629		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AIFM1	Cowchock syndrome, OMIM:310490;Combined oxidative phosphorylation deficiency 6, OMIM:300816;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, OMIM:300232		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AIMP1	Leukodystrophy, hypomyelinating, 3, OMIM:260600		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, OMIM:615937		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALDH18A1	Cutis laxa, autosomal recessive, type IIIA, 219150;SPASTIC PARAPLEGIA 9, AUTOSOMAL DOMINANT; SPG9		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALDH3A2	Sjogren-Larsson syndrome, OMIM:270200		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALDH4A1	Hyperprolinemia, type II, 239510;HYPERPROLINEMIA TYPE 2 (HP-2)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency OMIM:271980;succinic semialdehyde dehydrogenase deficiency MONDO:0010083		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALDH7A1	PYRIDOXINE-DEPENDENT EPILEPSY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALG1	Congenital disorder of glycosylation, type Ik, 608540;ALG1-CDG (CDG-IK)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALG11	Congenital disorder of glycosylation, type Ip 613661;ALG11-CDG (CDG-IP)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALG12	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1G (CDG1G)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALG13	EPILEPTIC ENCEPHALOPATHIES.		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALG3	Congenital disorder of glycosylation, type Id, 601110;ALG3-CDG (CDG-ID)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALG6	Congenital disorder of glycosylation, type Ic, 603147;ALG6-CDG (CDG-IC)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALG8	Congenital disorder of glycosylation, type Ih, 608104;ALG8-CDG (CDG-IH)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALG9	Developmental delay;Congenital disorder of glycosylation, type II 608776;Gillessen-Kaesbach-Nishimura syndrome 263210		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALMS1	ALSTROM SYNDROME, 203800		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AMER1	Osteopathia striata with cranial sclerosis 300373		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AMT	Glycine encephalopathy, 605899;GLYCINE ENCEPHALOPATHY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ANK2	Complex neurodevelopmental disorder, MONDO:0100038		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ANK3	Intellectual developmental disorder, autosomal recessive 37, OMIM:615493;intellectual disability-hypotonia-spasticity-sleep disorder syndrome, MONDO:0014210		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ANKRD11	KBG syndrome, 148050;KBG SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AP1S1	MEDNIK syndrome, 609313;MEDNIK syndrome;mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis and keratoderma syndrome		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AP1S2	Pettigrew syndrome, OMIM:304340		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AP3B1	Hermansky-Pudlak syndrome 2 608233		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AP3B2	Epileptic encephalopathy, early infantile, 48 617276		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AP4B1	Spastic paraplegia 47, autosomal recessive, OMIM:614066;Hereditary spastic paraplegia 47, MONDO:0013551		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AP4E1	Spastic paraplegia 51, autosomal recessive, OMIM:613744;Hereditary spastic paraplegia 51, MONDO:0013401		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AP4M1	Spastic paraplegia 50, autosomal recessive, 612936;CEREBRAL PALSY SPASTIC QUADRIPLEGIC TYPE 3 (CPSQ3)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AP4S1	Spastic paraplegia 52, autosomal recessive, 614067;CEREBRAL PALSY SPASTIC QUADRIPLEGIC TYPE 6		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARFGEF2	Periventricular heterotopia with microcephaly, 608097;PERIVENTRICULAR HETEROTOPIA WITH MICROCEPHALY□		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARG1	Argininemia, OMIM:207800		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARHGEF9	Developmental and epileptic encephalopathy 8, OMIM:300607		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARID1A	Mental retardation, autosomal dominant 14, 614607;COFFIN-SIRIS SYNDROME; CSS		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARID1B	Mental retardation, autosomal dominant 12, 614562;COFFIN SIRIS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARL13B	Joubert syndrome 8, 612291;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARL6	Bardet-Biedl syndrome 3, 209900{Bardet-Biedl syndrome 1, modifier of}, 209900Retinitis pigmentosa 55, 613575;BARDET-BIEDL SYNDROME TYPE 3 (BBS3)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARSA	ARYLSULFATASE A DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARSB	MUCOPOLYSACCHARIDOSIS TYPE 6 (MPS6)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARSE	CHONDRODYSPLASIA PUNCTATA 1, X-LINKED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARX	Epileptic encephalopathy, early infantile, 1, 308350Lissencephaly, X-linked 2, 300215Mental retardation, X-linked 29 and others, 300419Proud syndrome, 300004Partington syndrome, 309510Hydranencephaly with abnormal genitalia, 300215;MENTAL RETARDATION X-LINKED ARX-RELATED (MRXARX)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ASAH1	FARBER LIPOGRANULOMATOSIS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ASL	Argininosuccinic aciduria, OMIM:207900;argininosuccinic aciduria, MONDO:0008815		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ASNS	Asparagine synthetase deficiency, 615574;congenital microcephaly, intellectual disability, progressive cerebral atrophy, intractable seizures		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ASPA	Canavan disease, 271900;CANAVAN DISEASE		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ASPM	Microcephaly 5, primary, autosomal recessive, OMIM:608716		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ASS1	Citrullinemia, 215700;intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ASXL1	Bohring-Opitz syndrome, 605039Myelodysplastic syndrome, somatic, 614286;BOHRING-OPITZ SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATG7	Spinocerebellar ataxia, autosomal recessive 31, OMIM:619422		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688;AICA-RIBOSURIA (AICAR)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATM	Ataxia-telangiectasia, OMIM:208900		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATN1	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, OMIM:618494		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP13A2	PARKINSON DISEASE 9		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP1A2	Migraine, familial hemiplegic, 2 602481;Alternating hemiplegia of childhood 1, 104290		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP1A3	Alternating Hemiplegia of Childhood (AHC), intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP6AP2	Mental retardation, X-linked, syndromic, Hedera type, 300423;MENTAL RETARDATION X-LINKED WITH EPILEPSY;X-linked intellectual disability, Hedera type		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP6V0A1	ATP6V0A1-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA 219200; Wrinkly skin syndrome 278250		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP7A	Menkes disease, 309400Occipital horn syndrome, 304150Spinal muscular atrophy, distal, X-linked 3, 300489;OCCIPITAL HORN SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP8A2	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4 OMIM:615268;cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4 MONDO:0014104		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATR	Seckel syndrome 1, OMIM:210600		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATRX	Alpha-thalassemia/mental retardation syndrome, 301040;Mental retardation-hypotonic facies syndrome, X-linked, 309580		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AUH	3-methylglutaconic aciduria, type I, 250950;3-METHYLGLUTACONIC ACIDURIA TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AUTS2	SYNDROMIC INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
B4GALT7	Ehlers-Danlos syndrome, progeroid type, 1, 130070;EHLERS-DANLOS SYNDROME PROGEROID TYPE (EDSP)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
B9D2	Joubert syndrome 34, OMIM:614175;Meckel syndrome 10, OMIM:614175;Meckel syndrome, type 10, MONDO:0013609		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BAP1	Kury-Isidor syndrome, OMIM:619762		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BBS1	Bardet-Biedl syndrome 1 OMIM:209900;Bardet-Biedl syndrome 1 MONDO:0008854		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BBS10	Bardet-Biedl syndrome 10, 209900;BARDET-BIEDL SYNDROME TYPE 10 (BBS10)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BBS12	Bardet-Biedl syndrome 12, 209900;BARDET-BIEDL SYNDROME TYPE 12 (BBS12)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BBS2	Bardet-Biedl syndrome 2, 209900;BARDET-BIEDL SYNDROME TYPE 2 (BBS2)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BBS4	Bardet-Biedl syndrome 4, 209900;BARDET-BIEDL SYNDROME TYPE 4 (BBS4)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BBS5	Bardet-Biedl syndrome 5, 209900;BARDET-BIEDL SYNDROME TYPE 5 (BBS5)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BBS7	Bardet-Biedl syndrome 7, 209900;BARDET-BIEDL SYNDROME TYPE 7 (BBS7)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BBS9	Bardet-Biedl syndrome 9, 209900;BARDET-BIEDL SYNDROME TYPE 9 (BBS9)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BCAP31	DEAFNESS, DYSTONIA, AND CENTRAL HYPOMYELINATION WITH DISORGANIZATION OF THE GOLGI APPARATUS		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BCKDHA	Maple syrup urine disease, type Ia, 248600;MAPLE SYRUP URINE DISEASE		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BCKDHB	Maple syrup urine disease, type Ib, 248600;MAPLE SYRUP URINE DISEASE		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BCL11A	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BCOR	Microphthalmia, syndromic 2, 300166;MICROPHTHALMIA SYNDROMIC TYPE 2 (MCOPS2)		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BCS1L	Mitochondrial complex III deficiency, nuclear type 1, 124000;Leigh syndrome, 256000;Bjornstad syndrome, 262000		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BLM	Bloom syndrome, OMIM:210900		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BMP4	Microphthalmia, syndromic 6 607932;Global developmental delay		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BOLA3	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 2		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BRAF	Melanoma, malignant, somaticColorectal cancer, somaticAdenocarcinoma of lung, somatic, 211980Nonsmall cell lung cancer, somaticCardiofaciocutaneous syndrome, 115150Noonan syndrome 7, 613706LEOPARD syndrome 3, 613707;NOONAN SYNDROME TYPE 7 (NS7)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BRAT1	LETHAL NEONATAL RIGIDITY AND SEIZURE SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BRSK2	Global developmental delay;Autism;Behavioral abnormality;Global developmental delay, Intellectual disability, Autism, Behavioral abnormality;Intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BRWD3	Mental Retardation, X-linked;Mental retardation, X-linked 93, 300659;MENTAL RETARDATION X-LINKED TYPE 93 (MRX93)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BSCL2	Encephalopathy, progressive, with or without lipodystrophy, OMIM:615924;Lipodystrophy, congenital generalized, type 2, OMIM:269700		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BTD	Biotinidase deficiency, 253260;BIOTINIDASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BUB1B	Colorectal cancer, somatic, 114500Mosaic variegated aneuploidy syndrome 1, 257300[Premature chromatid separation trait], 176430;MOSAIC VARIEGATED ANEUPLOIDY SYNDROME 1; MVA1		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
C12orf57	Temtamy syndrome, 218340;TEMTAMY SYNDROME; COLOBOMA, HYPOPLASTIC CORPUS CALLOSUM AND INTELLECTUAL DISABILITY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
C12orf65	Combined oxidative phosphorylation deficiency 7, OMIM:613559;Spastic paraplegia 55, autosomal recessive, OMIM:615035		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
C5orf42	JOUBERT SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730;OSTEOPETROSIS AUTOSOMAL RECESSIVE TYPE 3 (OPTB3);carbonic anhydrase II deficiency;intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227;CEREBELLAR ATAXIA MENTAL RETARDATION AND DYSEQUILIBRIUM SYNDROME TYPE 3 (CMARQ3)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNA1A	Developemental and epileptic encephalopathy 42, OMIM:617106;developmental and epileptic encephalopathy, 42, MONDO:0014917		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNA1C	Timothy syndrome, OMIM:601005;Timothy syndrome, MONDO:0010979;Long QT syndrome 8, OMIM:618447;long qt syndrome 8, MONDO:0032756;Brugada syndrome 3, OMIM:611875;Brugada syndrome 3, MONDO:0012742;CACNA1C-related disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities 615474 AD; Sinoatrial node dysfunction and deafness 614896 AR		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNA1E	Global developmental delay;Intellectual disability;Seizures;Dystonia;Congenital contracture;Macrocephaly;Epileptic encephalopathy, early infantile, 69, 618285		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNA1G	Spinocerebellar ataxia 42 616795;Cerebellar atrophy, epilepsy, intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNA2D1	Abnormal muscle tone;Feeding difficulties;Global developmental delay;Intellectual disability;Seizures;Microcephaly;Abnormality of the corpus callosum;Cerebral atrophy;Abnormality of movement;Cortical visual impairment;Pain insensitivity		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CAMK4	Global developmental delay;Intellectual disability;Autism;Behavioral abnormality;Abnormality of movement;Dystonia;Ataxia;Chorea;Myoclonus		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CAMTA1	CEREBELLAR ATAXIA, NONPROGRESSIVE, WITH MENTAL RETARDATION		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CASK	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749FG syndrome 4, 300422Mental retardation, with or without nystagmus, 300422;MENTAL RETARDATION X-LINKED CASK-RELATED (MRXCASK)		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CASP2	Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly, OMIM:620653		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;NOONAN SYNDROME-LIKE DISORDER WITH OR WITHOUT JUVENILE MEYLOMONOCYTIC LEUKEMIA		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200;Thrombosis, hyperhomocysteinemic, 236200;CYSTATHIONINE BETA-SYNTHASE DEFICIENCY (CBSD)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CC2D1A	Intellectual developmental disorder, autosomal recessive 3, OMIM:608443		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CC2D2A	Joubert syndrome 9, 612285Meckel syndrome 6, 612284COACH syndrome, 216360;MECKEL SYNDROME, TYPE 6; MKS6		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, OMIM:235510		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CCDC22	SYNDROMIC X-LINKED INTELLECTUAL DISABILITY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CCDC88C	?Spinocerebellar ataxia 40 616053 AD; Hydrocephalus, nonsyndromic, autosomal recessive 236600 AR		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CDC6	Meier-Gorlin syndrome 5, OMIM:613805		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CDK5RAP2	Microcephaly 3, primary, autosomal recessive, 604804;PRIMARY AUTOSOMAL RECESSIVE MICROCEPHALY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672Angelman syndrome-like, 105830;EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 2 (EIEE2)		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CDON	Holoprosencephaly 11, 614226;HOLOPROSENCEPHALY 11		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CENPJ	Microcephaly 6, primary, autosomal recessive, 608393Seckel syndrome 4, 613676;MICROCEPHALY PRIMARY TYPE 6 (MCPH6)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CEP135	Microcephaly 8, primary, autosomal recessive, 614673;PRIMARY MICROCEPHALY AND DISTURBED CENTROSOMAL FUNCTION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CEP152	Microcephaly 9, primary, autosomal recessive, 614852Seckel syndrome 5, 613823;MICROCEPHALY PRIMARY TYPE 4 (MCPH4)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CEP290	Joubert syndrome 5, 610188Senior-Loken syndrome 6, 610189Leber congenital amaurosis 10, 611755Meckel syndrome 4, 611134Bardet-Biedl syndrome 14, 209900;BARDET-BIEDL SYNDROME TYPE 14 (BBS14)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CEP41	Joubert syndrome 15, 614464;JOUBERT SYNDROME 15		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CEP57	MOSAIC VARIEGATED ANEUPLOIDY SYNDROME 2		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CEP85L	Lissencephaly 10, OMIM:618873;Lissencephaly 10, MONDO:0030031		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHD2	Epileptic encephalopathy, childhood-onset, 615369;EPILEPTIC ENCEPHALOPATHY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHD3	Global developmental delay;Intellectual disability;Macrocephaly;Snijders Blok-Campeau syndrome, 618205		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHD7	CHARGE syndrome, 214800{Scoliosis, idiopathic 3}, 608765Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;KALLMANN SYNDROME TYPE 5 (KAL5)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHD8	Overgrowth with Intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHKB	Muscular dystrophy, congenital, megaconial type, 602541		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CIC	Mental retardation, autosomal dominant 45 617600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200;NEURONAL CEROID LIPOFUSCINOSIS TYPE 3 (CLN3)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CLN5	Ceroid lipofuscinosis, neuronal, 5 OMIM:256731;neuronal ceroid lipofuscinosis 5 MONDO:0009745		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300;CEROID LIPOFUSCINOSIS, NEURONAL, 6		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;NEURONAL CEROID LIPOFUSCINOSIS TYPE 8 (CLN8)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CNKSR2	Intellectual developmental disorder, X-linked, syndromic, Houge type, OMIM:301008;intellectual disability, X-linked, syndromic, Houge type, MONDO:0030909		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CNNM2	Hypomagnesemia, seizures, and mental retardation, OMIM:616418;Hypomagnesemia, seizures, and mental retardation, MONDO:0014631		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CNOT3	CNOT3 syndrome;intellectual disability, global developmental delay;Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome, 610042{Autism susceptibility 15}, 612100Pitt-Hopkins like syndrome 1, 610042;CORTICAL DYSPLASIA-FOCAL EPILEPSY SYNDROME (CDFES)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COG1	Congenital disorder of glycosylation, type IIg, 611209;COG1-CDG (CDG-IIG)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COG4	Congenital disorder of glycosylation, type IIj, 613489;COG4-CDG (CDG-IIJ)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COG5	Congenital disorder of glycosylation, type III, 613612;COG5-CDG;CDG-III		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43

COG6	Congenital disorder of glycosylation, type III, 614576;Shaheen syndrome, 615328		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COG7	Congenital disorder of glycosylation, type IIe, 608779;COG7-CDG (CDG-IIE)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COG8	Congenital disorder of glycosylation, type IIh, 611182;COG8-CDG (CDG-IIH)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL4A1	Porencephaly 1, 175780Brain small vessel disease with hemorrhage, 607595Angiopathy, hereditary, with nephropathy, aneurysms, and muscle, 611773Brain small vessel disease with Axenfeld-Rieger anomaly, 607595{Hemorrhage, intracerebral, susceptibility to}, 614519;PORENCEPHALY 1		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL4A2	Porencephaly 2, 614483{Hemorrhage, intracerebral, susceptibility to}, 614519;PORENCEPHALY 2		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COLEC11	3MC syndrome 2, 265050;3MC SYNDROME 2		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COQ4	COENZYME Q10 DEFICIENCY, PRIMARY, 7		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COX10	Mitochondrial complex IV deficiency, nuclear type 3, OMIM:619046		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COX15	Mitochondrial complex IV deficiency, nuclear type 6, OMIM:615119		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CPE	Intellectual developmental disorder and hypogonadotropic hypogonadism, OMIM:619326		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CPS1	Carbamoylphosphate synthetase I deficiency, 237300{Pulmonary hypertension, neonatal, susceptibility to}, 615371{Venoocclusive disease after bone marrow transplantation};CARBAMOYL PHOSPHATE SYNTHETASE 1 DEFICIENCY (CPS1D)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRADD	Mental retardation, autosomal recessive 34, with variant lissencephaly, OMIM:614499		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CREBBP	Rubinstein-Taybi syndrome, 180849;RUBINSTEIN-TAYBI SYNDROME TYPE 1 (RSTS1)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CSDE1	Autism;Global developmental delay;Intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), OMIM:254800		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168;CONGENITAL CATARACTS FACIAL DYSMORPHISM AND NEUROPATHY SYNDROME (CCFDN)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CTNNB1	Neurodevelopmental disorder with spastic diplegia and visual defects, OMIM:615075		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CTNND1	developmental delay		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CTSA	Galactosialidosis, 256540;GALACTOSIALIDOSIS (GSL)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127;NEURONAL CEROID LIPOFUSCINOSIS TYPE 10 (CLN10)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CUL3	Neurodevelopmental disorder with or without autism or seizures, OMIM:619239		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354;MENTAL RETARDATION SYNDROMIC X-LINKED CABEZAS TYPE (MRXC)		X-LINKED: hemizygous mutation in males, biallelic mutations in females		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CYB5R3	Methemoglobinemia, type I, 250800Methemoglobinemia, type II, 250800;METHEMOGLOBINEMIA DUE TO DEFICIENCY OF METHEMOGLOBIN REDUCTASE		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
D2HGDH	D-2-hydroxyglutaric aciduria, 600721		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DAG1	Walker-Warburg syndrome;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9 613818;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9 616538		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, OMIM:611105		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DBT	Maple syrup urine disease, type II, 248600;MAPLE SYRUP URINE DISEASEQ		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DCAF17	Woodhouse-Sakati syndrome, 241080;WOODHOUSE-SAKATI SYNDROME (WOSAS)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DCX	Lissencephaly, X-linked, OMIM:300067;Subcortical laminal heterotopia, X-linked, OMIM:300067		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DDC	Aromatic L-amino acid decarboxylase deficiency, OMIM:608643;Aromatic L-amino acid decarboxylase deficiency, MONDO:0012084		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DDX11	WARSAW BREAKAGE SYNDROME (WBRS)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171;Mental retardation, autosomal dominant 24, 615828		BOTH monoallelic and biallelic, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DHCR24	Desmosterolosis, 602398;DESMOSTEROLOSIS		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DHCR7	Smith-Lemli-Opitz syndrome, 270400;SMITH-LEMLI-OPITZ SYNDROME (SLOS)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DHDDS	Developmental delay and seizures with or without movement abnormalities, OMIM:617836		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839;MEGALOBLASTIC ANEMIA DUE TO DIHYDROFOLATE REDUCTASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DHX37	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, MIM#618731		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DIAPH1	Seizures, cortical blindness, microcephaly syndrome, 616632; developmental delay; intellectual disability		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DIS3L2	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DKC1	Dyskeratosis congenita, X-linked, 305000;DKC1-RELATED DYSKERATOSIS CONGENITA		X-LINKED: hemizygous mutation in males, biallelic mutations in females		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900;DIHYDROLIPOAMIDE DEHYDROGENASE (E3) DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DLG3	Mental retardation, X-linked 90, 300850;MENTAL RETARDATION X-LINKED TYPE 90 (MRX90)		X-LINKED: hemizygous mutation in males, biallelic mutations in females		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DLL1	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, OMIM:618709		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DMD	Duchenne muscular dystrophy, 310200Becker muscular dystrophy, 300376Cardiomyopathy, dilated, 3B, 302045;BECKER MUSCULAR DYSTROPHY (BMD)		X-LINKED: hemizygous mutation in males, biallelic mutations in females		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DNAJC19	3-methylglutaconic aciduria, type V, 610198		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DNM1	Developmental and epileptic encephalopathy 31, OMIM:616346		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DNM1L	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, MIM#614388		BOTH monoallelic and biallelic, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DNMT3A	Tatton-Brown-Rahman syndrome OMIM:615879;Heyn-Sproul-Jackson syndrome OMIM:618724;MONDO:0032882		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860;IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME 1		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DOCK3	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia OMIM:618292;neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia MONDO:0032661		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DOCK4	neuronevelopmental disorder, MONDO:0700092;intellectual disability, MONDO:0001071		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DOCK6	Adams-Oliver syndrome 2, 614219;intellectual disability, developmental delay		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DOLK	CONGENITAL DISORDERS OF GLYCOSYLATION		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093Myasthenic syndrome, congenital, with tubular aggregates 2, 614750;MYASTHENIC SYNDROME, CONGENITAL, WITH TUBULAR AGGREGATES 2		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DPM1	Congenital disorder of glycosylation, type Ie, OMIM:608799		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DPYD	Dihydropyrimidine dehydrogenase deficiency, 2742705-fluorouracil toxicity, 274270		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DPYS	Dihydropyrimidinuria, OMIM:222748		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DYM	Dyggve-Melchior-Clausen disease, 223800Smith-McCort dysplasia, 607326;SMITH-MCCORT DYSPLASIA (SMC)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DYNC1H1	Mental Retardation, Dominant;Charcot-Marie-Tooth disease, axonal, type 20, 614228;Mental retardation, autosomal dominant 13, 614563;Spinal muscular atrophy, lower extremity-predominant, AD, 158600;SPINAL MUSCULAR ATROPHY, LOWER EXTREMITY-PREDOMINANT, AD		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DYRK1A	Mental retardation, autosomal dominant 7, 614104;MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 7		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EARS2	Combined oxidative phosphorylation deficiency 12, MIM#614924		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EBP	CHONDRODYSPLASIA PUNCTATA 2, X-LINKED		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type, 610536;MANDIBULOFACIAL DYSOSTOSIS WITH MICROCEPHALY; MFDM		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EHMT1	Kleefstra syndrome, 610253;9Q SUBTELOMERIC DELETION SYNDROME (9Q- SYNDROME)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EIF2AK3	Wolcott-Rallison syndrome, 226980;WOLCOTT-RALLISON SYNDROME (WRS)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ELAC2	INFANTILE HYPERTROPHIC CARDIOMYOPATHY, LACTIC ACIDOSIS, AND ISOLATED COMPLEX I DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ELOVL4	Stargardt disease 3, 600110Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110Ichthyosis, spastic quadriplegia, and mental retardation, 614457;ICHTHYOSIS, SPASTIC QUADRIPLEGIA, AND MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ELP2	Mental retardation, autosomal recessive 58 617270		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EMX2	Schizencephaly 269160		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ENTPD1	Spastic paraplegia 64, autosomal recessive, OMIM:615683		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EP300	RUBINSTEIN-TAYBI SYNDROME TYPE 2 (RSTS2)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ERBB4	intellectual disability MONDO:0001071		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ERCC1	FANCONI ANEMIA		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ERCC2	Xeroderma pigmentosum, group D, 278730Trichothiodystrophy, 601675Cerebrooculofacioskeletal syndrome 2, 610756;CEREBRO-OCULO-FACIO-SKELETAL SYNDROME TYPE 2 (COFS2)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ERCC3	Xeroderma pigmentosum, group B, 610651Trichothiodystrophy, 601675;XERODERMA PIGMENTOSUM COMPLEMENTATION GROUP B (XP-B)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ERCC5	Xeroderma pigmentosum, group G, 278780Xeroderma pigmentosum, group G/Cockayne syndrome, 278780;XERODERMA PIGMENTOSUM COMPLEMENTATION GROUP G (XP-G)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ERCC6	De Sanctis-Cacchione syndrome, OMIM:278800		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ERCC8	Cockayne syndrome, type A, 216400UV-sensitive syndrome 2, 614621;COCKAYNE SYNDROME TYPE A (CSA)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ERLIN2	Spastic paraplegia 18, autosomal recessive, 611225;intellectual disability;AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ESAM	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, OMIM:620371		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ESCO2	Roberts syndrome, 268300SC phocomelia syndrome, 269000;SC PHOCOMELIA SYNDROME (SCPS)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ETFA	Glutaric acidemia IIA, OMIM:231680;multiple acyl-CoA dehydrogenase deficiency, MONDO:0009282		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ETFB	Glutaric acidemia IIB, OMIM:231680;multiple acyl-CoA dehydrogenase deficiency, MONDO:0009282		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ETFDH	GLUTARIC ACIDURIA TYPE 2C		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ETHE1	Ethylmalonic encephalopathy, 602473;ETHYLMALONIC ENCEPHALOPATHY		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EXOSC3	Pontocerebellar hypoplasia, type 1B, OMIM:614678		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EXT2	Seizures, scoliosis, and macrocephaly syndrome, 616682;autosomal recessive EXT2-related syndrome		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities, OMIM:617425		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EZH2	WEAVER SYNDROME 2		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532;LEUKODYSTROPHY HYPOMYELINATING TYPE 5 (HLD5)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FAM20C	RAINE SYNDROME		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FARS2	Spastic paraplegia 77, autosomal recessive, 617046;Combined oxidative phosphorylation deficiency 14, 614946		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FBXW11	Neurodevelopmental, jaw, eye, and digital syndrome, OMIM:618914;neurodevelopmental, jaw, eye, and digital syndrome, MONDO:003005		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FBXW7	Developmental delay, hypotonia, and impaired language, OMIM:620012		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FGD1	Aarskog-Scott syndrome, 305400Mental retardation, X-linked syndromic 16, 305400;AARSKOG-SCOTT SYNDROME (AAS)		X-LINKED: hemizygous mutation in males, biallelic mutations in females		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FH	Fumarase deficiency, OMIM:606812		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FIG4	Charcot-Marie-Tooth disease, type 4J, OMIM:611228;Charcot-Marie-Tooth disease type 4J MONDO:0012640;Yunis Varon syndrome OMIM:216340;Yunis-Varon syndrome MONDO:0008995		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5,606612Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C5 (MDDGC5)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FKTN	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152Cardiomyopathy, dilated, 1X, 611615Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL W		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FLVCR2	PROLIFERATIVE VASCULOPATHY AND HYDRAENCEPHALY-HYDROCEPHALY SYNDROME		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FMN2	NONSYNDROMIC AUTOSOMAL-RECESSIVE INTELLECTUAL DISABILITY		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FMR1	Fragile X syndrome, OMIM:300624;Fragile X tremor/ataxia syndrome, OMIM:300623		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOLR1	NEURODEGENERATION DUE TO CEREBRAL FOLATE TRANSPORT DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOXG1	Rett Syndrome, congenital variant OMIM:613454;Rett syndrome, congenital variant MONDO:0013270		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOXP1	Mental retardation with language impairment and autistic features, 613670;Mental Retardation with Language Impairment and Autistic Features;MENTAL RETARDATION WITH LANGUAGE IMPAIRMENT AND AUTISTIC FEATURES (MRLIAF)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOXP2	Speech-Language Disorder 1;SPEECH-LANGUAGE DISORDER 1		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19, OMIM:618241;mitochondrial complex 1 deficiency, nuclear type 19, MONDO:0032624		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FRMPD4	Mental retardation, X-linked 104, 300983;global developmental delay;intellectual disability		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FTCD	GLUTAMATE FORMIMINOTRANSFERASE DEFICIENCY (FIGLU-URIA)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FTSJ1	Mental retardation, X-linked 9, 309549;MENTAL RETARDATION X-LINKED TYPE 44 (MRX44)		X-LINKED: hemizygous mutation in males, biallelic mutations in females		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FUCA1	Fucosidosis, 230000;FUCOSIDOSIS (FUCA1D);intellectual disability		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FUT8	Congenital disorder of glycosylation with defective fucosylation, 618005;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GABRA1	JUVENILE MYOCLONIC EPILEPSY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GABRA5	Epileptic encephalopathy, early infantile, 79, 618559;developmental delay		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GABRB3	CHILDHOOD ABSENCE EPILEPSY TYPE 5		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GABRD	{Epilepsy, idiopathic generalized, 10}, OMIM:613060;{Epilepsy, juvenile myoclonic, susceptibility to}, OMIM:613060		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GABRG2	Epilepsy, generalized, with febrile seizures plus, type 3 611277;Febrile seizures, familial, 8 611277;{Epilepsy, childhood absence, susceptibility to, 2} 607681		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GAD1	?Cerebral palsy, spastic quadriplegic, 1603513;Developmental and epileptic encephalopathies, cleft palate, joint contractures and/or omphalocele		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GALC	Krabbe disease, OMIM:245200		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GALE	Galactose epimerase deficiency, 230350;EPIMERASE-DEFICIENCY GALACTOSEMIA (EDG)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GALNT2	Congenital disorder of glycosylation, type IIt 618885		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GALT	Galactosemia, 230400;GALACTOSEMIA (GALCT)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GAMT	Cerebral creatine deficiency syndrome 2, 612736;GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY (GAMT DEFICIENCY)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GAN	Giant axonal neuropathy-1, OMIM:256850		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GATM	Cerebral creatine deficiency syndrome 3, OMIM:612718		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GCDH	GLUTARICACIDEMIA TYPE 1		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230Hyperphenylalaninemia, BH4-deficient, B, 233910;DYSTONIA TYPE 5 (DYT5)		BOTH monoallelic and biallelic, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GCSH	Multiple mitochondrial dysfunctions syndrome 7, OMIM:620423;Glycine encephalopathy;Transient neonatal hyperglycinemia		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GDI1	Mental retardation, X-linked 41, 300849;MENTAL RETARDATION X-LINKED TYPE 41 (MRX41)		X-LINKED: hemizygous mutation in males, biallelic mutations in females		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GFAP	Alexander disease, 203450;ALEXANDER DISEASE		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GFM1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GJC2	Leukodystrophy, hypomyelinating, 2, OMIM:608804		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GK	Glycerol kinase deficiency, 307030;GLYCEROL KINASE DEFICIENCY		X-LINKED: hemizygous mutation in males, biallelic mutations in females		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLB1	GM1-gangliosidosis, type I, 230500GM1-gangliosidosis, type II, 230600GM1-gangliosidosis, type III, 230650Mucopolysaccharidosis type IVB (Morquio), 253010;MUCOPOLYSACCHARIDOSIS TYPE 4B (MPS4B)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLDC	Glycine encephalopathy, 605899;GLDC-RELATED GLYCINE ENCEPHALOPATHY		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLI2	Holoprosencephaly-9, 610829;GLI2-RELATED HOLOPROSENCEPHALY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLUL	CONGENITAL SYSTEMIC GLUTAMINE DEFICIENCY (CSGD)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLYCTK	D-glyceric aciduria 220120		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GM2A	GM2-gangliosidosis, AB variant, 272750;GM2-GANGLIOSIDOSIS TYPE AB (GM2GAB)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GNAS	Pseudohypoparathyroidism Ia, OMIM:103580;pseudohypoparathyroidism type 1A, MONDO:0007078;Pseudohypoparathyroidism Ic, OMIM:612462;pseudohypoparathyroidism type 1C, MONDO:0012911;Pseudopseudohypoparathyroidism, OMIM:612463;pseudopseudohypoparathyroidism, MONDO:0012912		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GNB5	Intellectual developmental disorder with cardiac arrhythmia, OMIM:617173;Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, OMIM:617182		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GNPAT	Chondrodysplasia punctata, rhizomelic, type 2, 222765;RHIZOMELIC CHONDRODYSPLASIA PUNCTATA TYPE 2 (RCDP2)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GNPTAB	MUCOLIPIDOSIS TYPE II (MLII)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GNPTG	MUCOLIPIDOSIS TYPE III COMPLEMENTATION GROUP C (MLIIIC)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GNS	Mucopolysaccharidosis type IIID, 252940;MUCOPOLYSACCHARIDOSIS TYPE 3D (MPS3D)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870Wilms tumor, somatic, 194070;SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 1		X-LINKED: hemizygous mutation in males, biallelic mutations in females		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GPC4	Keipert syndrome OMIM# 301026		X-LINKED: hemizygous mutation in males, biallelic mutations in females		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GRIA3	Mental retardation, X-linked 94, 300699;MENTAL RETARDATION X-LINKED TYPE 94 (MRX94)		X-LINKED: hemizygous mutation in males, biallelic mutations in females		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GRIK2	Mental retardation, autosomal recessive, 6, OMIM:611092;non-syndromic neurodevelopmental disorder (NDD), autosomal dominant		BOTH monoallelic and biallelic, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant OMIM:614254;intellectual disability, autosomal dominant 8 MONDO:0013655;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive OMIM:617820;neurodevelopmental disorder with or without hyperkinetic movements and seizu		BOTH monoallelic and biallelic, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GRIN2A	Epilepsy with neurodevelopmental defects, 613971;EPILEPSY WITH NEURODEVELOPMENTAL DEFECTS		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GRIN2B	Intellectual developmental disorder, autosomal dominant 6, with or without seizures, OMIM:613970;Developmental and epileptic encephalopathy 27, OMIM:616139		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GRM1	Spinocerebellar ataxia, autosomal recessive 13, OMIM:614831		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GRM7	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, OMIM:618922		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GTF2H5	Trichothiodystrophy, complementation group A, 601675;TRICHOTHIODYSTROPHY PHOTOSENSITIVE		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GUSB	Mucopolysaccharidosis VII, 253220;MUCOPOLYSACCHARIDOSIS TYPE 7 (MPS7)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures 616756		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HADHA	LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HCCS	Microphthalmia, syndromic 7, 309801;MICROPHTHALMIA SYNDROMIC TYPE 7 (MCOPS7)		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HCN1	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 24; EIEE24		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HDAC4	Brachydactyly-mental retardation syndrome, 600430;BRACHYDACTYLY-MENTAL RETARDATION SYNDROME (BDMR)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HDAC8	Wilson-Turner syndrome, 309585Cornelia de Lange syndrome 5, 300882;CORNELIA DE LANGE-LIKE SYNDROME		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, OMIM:613925;Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, OMIM:613926		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HERC2	Mental retardation, autosomal recessive 38, OMIM:615516		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HESX1	Growth hormone deficiency with pituitary anomalies, OMIM:182230;Pituitary hormone deficiency, combined, 5, OMIM:182230;Septooptic dysplasia, OMIM:182230		BOTH monoallelic and biallelic, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HEXA	Tay-Sachs disease, 272800;GM2-gangliosidosis, several forms, 272800;GM2-GANGLIOSIDOSIS TYPE 1 (GM2G1)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, OMIM:268800		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HGSNAT	MUCOPOLYSACCHARIDOSIS TYPE 3C (MPS3C)		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HIBCH	HIBCH DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HK1	Neurodevelopmental disorder with visual defects and brain anomalies, OMIM:618547		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HLCS	Holocarboxylase synthetase deficiency, 253270;HOLOCARBOXYLASE SYNTHETASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HMGCL	HMG-CoA lyase deficiency, 246450;3-Hydroxy-3-methylglutaryl-CoA lyase deficiency;HMGCLD;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HNMT	Mental retardation, autosomal recessive 51, OMIM:616739		BIALLELIC, autosomal or pseudoautosomal		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HNRNPU	Epileptic encephalopathy, early infantile, 54, 617391;intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted		Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43

HOXA1	Bosley-Salih-Alorainy syndrome, 601536Athabaskan brainstem dysgenesis syndrome, 601536;BOSLEY-SALIH-ALORAINY SYNDROME (BSAS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HPD	Tyrosinemia, type III, 276710Hawkinsinuria, 140350;HAWKINSINURIA (HAWK)		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HPRT1	Lesch-Nyhan syndrome, OMIM:300322		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HRAS	{Bladder cancer, somatic}, 109800Costello syndrome, 218040{Thyroid carcinoma, follicular, somatic}, 188470Congenital myopathy with excess of muscle spindles, 218040{Nevus sebaceous, somatic}, 162900Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;COSTELLO SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HSD17B10	HSD10 mitochondrial disease, OMIM:300438		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HSD17B4	PERRAULT SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HSPD1	Leukodystrophy, hypomyelinating, 4, OMIM:612233		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HTRA2	3-methylglutaconic aciduria, type VIII617248		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HUWE1	Intellectual developmental disorder, X-linked syndromic, Turner type, OMIM:309590		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IDH2	D-2-hydroxyglutaric aciduria 2613657		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IDS	Mucopolysaccharidosis II, 309900;MUCOPOLYSACCHARIDOSIS TYPE 2 (MPS2)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IDUA	Mucopolysaccharidosis Ih, 607014Mucopolysaccharidosis Is, 607016Mucopolysaccharidosis Ih/s, 607015;MUCOPOLYSACCHARIDOSIS TYPE 1S (MPS1S)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231; MEDS; Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IFIH1	Aicardi-Goutieres syndrome 7, OMIM:615846		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IGF1	Insulin-like growth factor I deficiency, OMIM:608747		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IGF1R	Insulin-like growth factor I, resistance to, OMIM:270450		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IKBKG	Incontinentia pigmenti, OMIM:308300		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IL1RAPL1	Mental Retardation, X-linked;Mental retardation, X-linked 21/34, 300143;MENTAL RETARDATION X-LINKED TYPE 21 (MRX21)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IMPDH2	Neurodevelopmental disorder with dystonia		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IQSEC2	Intellectual developmental disorder, X-linked 1, OMIM:309530		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IRX5	HYPERTELORISM, SEVERE, WITH MIDFACE PROMINENCE, MYOPIA, MENTAL RETARDATION, AND BONE FRAGILITY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643;WALKER WARBURG SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ITPA	Epileptic encephalopathy, early infantile, 35, 616647		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ITPR1	Gillespie syndrome, OMIM:206700;Spinocerebellar ataxia 15, OMIM:606658;Spinocerebellar ataxia 29, congenital nonprogressive, OMIM:117360		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IVD	ISOVALERIC ACIDEMIA		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730;HEMORRHAGIC DESTRUCTION OF THE BRAIN, SUBEPENDYMAL CALCIFICATION, AND CATARACTS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KANSL1	Koolen-De Vries syndrome, 610443;Intellectual Disability Syndrome;CHROMOSOME 17Q21.31 MICRODELETION SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KARS	Global developmental delay;Intellectual disability;Seizures;?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641;Deafness, autosomal recessive 89, 613916		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KAT6B	SBBYSS syndrome, 603736Genitopatellar syndrome, 606170;BLEPHAROPHIMOSIS/INTELLECTUAL DISABILITY PHENOTYPE WHICH IS NOONAN-LIKE		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNA3	Neurodevelopmental disorder, MONDO:0700092;intellectual disability, MONDO:0001071		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCND2	global developmental delay, HP:0001263		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNJ10	Seizures, Sensorineural Deafness, Ataxia, Mental Retardation, andElectrolyte Imbalance Syndrome;SESAME syndrome, 612780Enlarged vestibular aqueduct, digenic, 600791;SEIZURES-SENSORINEURAL DEAFNESS-ATAXIA-MENTAL RETARDATION-ELECTROLYTE IMBALANCE (SESAME)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNJ11	Hyperinsulinemic hypoglycemia, familial, 2, 601820Diabetes, permanent neonatal, 606176Diabetes mellitus, permanent neonatal, with neurologic features, 606176{Diabetes mellitus, type 2, susceptibility to}, 125853Diabetes mellitus, transient neonatal, 3, 610582;FAMILIAL HYPERINSULINISM		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNJ6	Keppen-Lubinsky syndrome 614098		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNK9	Birk-Barel mental retardation dysmorphism syndrome 612292		MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNMA1	Cerebellar atrophy, developmental delay, and seizures, OMIM:617643;Cerebellar atrophy, developmental delay, and seizures, MONDO:0060551;Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, OMIM:609446;Generalized epilepsy-paroxysmal dyskinesia syndrome, MONDO:0012276;Liang-Wang syndrome, OMIM:618729;Liang-Wang syndrome, MON		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNN3	Zimmermann-Laband syndrome 3, OMIM:618658		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNQ2	Developmental and epileptic encephalopathy 7, OMIM:613720		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNQ3			MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726;NEURONAL CEROID LIPOFUSCINOSIS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KDM5A	autism spectrum disorder, MONDO:0005258;intellectual disability, MONDO:0001071;El Hayek-Chahrour neurodevelopmental syndrome, OMIM:620820		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type, OMIM:300534		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KDM6A	Kabuki syndrome 2, OMIM:300867		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KDM6B	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, OMIM:618505;neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, MONDO:0032790		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950;AUTOSOMAL-DOMINANT MICROCEPHALY ASSOCIATED WITH LYMPHEDEMA AND/OR CHORIORETINOPATHY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KIF1A	NESCAV syndrome, OMIM:614255;Spastic paraplegia 30, autosomal dominant, OMIM:610357		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KIF5A	Myoclonus, intractable, neonatal, 617235;intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KIF7	Hydrolethalus syndrome 2, 614120Acrocallosal syndrome, 200990Joubert syndrome 12, 200990;ACROCALLOSAL SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KIRREL3	Intellectual developmental disorder, autosomal dominant 4, OMIM:612581;intellectual disability, autosomal dominant 4, MONDO:0012947		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KLF7	Intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KLHL7	BOS-like phenotype		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KMT2C	Kleefstra syndrome 2, 617768		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KMT2D	Kabuki syndrome 1, OMIM:147920		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KMT2E	O'Donnell-Luria-Rodan syndrome, 618512;Global developmental delay;Intellectual disability;Autism;Seizures;Abnormality of skull size		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KRAS	Lung cancer, somatic, 211980Bladder cancer, somatic, 109800Pancreatic carcinoma, somatic, 260350Gastric cancer, somatic, 137215Leukemia, acute myelogenousNoonan syndrome 3, 609942Cardiofaciocutaneous syndrome 2, 615278Breast cancer, somatic, 114480SFM syndrome, somatic mosaic, 163200;CARDIOFACIOCUTANEOUS SYNDROME (CFC SYNDROME)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
L1CAM	Corpus callosum, partial agenesis of, OMIM:304100;CRASH syndrome, OMIM:303350;MASA syndrome, OMIM:303350;Hydrocephalus due to aqueductal stenosis, OMIM:307000;Hydrocephalus with congential idiopathic intestinal pseudoobstruction, OMIM:307000;Hydrocephalus with Hirschsprung disease, OMIM:307000		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
L2HGDH	L-2-hydroxyglutaric aciduria, 236792;L-2-HYDROXYGLUTARIC ACIDURIA (L2HGA)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LAMA1	Poretti-Boltshauser syndrome OMIM:615960;ataxia - intellectual disability - oculomotor apraxia - cerebellar cysts syndrome MONDO:0014419		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LAMA2	Muscular dystrophy, congenital merosin-deficient, 607855Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855;CONGENITAL MUSCULAR DYSTROPHY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LAMB1	Lissencephaly 5, OMIM:615191		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LAMC3	Cortical malformations, occipital, 614115;OCCIPITAL CORTICAL MALFORMATIONS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LAMP2	Danon disease, 300257;DANON DISEASE (DAND)		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LETM1	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, OMIM:620089		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LIAS	Hyperglycinemia, lactic acidosis, and seizures, 614462		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LIG4	LIG4 syndrome, OMIM:606593		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LMNB1	Microcephaly 26, primary, autosomal dominant, OMIM:619179		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LRP2	Donnai-Barrow syndrome, 222448;INTELLECTUAL DISABILITY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LRPPRC	Leigh syndrome, French-Canadian type, 220111;LEIGH SYNDROME, FRENCH-CANADIAN TYPE		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MADD	DEEAH syndrome, OMIM:619004;deeah syndrome, MONDO:0033561; Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, OMIM:619005;neurodevelopmental disorder with dysmorphic facies, impaired speech, and hypotonia, MONDO:0033562		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAF	CATARACT, DEAFNESS, INTELLECTUAL DISABILITY, SEIZURES, AND A DOWN SYNDROME-LIKE FACIES		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAGEL2	PRADER WILLI SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAN1B1	Mental Retardation, Recessive;Mental retardation, autosomal recessive 15, 614202;AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAN2B1	Mannosidosis, alpha-, types I and II, 248500;LYSOSOMAL ALPHA-MANNOSIDOSIS (AM)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MANBA	Mannosidosis, beta, 248510;LYSOSOMAL BETA-MANNOSIDOSIS (LYSBMAN)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAOA	Brunner syndrome, 300615;BRUNNER SYNDROME (BRUNS)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279;CARDIOFACIOCUTANEOUS SYNDROME (CFC SYNDROME)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280;CARDIOFACIOCUTANEOUS SYNDROME (CFC SYNDROME)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MASP1	3MC SYNDROME 1; 3MC1		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAST4	neurodevelopmental disorder MONDO:0700092, MAST4-related		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/IIIdeficiency, 250850Methionine adenosyltransferase deficiency, autosomal recessive, 250850;METHIONINE ADENOSYLTRANSFERASE DEFICIENCY		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MBD5	Mental Retardation, Dominant;Autosomal Dominant Mental Retardation syndrome type 1;Mental retardation, autosomal dominant 1, 156200;EHMT1-LIKE INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MBTPS2	IFAP syndrome with or without BRESHECK syndrome 308205;IFAP syndrome with or without BRESHECK syndrome,308205;Ichthyosis follicularis, atrichia, and photophobia with or without brain anomalies, retardation, ectodermal dysplasia, skeletal malformations, hirschsprung disease, ear/eye anomalies, cleft palate/cryptorchidism, and kidney dysplasia/hypoplasia		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200;3-METHYLCROTONYL-COA CARBOXYLASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210;3-METHYLCROTONYL-COA CARBOXYLASE 2 DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MCM3AP	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MCOLN1	Mucolipidosis IV, 252650;MUCOLIPIDOSIS IV		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MCPH1	Microcephaly 1, primary, autosomal recessive, OMIM:251200		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MECP2	Rett syndrome, 312750Mental retardation, X-linked, syndromic 13, 300055Rett syndrome, preserved speech variant, 312750Encephalopathy, neonatal severe, 300673{Autism susceptibility, X-linked 3}, 300496Angelman syndrome, 105830Mental retardation, X-linked syndromic, Lubs type, 300260;RETT SYNDROME (RTT)[		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MED12	Opitz-Kaveggia syndrome, 305450;Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;OPITZ-KAVEGGIA SYNDROME (OKS)		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MED13	Delayed speech and language development;Motor delay;Intellectual disability;Autistic behavior;Attention deficit hyperactivity disorder;Abnormality of the eye;Constipation		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MED13L	Impaired intellectual development and distinctive facial features with or without cardiac defects, OMIM:616789		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668;MICROCEPHALY, POSTNATAL PROGRESSIVE, WITH SEIZURES AND BRAIN ATROPHY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MED23	Mental retardation, autosomal recessive 18, 614249;MRT18;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MED25	Basel-Vanagait-Smirin-Yosef syndrome, 616449;BVSYS;Syndromic intellectual disability;Congenital cataract-microcephaly-nevus flammeus simplex-severe intellectual disability syndrome		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MEF2C	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443;MRSME;Chromosome 5q14.3 deletion syndrome, 613443		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MEIS2	Cleft palate, cardiac defects, and mental retardation, OMIM:600987;Cardiac malformation, cleft lip/palate, microcephaly, and digital anomalies, MONDO:0010970		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MFF	Encephalopathy due to defective mitochondrial and peroxisomal fission 2 617086		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MFSD2A	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain imaging abnormalities, 616486		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MFSD8	MFSD8-RELATED NEURONAL CEROID-LIPOFUSCINOSIS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MGAT2	Congenital disorder of glycosylation, type IIa, 212066;CONGENITAL DISORDER OF GLYCOSYLATION TYPE 2A (CDG2A)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MID1	Opitz GBBB syndrome, type I, 300000;OPITZ G/BBB SYNDROME, X-LINKED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MINPP1	Pontocerebellar hypoplasia		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MKKS	McKusick-Kaufman syndrome, 236700Bardet-Biedl syndrome 6, 209900;MCKUSICK-KAUFMAN SYNDROME (MKKS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MKS1	MECKEL SYNDROME TYPE 1 (MKS1)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MLC1	LEUKOENCEPHALOPATHY MEGALENCEPHALIC WITH SUBCORTICAL CYSTS (MLC)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360;MALONYL-COA DECARBOXYLASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100;METHYLMALONIC ACIDURIA TYPE CBLA		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MMAB	METHYLMALONIC ACIDURIA TYPE CBLB		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400;METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, CBLC TYPE		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MMADHC	Homocystinuria, cblD type, variant 1, 277410Methylmalonic aciduria, cblD type, variant 2, 277410Methylmalonic aciduria and homocystinuria, cblD type, 277410;METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA TYPE CBLD (MMADHC)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MOCS1	Molybdenum cofactor deficiency, type A, 252150;MOLYBDENUM COFACTOR DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MOCS2	Molybdenum cofactor deficiency, type B, 252150;MOLYBDENUM COFACTOR DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MOGS	Congenital disorder of glycosylation, type IIb, 606056		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MPDU1	Congenital disorder of glycosylation, type If, 609180;CONGENITAL DISORDERS OF GLYCOSYLATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MPLKIP	Trichothiodystrophy, nonphotosensitive 1, 234050;TRICHOTHIODYSTROPHY NON-PHOTOSENSITIVE TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719;COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 5		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis, OMIM:616834;Microcephaly-congenital cataract-psoriasiform dermatitis syndrome, MONDO:0014793		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MTFMT	Combined oxidative phosphorylation deficiency 15 OMIM:614947;combined oxidative phosphorylation defect type 15 MONDO:0013987;Mitochondrial complex I deficiency, nuclear type 27 OMIM:618248;mitochondrial complex 1 deficiency, nuclear type 27 MONDO:0032631		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MTHFR	METHYLENETETRAHYDROFOLATE REDUCTASE DEFICIENCY (MTHFRD)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MTHFS	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MTO1	Combined oxidative phosphorylation deficiency 10, 614702		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940;{Neural tube defects, folate-sensitive, susceptibility to}, 601634;METHYLCOBALAMIN DEFICIENCY TYPE G (CBLG)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270{Neural tube defects, folate-sensitive, susceptibility to}, 601634;HOMOCYSTINURIA-MEGALOBLASTIC ANEMIA, CBL E TYPE		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MUT	Methylmalonic aciduria, mut(0) type, 251000;METHYLMALONIC ACIDURIA TYPE MUT		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MVK	Mevalonic aciduria, OMIM:610377		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MYCN	Feingold syndrome 1, OMIM:164280		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MYO5A	Griscelli syndrome, type 1, 214450;ELEJALDE SYNDROME (ELEJAS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MYT1L	Mental retardation, autosomal dominant 39, 616521;MRD39;Intellectual disability;obesity		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NAA10	N-terminal acetyltransferase deficiency, 300855;NONPECIFIC SEVERE ID		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NAGA	Schindler disease, type I, 609241Kanzaki disease, 609242Schindler disease, type III, 609241;KANZAKI DISEASE (KANZD)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920;MUCOPOLYSACCHARIDOSIS TYPE 3B (MPS3B)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NBEA	Global developmental delay;Intellectual disability;Seizures;No OMIM number		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NCKAP1	Intellectual disability;Autism		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDE1	Lissencephaly 4 (with microcephaly), 614019;LISSENCEPHALY 4 (WITH MICROCEPHALY)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDP	Norrie disease, 310600Exudative vitreoretinopathy, X-linked, 305390;NORRIE DISEASE		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDST1	Mental retardation, autosomal recessive 46, 616116;MRT46;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFA1	Mitochondrial complex I deficiency, 252010;MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY (NUCLEAR GENES)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFA2	Mitochondrial complex I deficiency, nuclear type 13, MIM#618235		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFS1	Mitochondrial complex I deficiency, nuclear type 5, OMIM:618226		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFS4	Leigh syndrome, 256000Mitochondrial complex I deficiency, 252010;MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY (NUCLEAR GENES)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFS7	Leigh syndrome, 256000;MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY (NUCLEAR GENES)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency, 256000;MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY (NUCLEAR GENES)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFV1	Mitochondrial complex I deficiency, 252010;MITOCHONDRIAL COMPLEX I DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NEDD4L	Periventricular nodular heterotopia 7, 617201		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NEU1	Sialidosis, type I, 256550Sialidosis, type II, 256550;SIALIDOSIS (SIALIDOSIS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NF1	Neurofibromatosis, type 1, 162200Leukemia, juvenile myelomonocytic, 607785Melanoma, desmoplastic neurotrophic (2)Neurofibromatosis, familial spinal, 162210Neurofibromatosis-Noonan syndrome, 601321Watson syndrome, 193520;NEUROFIBROMATOSIS-NOONAN SYNDROME (NFNS)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NFIA	Brain malformations with or without urinary tract defects, 613735;BRMUTD;Intellectual disability;Chromosome 1p32-p31 deletion syndrome, included		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NFIX	SOTOS-LIKE SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NFU1	Multiple mitochondrial dysfunctions syndrome 1, OMIM:605711		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NHS	Nance-Horan syndrome, 302350Cataract 40, X-linked, 302200;NANCE-HORAN SYNDROME (NHS)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NIPBL	Cornelia de Lange syndrome 1, 122470;CORNELIA DE LANGE SYNDROME TYPE 1 (CDLS1)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NKX2-1	Goiter, familial, due to TTF-1 defect (1)Chorea, hereditary benign, 118700Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978;BENIGN HEREDITARY CHOREA		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NLGN3	AUTISM SPECTRUM DISORDERS		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NPC1	NIEMANN-PICK DISEASE, TYPE C1		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NPC2	NIEMANN-PICK DISEASE, TYPE C2		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NPHP1	Nephronophthisis 1, juvenile, 256100Senior-Loken syndrome-1, 266900Joubert syndrome 4, 609583;SENIOR-LOKEN SYNDROME TYPE 1 (SLSN1)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NR2F1	BOSCH-BOONSTRA OPTIC ATROPHY SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NR4A2	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, OMIM:619911		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NRAS	Autoimmune lymphoproliferative syndrome type IV, 614470Noonan syndrome 6, 613224Epidermal nevus, somatic, 162900Thyroid carcinoma, follicular, somatic, 188470Colorectal cancer, somatic, 114500;NOONAN SYNDROME TYPE 6		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NRXN1	Pitt-Hopkins-like syndrome 2, OMIM:614325 (AR);Complex neurodevelopmental disorder (AD)		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NSD1	Sotos syndrome 1, 117550Leukemia, acute myeloid, 601626 (1)Beckwith-Wiedemann syndrome, 130650;WEAVER SYNDROME (WES)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NSDHL	CHILD syndrome, 308050CK syndrome, 300831;CONGENITAL HEMIDYSPLASIA WITH ICHTHYOSIFORM ERYTHRODERMA AND LIMB DEFECTS (CHILD)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NSUN2	Mental Retardation, Recessive;Mental retardation, autosomal recessive 5, 611091;AUTOSOMAL- RECESSIVE INTELLECTUAL DISABILITY MRT5		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NTRK2	Epileptic encephalopathy, early infantile, 58, 617830;Obesity, hyperphagia, and developmental delay, 613886		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NUBPL	Mitochondrial complex I deficiency, nuclear type 21, OMIM:618242		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
OCLN	Pseudo-TORCH syndrome 1, 251290 (includes profound developmental delay);Band-like calcification with simplified gyration and polymicrogyria		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
OCRL	Lowe syndrome, 309000Dent disease 2, 300555;LOWE OCULOCEREBRORENAL SYNDROME (OCRL)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ODC1	Global developmental delay;Intellectual disability;Macrocephaly;Alopecia;Ectodermal dysplasia		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
OFD1	Oral-facial-digital syndrome 1, 311200Simpson-Golabi-Behmel syndrome, type 2, 300209Joubert syndrome 10, 300804;ORAL-FACIAL-DIGITAL SYNDROME TYPE 1 (OFD1)		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
OPA3	3-methylglutaconic aciduria, type III, 258501;Costeff syndrome; Cognitive regression		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486;MENTAL RETARDATION X-LINKED OPHN1-RELATED (MRXSO)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
OTC	CGD Ornithine transcarbamylase deficiency, 311250;ORNITHINE TRANSCARBAMYLASE DEFICIENCY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
OTX2	MICROPHTHALMIA SYNDROMIC TYPE 5 (MCOPS5)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PAFAH1B1	Lissencephaly 1, OMIM:607432;Subcortical laminar heterotopia, OMIM:607432		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PAH	Phenylketonuria, 261600[Hyperphenylalaninemia, non-PKU mild], 261600;NON-PHENYLKETONURIA HYPERPHENYLALANINEMIA (NON-PKU HPA)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43

PAK3	Mental retardation, X-linked 30/47, 300558;Mental Retardation, X-linked;AGENESIS OF THE CORPUS CALLOSUM		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700;CONGENITAL HYPOTHYROIDISM NON-GOITROUS TYPE 2 (CHNG2)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PC	Pyruvate carboxylase deficiency, OMIM:266150		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PCCA	PROPIONIC ACIDEMIA		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PCCB	PROPIONIC ACIDEMIA		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088;EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 9 (EIEE9)		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, OMIM:210720		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PDE4D	Acrodysostosis 2, with or without hormone resistance, 614613;Acrodysostosis Orphanet:950		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170Leigh syndrome, X-linked, 308930;PYRUVATE DEHYDROGENASE E1-ALPHA DEFICIENCY IN FEMALES		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PDHB	Pyruvate dehydrogenase E1-beta deficiency, OMIM:614111		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PDHX	Lacticacidemia due to PDX1 deficiency, OMIM:245349		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651;COENZYME Q10 DEFICIENCY, PRIMARY, 2		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652;COENZYME Q10 DEFICIENCY, PRIMARY, 3		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEPD	Prolidase deficiency, 170100;PROLIDASE DEFICIENCY (PD)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX1	Peroxisome biogenesis disorder 1A (Zellweger), 214100Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 1 (PBD-CG1)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870Peroxisome biogenesis disorder 6B, 614871;ADRENOLEUKODYSTROPHY NEONATAL (NALD)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX11B	Peroxisome biogenesis disorder 14B, 614920;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859Peroxisome biogenesis disorder 3B, 266510;ZELLWEGER SYNDROME (ZWS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883Peroxisome biogenesis disorder 11B, 614885;ADRENOLEUKODYSTROPHY NEONATAL (NALD)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX14	ZELLWEGER SYNDROME (ZWS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX16	Peroxisome biogenesis disorder 8A, (Zellweger), 614876Peroxisome biogenesis disorder 8B, 614877;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 9 (PBD-CG9)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886;ZELLWEGER SYNDROME (ZWS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866Peroxisome biogenesis disorder 5B, 614867;ZELLWEGER SYNDROME (ZWS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872Peroxisome biogenesis disorder 7B, 614873;INFANTILE REFSUM DISEASE (IRD)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 12 (PBD-CG12)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110Peroxisome biogenesis disorder 2B, 202370;ADRENOLEUKODYSTROPHY NEONATAL (NALD)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX6	Peroxisome biogenesis disorder 4A (Zellweger), OMIM:614862;Peroxisome biogenesis disorder 4B, OMIM:614863		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PEX7	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879;REFSUM DISEASE (RD)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PGK1	Phosphoglycerate kinase 1 deficiency, 300653;PHOSPHOGLYCERATE KINASE 1 DEFICIENCY (PGK1D)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PHF6	Borjeson-Forssman-Lehmann syndrome, OMIM:301900		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PHF8	Mental retardation syndrome, X-linked, Siderius type, 300263;MENTAL RETARDATION SYNDROMIC X-LINKED SIDERIUS TYPE (MRXSSD)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PHGDH	Phosphoglycerate dehydrogenase deficiency, 601815;NEU-LAXOVA SYNDROME; NLS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PHIP	INTELLECTUAL DISABILITY;Developmental delay, intellectual disability, obesity, and dysmorphic features, 617991		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PIGA	MULTIPLE CONGENITAL ANOMALIES-HYPOTONIA-SEIZURES SYNDROME 2; MCAHS2		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PIGL	Coloboma, Congenital Heart Disease, Ichthyosiform Dermatosis,Mental Retardation, and Ear Anomalies Syndrome;ZUNICH NEUROECTODERMAL SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080;MULTIPLE CONGENITAL ANOMALIES-HYPOTONIA-SEIZURES SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749;Hyperphosphatasia with Mental Retardation Syndrome;HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 2; HPMRS2		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300;Hyperphosphatasia with Mental Retardation Syndrome;HYPERPHOSPHATASIA WITH MENTAL RETARDATION (HPMR)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PIK3CA	Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Megalencephaly-capillary malformation-polymicrogyria syndrome, Orphanet:60040		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, OMIM:603387		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PIP5K1C	neurodevelopmental disorder, MONDO:0700092;intellectual disability, MONDO:0001071		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600Neurodegeneration with brain iron accumulation 2B, 610217Parkinson disease 14, 612953;NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 2B		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722;EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 12		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PLP1	Pelizaeus-Merzbacher disease, 312080Spastic paraplegia 2, X-linked, 312920;LEUKODYSTROPHY HYPOMYELINATING TYPE 1 (HLD1)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PMM2	Congenital disorder of glycosylation, type Ia, 212065;CONGENITAL DISORDERS OF GLYCOSYLATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PNKP	Epileptic encephalopathy, early infantile, 10, 613402;EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 10		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PNPLA6	Boucher-Neuhauser syndrome, 215470;Oliver-McFarlane syndrome, 275400;Trichomegaly-retina pigmentary degeneration-dwarfism syndrome		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POGZ	White-Sutton syndrome, OMIM:616364		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POLG	MITOCHONDRIAL DNA DEPLETION SYNDROME 4A (ALPERS TYPE)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POLR1C	Leukodystrophy, hypomyelinating, 11, OMIM:616494		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropichypogonadism, 607694;LEUKODYSTROPHY, HYPOMYELINATING, 7, WITH OR WITHOUT OLIGODONTIA AND/OR HYPOGONADOTROPIC HYPOGONADISM		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, OMIM:614381;POLR3B-related neurodevelopmental disorder;Ataxia, spasticity, and demyelinating neuropathy		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POLRMT	Combined oxidative phosphorylation deficiency 55, OMIM:619743		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH MENTAL RETARDATION TYPE B3 (		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH BRAIN AND EYE ANOMALIES TYPE		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH MENTAL RETARDATION TYPE B2 (		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PORCN	Focal dermal hypoplasia, 305600;FOCAL DERMAL HYPOPLASIA (FODH)		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PPM1D	Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450;IDDGIP		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PPP2R1A	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730;NEURONAL CEROID LIPOFUSCINOSIS TYPE 1 (CLN1)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PQBP1	Renpenning syndrome, 309500;RENPENNING S(YNDROME 1 (RENS1)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRICKLE2	Neurodevelopmental disorder;global developmental delay		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures617157		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRPF8	PRPF8-related developmental disorder (monoallelic);Retinitis pigmentosa 13, OMIM:600059		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRPS1	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Arts syndrome, 301835;Deafness, X-linked 1, 304500;CHARCOT-MARIE-TOOTH DISEASE X-LINKED RECESSIVE TYPE 5 (CMTX5);Gout, PRPS-related, 300661;Phosphoribosylpyrophosphate synthetase superactivity, 300661		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRSS12	Mental retardation, autosomal recessive 1, 249500;Mental Retardation, Recessive;MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 1 (MRT1)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PSAP	Metachromatic leukodystrophy due to SAP-b deficiency, 249900Gaucher disease, atypical, 610539Combined SAP deficiency, 611721Krabbe disease, atypical, 611722;ATYPICAL KRABBE DISEASE		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PSPH	PHOSPHOSERINE PHOSPHATASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PTCH1	Holoprosencephaly 7, OMIM:610828		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PTCHD1	AUTISM/ID		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PTEN	Cowden syndrome 1, 158350Lhermitte-Duclos syndrome, 158350Bannayan-Riley-Ruvalcaba syndrome, 153480{Meningioma}, 607174{Glioma susceptibility 2}, 613028Macrocephaly/autism syndrome, 605309PTEN hamartoma tumor syndromeVATER association with macrocephaly and ventriculomegaly, 276950{Prostate cancer, somatic}, 176807Thyroid carcinoma, follicular, so		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PTF1A	DIABETES MELLITUS, PERMANENT NEONATAL, WITH CEREBELLAR AGENESIS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PTPN11	Noonan syndrome 1, 163950LEOPARD syndrome 1, 151100Leukemia, juvenile myelomonocytic, 607785Metachondromatosis, 156250;LEOPARD SYNDROME TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PTS	6-PYRUVOYLTETRAHYDROPTERIN SYNTHASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462;Mitochondrial myopathy and sideroblastic anemia 1; MLASA; Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PYCR1	Cutis laxa, autosomal recessive, type IIB, 612940;ARCL2B;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
QDPR	BH4-DEFICIENT HYPERPHENYLALANINEMIA C		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAB18	Warburg micro syndrome 3, 614222;WARBURG MICRO SYNDROME TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAB23	ACROCEPHALOPOLYSYNDACTYLY TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAB39B	Intellectual developmental disorder, X-linked 72, OMIM:300271;Waisman syndrome, OMIM:311510		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAB3GAP1	Warburg micro syndrome 1, 600118;WARBURG MICRO SYNDROME TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAB3GAP2	Martsolf syndrome 1, OMIM:212720;Warburg micro syndrome 2, OMIM:614225		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAC1	intellectual disability; developmental delay		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAD21	Cornelia de Lange syndrome 4, OMIM:614701		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAF1	Noonan syndrome 5, 611553LEOPARD syndrome 2, 611554;NOONAN SYNDROME 5		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAI1	Immunodeficiency 9, 612782;SMITH-MAGENIS SYNDROME (SMS)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RALGAPA1	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation 618797		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RARS2	Pontocerebellar hypoplasia, type 6, 611523;PONTOCEREBELLAR HYPOPLASIA TYPE 6		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RBBP8	Jawad syndrome, 251255;Microcephaly with mental retardation and digital anomalies;Seckel syndrome, 24389050;growth retardation, microcephaly with mental retardation, and a characteristic facial appearance		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RBM10	TARP syndrome, 311900;TARP SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RELN	Lissencephaly 2 (Norman-Roberts type), OMIM:257320;neurodevelopmental disorder, MONDO:0700092		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RFT1	Congenital disorder of glycosylation, type In, 612015;CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1N		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333;AICARDI-GOUTIERES SYNDROME 4		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RNASEH2B	Aicardi-Goutieres syndrome 2, OMIM:610181		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329;AICARDI-GOUTIERES SYNDROME 3		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951;LEUKOENCEPHALOPATHY, CYSTIC, WITHOUT MEGALENCEPHALY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RNF113A	Trichothiodystrophy 5, nonphotosensitive, OMIM:300953		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ROBO1	Intellectual disability, MONDO:0001071;Neurooculorenal syndrome, OMIM:620305		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ROGDI	Kohlschutter-Tonz syndrome, 226750;KOHLSCH TTER-T NZ SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RORA	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RPGRIP1L	Joubert syndrome 7, 611560Meckel syndrome 5, 611561COACH syndrome, 216360;COACH SYNDROME (COACHS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RPIA	Ribose 5-phosphate isomerase deficiency, OMIM:608611		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RPL10	Intellectual developmental disorder, X-linked syndromic 35, OMIM:300998		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RPS6KA3	Coffin-Lowry syndrome, OMIM:303600;Intellectual developmental disorder, X-linked 19, OMIM:300844		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RRM2B	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075;Mitochondrial depletion syndrome; Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RSRC1	Intellectual developmental disorder, autosomal recessive 70 618402		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SAMD9	MIRAGE syndrome, 617053		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SAMHD1	AICARDI-GOUTIERES SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis 613845		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SATB2	Cleft palate and mental retardation, 119540;CLEFT PALATE ISOLATED (CPI)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403Dravet syndrome, 607208Migraine, familial hemiplegic, 3, 609634Febrile seizures, familial, 3A, 604403;SCN1A-RELATED SEIZURE DISORDERS		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCN2A	Seizures, benign familial infantile, 3, 607745Epileptic encephalopathy, early infantile, 11, 613721;NONSPECIFIC SEVERE ID		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCN3A	Epileptic encephalopathy, early infantile, 62, 617938;intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCN8A	Cognitive impairment with or without cerebellar ataxia, OMIM:614306;Developmental and epileptic encephalopathy 13, OMIM:614558		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377;Myopia 6, 608908;FATAL INFANTILE CARDIOENCEPHALOMYOPATHY DUE TO CYTOCHROME C OXIDASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SDCCAG8	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SDHA	Leigh syndrome, 256000Mitochondrial respiratory chain complex II deficiency, 252011Cardiomyopathy, dilated, 1GG, 613642Paragangliomas 5, 614165;LEIGH SYNDROME (NUCLEAR DNA MUTATION)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SDHAF1	MITOCHONDRIAL COMPLEX II DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SETBP1	Schinzel-Giedion midface retraction syndrome, 269150;SCHINZEL-GIEDION MIDFACE RETRACTION SYNDROME (SGMFS)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SETD2	Luscan-Lumish syndrome, 616831;intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SGSH	MUCOPOLYSACCHARIDOSIS TYPE 3A (MPS3A)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SHANK2	SUSCEPTIBILITY TO AUTISM TYPE 17 (AUTS17)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SHANK3	Phelan-McDermid syndrome, 606232{Schizophrenia 15}, 613950;PHELAN-MCDERMID SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SHH	Holoprosencephaly-3, 142945Single median maxillary central incisor, 147250Microphthalmia with coloboma 5, 611638Schizencephaly, 269160;TRIPHALANGEAL THUMB-POLYSYNDACTYLY SYNDROME (TPTPS)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SHOC2	Noonan-like syndrome with loose anagen hair, 607721;NOONAN-LIKE SYNDROME WITH LOOSE ANAGEN HAIR		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SIL1	Marinesco-Sjogren syndrome, 248800;MARINESCO-SJOEGREN SYNDROME (MSS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SIX3	Holoprosencephaly-2, 157170;Schizensephaly, 269160;HOLOPROSENCEPHALY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SKI	Shprintzen-Goldberg syndrome, 182212;SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME; SGS		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, OMIM:218000		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC16A2	Allan-Herndon-Dudley syndrome, OMIM:300523		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC17A5	Salla disease, OMIM:604369;Sialic acid storage disorder, infantile, OMIM:269920		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC19A3	THIAMINE METABOLISM DYSFUNCTION SYNDROME 2 (BIOTIN- OR THIAMINE-RESPONSIVE ENCEPHALOPATHY TYPE 2)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC1A1	Dicarboxylic aminoaciduria 222730		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC1A2	Epileptic encephalopathy, early infantile, 41, OMIM:617105;developmental and epileptic encephalopathy, 41, MONDO:0014916		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC25A12	Epileptic encephalopathy, early infantile, 39,612949;Hypomyelination, global cerebral;Epileptic encephalopathy with global cerebral demyelination;Delayed psychomotor development		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, OMIM:238970		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304;EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 3		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC2A1	GLUT1 deficiency syndrome 1, 606777;GLUT1 deficiency syndrome 2, 612126;{Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847;Dystonia 9, 601042;GLUT1 DEFICIENCY SYNDROME TYPE 1 (GLUT1DS1)		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585;intellectual disability, ataxia, seizures, bleeding diathesis and proteinuria		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265;CONGENITAL DISORDER OF GLYCOSYLATION TYPE 2C (CDG2C)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC46A1	HEREDITARY FOLATE MALABSORPTION (HFM)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities, 604278;PROXIMAL RENAL TUBULAR ACIDOSIS WITH OCULAR ABNORMALITIES (PRTA-OA)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC6A1	Myoclonic-atonic epilepsy, OMIM:616421		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC6A19	Hartnup disorder234500; Iminoglycinuria, digenic242600		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC6A3	PARKINSONISM-DYSTONIA, INFANTILE		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352;X-LINKED CREATINE DEFICIENCY SYNDROME (XL-CDS)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC9A6	Mental retardation, X-linked syndromic, Christianson type, 300243;MENTAL RETARDATION SYNDROMIC X-LINKED CHRISTIANSON TYPE (MRXSC)		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLX4	FANCONI ANEMIA COMPLEMENTATION GROUP P		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMAD4	Myhre syndrome, 139210;Includes intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMARCA2	Nicolaides-Baraitser syndrome, OMIM:601358;Coffin-siris syndrome;Blepharophimosis intellectual disability syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMARCA4	Coffin-Siris syndrome 4, OMIM:614609		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMARCB1	Coffin-Siris syndrome 3, OMIM:614608		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMARCE1	Coffin-Siris syndrome 5, OMIM:616938		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMC1A	Cornelia de Lange syndrome 2, OMIM:300590;Cornelia de Lange syndrome 2, MONDO:0010370;Developmental and epileptic encephalopathy 85, with or without midline brain defects, OMIM:301044;Developmental and epileptic encephalopathy, 85, with or without midline brain defects, MONDO:0026771		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMC3	Cornelia de Lange syndrome 3, 610759;CDLS3		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMOC1	Microphthalmia with limb anomalies, 206920;OPHTHALMOACROMELIC SYNDROME (OAS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMPD1	Niemann-Pick disease, type A, 257200Niemann-Pick disease, type B, 607616;NIEMANN-PICK DISEASE TYPE A (NPDA)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMS	Mental retardation, X-linked, Snyder-Robinson type, 309583;SNYDER-ROBINSON SYNDROME (SRS)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528;CEDNIK SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism OMIM:614501;psychomotor retardation, epilepsy, and craniofacial dysmorphism MONDO:0013787		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SOS1	Fibromatosis, gingival, 135300Noonan syndrome 4, 610733;NOONAN SYNDROME 4		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SOX10	Waardenburg syndrome, type 4C, 613266Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584PCWH syndrome, 609136;PERIPHERAL DEMYELINATING NEUROPATHY, CENTRAL DYSMYELINATING LEUKODYSTROPHY, WAARDENBURG SYNDROME, AND HIRSCHSPRUNG DISEASE (PCWH)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SOX2	AEG SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SOX5	12P12.5 INTRAGENIC DELETIONS ASSOCIATED WITH INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SOX6	intellectual diability;ADHD;Craniosynostosis;Osteochondromas		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPECC1L	Opitz GBBB syndrome, type II,145410;Intellectual disability;Autosomal dominant Opitz G/BBB syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPG11	Spastic paraplegia 11, autosomal recessive, 604360;SPASTIC PARAPLEGIA-11		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPR	DOPA-RESPONSIVE DYSTONIA DUE TO SEPIAPTERIN REDUCTASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPRED1	Legius syndrome, 611431;LEGIUS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPTAN1	Developmental and epileptic encephalopathy 5, OMIM:613477;developmental and epileptic encephalopathy, 5, MONDO:0013277		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPTBN2	Spinocerebellar ataxia, autosomal recessive 14, OMIM:615386		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SRCAP	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, OMIM:619595;Floating-Harbor syndrome, OMIM:136140		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379Kahrizi syndrome, 612713;CONGENITAL DISORDERS OF GLYCOSYLATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ST3GAL3	Developmental and epileptic encephalopathy 15, OMIM:615006;developmental and epileptic encephalopathy, 15, MONDO:0014003;Intellectual developmental disorder, autosomal recessive 12, OMIM:611090;intellectual disability, autosomal recessive 12, MONDO:0012612		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43

ST3GAL5	Salt and pepper developmental regression syndrome, 609056;Developmental regression;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
STIL	Microcephaly 7, primary, autosomal recessive, 612703;MICROCEPHALY PRIMARY TYPE 7		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
STRA6	Microphthalmia, syndromic 9, 601186Microphthalmia, isolated, with coloboma 8, 601186;MICROPHTHALMIA SYNDROMIC TYPE 9 (MCOPS9)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, MIM:611087 (includes mental retardation); severe psychomotor retardation		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
STX1A	neurodevelopmental disorder, MONDO:0700092;intellectual disability, MONDO:0001071		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
STXBP1	Developmental and epileptic encephalopathy 4, OMIM:612164;developmental and epileptic encephalopathy, 4, MONDO:0012812		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SUCLG1	FATAL INFANTILE LACTIC ACIDOSIS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SUFU	Joubert syndrome 32, OMIM:617757		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SUMF1	SULFATIDOSIS, JUVENILE, AUSTIN TYPE		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SUOX	Sulfite oxidase deficiency, 272300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SUPT16H	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, OMIM:619480;Global developmental delay;Intellectual disability;Abnormality of the corpus callosum		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SURF1	Leigh syndrome, due to COX deficiency, 256000;LEIGH SYNDROME (NUCLEAR DNA MUTATION)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491;EPILEPSY, X-LINKED, WITH VARIABLE LEARNING DISABILITIES AND BEHAVIOR DISORDERS		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SYNGAP1	Mental retardation, autosomal dominant 5, 612621;EPILEPTIC ENCEPHALOPATHY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SYP	Mental Retardation, X-linked;Mental retardation, X-linked 96, 300802;MENTAL RETARDATION X-LINKED SYP-RELATED (MRXSYP)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TAF1	Mental retardation, X-linked, syndromic 33, 300966; global developmental delay; intellectual disability		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TAF2	Mental retardation, autosomal recessive 40, OMIM:615599		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TAT	Tyrosinemia, type II, 276600;TYROSINEMIA TYPE 2 (TYRO2)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TAZ	BARTH SYNDROME; BTHS		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBC1D23	Pontocerebellar hypoplasia, type 11, 617695;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBC1D24	Myoclonic epilepsy, infantile, familial, 605021Epileptic encephalopathy, early infantile, 16, 615338;NON SYNDROMAL HEARING LOSS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBCE	Kenny-Caffey syndrome, type 1, OMIM:244460;autosomal recessive Kenny-Caffey syndrome, MONDO:0009486;Hypoparathyroidism-retardation-dysmorphism syndrome, OMIM:241410;hypoparathyroidism-retardation-dysmorphism syndrome, MONDO:0009426		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBL1XR1	AUTISM		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TCF4	Pitt-Hopkins syndrome, 610954;PITT-HOPKINS SYNDROME (PTHS)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TCF7L2	Developmental disorders;Global developmental delay;Intellectual disability;Autism;Attention deficit hyperactivity disorder;Myopia;Abnormality of skeletal system		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TCN2	Transcobalamin II deficiency;can have a presentation similar to severe combined immunodeficiency;pancytopenia;neutropenic colitis;Agammaglobulinemia;megaloblastic bone marrow;thrombocytopenia;neutropenia;failure to thrive;hypotonia, myoclonic like movements, pallor, purpura, anaemia, thrombocytopenia, megaloblastosis, aplastic bone marrow.		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TCTN2	JOUBERT SYNDROME AND RELATED DISORDERS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TGIF1	HOLOPROSENCEPHALY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TH	DOPA-RESPONSIVE DYSTONIA		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
THRA	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 6		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 614132;CRANIOFACIAL DYSMORPHISM, SKELETAL ANOMALIES, AND MENTAL RETARDATION SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TMEM165	Congenital disorder of glycosylation, type IIk, 614727;CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IIK		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TMEM216	JOUBERT SYNDROME 2		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TMEM237	Joubert syndrome 14, 614424;JOUBERT SYNDROME 14		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TMEM67	Meckel syndrome 3, 607361Joubert syndrome 6, 610688{Bardet-Biedl syndrome 14, modifier of}, 209900COACH syndrome, 216360Nephronophthisis 11, 613550;COACH SYNDROME(COACHS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TMEM70	MITOCHONDRIAL COMPLEX V (ATP SYNTHASE) DEFICIENCY, NUCLEAR TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TOR1A	Arthrogryposis multiplex congenita 5, OMIM:618947;Arthrogryposis multiplex congenita 5, MONDO:0100218		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TP73	Ciliary dyskinesia, primary, 47, and lissencephaly, OMIM:619466		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500;NEURONAL CEROID LIPOFUSCINOSIS TYPE 2 (CLN2)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TPP2	Immunodeficiency 78 with autoimmunity and developmental delay, OMIM:619220		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRAPPC9	Mental retardation, autosomal recessive 13, OMIM:613192		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750Chilblain lupus, 610448Vasculopathy, retinal, with cerebral leukodystrophy, 192315{Systemic lupus erythematosus, susceptibility to}, 152700;AICARDI-GOUTIERES SYNDROME 1, DOMINANT AND RECESSIVE		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRIO	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, OMIM:617061;Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, OMIM:618825		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRIP12	Mental retardation, autosomal dominant 49617752		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRMT1	autosomal recessive intellectual disorder;ARID;Mental retardation, autosomal recessive 68, 618302;Global developmental delay;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRPM3	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, OMIM:620224		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRRAP	Microcephaly;Seizures;Abnormal heart morphology;Autism;Developmental delay with or without dysmorphic facies and autism, 603015;Intellectual disability;Abnormality of the urinary system;Global developmental delay		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TSC1	Tuberous sclerosis-1, 191100Lymphangioleiomyomatosis, 606690;Focal cortical dysplasia, Taylor balloon cell type, 607341;TUBEROUS SCLEROSIS TYPE 1 (TSC1)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TSC2	Tuberous sclerosis-2, 613254Lymphangioleiomyomatosis, somatic, 606690;LYMPHANGIOLEIOMYOMATOSIS (LAM)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TSEN2	PONTOCEREBELLAR HYPOPLASIA TYPE 2 AND TYPE 4		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TSEN34	PONTOCEREBELLAR HYPOPLASIA TYPE 2 AND TYPE 4		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TSEN54	?Pontocerebellar hypoplasia type 5, OMIM:610204;Pontocerebellar hypoplasia type 2A, OMIM:277470;Pontocerebellar hypoplasia type 4, OMIM:225753		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TSFM	Combined oxidative phosphorylation deficiency 3 610505		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TSHB	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS 4		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TSPAN7	Mental retardation, X-linked 58, 300210;MENTAL RETARDATION X-LINKED TYPE 58		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TTC19	MITOCHONDRIAL COMPLEX III DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TTC37	Trichohepatoenteric syndrome 1, 222470		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TTC8	Bardet-Biedl syndrome 8, 209900Retinitis pigmentosa 51, 613464;RETINITIS PIGMENTOSA TYPE 51 (RP51)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TTI2	AUTOSOMAL RECESSIVE MENTAL RETARDATION;Mental retardation, autosomal recessive 39, 615541		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TUBA1A	Lissencephaly 3, 611603;INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TUBB2B	Cortical dysplasia, complex, with other brain malformations 7, OMIM:610031		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TUBB3	CONGENITAL FIBROSIS OF THE EXTRAOCULAR MUSCLES		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TUBGCP6	MICROCEPHALY AND CHORIORETINOPATHY WITH OR WITHOUT MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TUSC3	Intellectual developmental disorder, autosomal recessive 7, OMIM:611093		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TWIST1	SAETHRE-CHOTZEN SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UBE2A	Mental retardation, X-linked syndromic, Nascimento-type, 300860;UBE2A-RELATED X-LINKED SYNDROMIC MENTAL RETARDATION		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UBE3A	Angelman syndrome, 105830;ANGELMAN SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UBR1	Johanson-Blizzard syndrome, 243800;JOHANSON-BLIZZARD SYNDROME (JBS)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UBR7	Intellectual disability;epilepsy;hypothyroidism;congenital anomalies;dysmorphic features		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UMPS	OROTIC ACIDURIA TYPE 1 (ORAC1)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, OMIM:616801		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UPF3B	Mental Retardation, X-linked;Mental retardation, X-linked, syndromic 14, 300676;MENTAL RETARDATION SYNDROMIC X-LINKED TYPE 14 (MRXS14)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UROC1	UROCANASE DEFICIENCY (UROD)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
USP7	Hao-Fountain syndrome, 616863		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
USP9X	Mental retardation, X-linked 99 300919 XLR;Mental retardation, X-linked 99, syndromic, female-restricted 300968		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
VCP	neurodevelopmental disorder, MONDO:0700092;intellectual disability, MONDO:0001071		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050;CEREBELLAR ATAXIA MENTAL RETARDATION AND DYSEQUILIBRIUM SYNDROME TYPE 1 (CMARQ1)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
VPS13B	Cohen syndrome, 216550;COHEN SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
VRK1	Pontocerebellar hypoplasia type 1A, 607596;PONTOCEREBELLAR HYPOPLASIA TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WDFY3	Microcephaly 18, primary, autosomal dominant, 617520		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WDPCP	BARDET-BIEDL SYNDROME TYPE 15 (BBS15)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WDR4	Galloway-Mowat syndrome 6, OMIM:61834;Microcephaly, growth deficiency, seizures, and brain malformations, OMIM:618347		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WDR45	Neurodegeneration with brain iron accululation 5, 300894;NEURODEGENERATION WITH BRAIN IRON ACCUMULATION (NBIA)		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WDR45B	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, OMIM:617977		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, OMIM:604317		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WDR81	cerebellar ataxia, intellectual disability and quadrupedal locomotion;Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WWOX	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
XRCC4	Short stature, microcephaly, and endocrine dysfunction, OMIM:616541		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
XYLT1	Desbuquois dysplasia 2, OMIM:615777;Desbuquois dysplasia 2, MONDO:0014343		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
YY1	Gabriele-de Vries syndrome617557		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZBTB18	intellectual disability with variable features; global developmental delay; Mental retardation, autosomal dominant 22, 612337		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069;intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type, OMIM:300799		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZEB2	Mowat-Wilson syndrome, 235730;MOWAT-WILSON SYNDROME (MWIS)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZFHX3	syndromic intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZFHX4	Developmental disorders;intellectual disability, dysmorphic features		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZFYVE26	Spastic paraplegia 15, autosomal recessive, OMIM:270700		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZIC1	Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736;?Craniosynostosis 6, 616602		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZIC2	Holoprosencephaly-5, 609637;HOLOPROSENCEPHALY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZMYND11	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZNF335	Microcephaly 10, primary, autosomal recessive, 615095		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZNF526	Intellectual disability;Microcephaly;Cataracts;Epilepsy;Hypertonia;Dystonia		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZNF711	Mental Retardation, X-linked;Mental retardation, X-linked 97, 300803;MENTAL RETARDATION X-LINKED ZNF711-RELATED (MRXZ)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACADSB	2-methylbutyrylglycinuria, 610006		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACADVL	VLCAD deficiency, 201475		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACAT1	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACP5	Spondyloenchondrodysplasia with immune dysregulation, 607944		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACTA2	Aortic aneurysm, familial thoracic 6, 611788;Multisystemic; smooth muscle dysfunction syndrome, 613834;Moyamoya disease 5, 614042		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACVR1	Fibrodysplasia ossificans progressiva 135100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ADA	Severe combined immunodeficiency due to ADA deficiency, OMIM:102700;Adenosine deaminase deficiency, partial, OMIM:102700		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ADAMTS10	Weill-Marchesani syndrome 1, recessive, MIM#277600		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AGMO	microcephaly;intellectual disability;epilepsy;developmental delay		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AGPS	Rhizomelic chondrodysplasia punctata, type 3, 600121		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AGXT	Hyperoxaluria, primary, type 1, 259900		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AIPL1	Leber congenital amaurosis 4, 604393;Retinitis pigmentosa, juvenile, 604393;Cone-rod dystrophy, 604393		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without,reversible metaphyseal dysplasia, 240300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AK2	NA		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AKR1D1	Bile acid synthesis defect, congenital, 2, 235555		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AKT1	Cowden syndrome 6 OMIM:164730		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALAD	Porphyria, acute hepatic, 612740; {Lead poisoning, susceptibility to}, 612740		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALDOA	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALPL	Hypophosphatasia, infantile, 241500; Hypophosphatasia, childhood, 241510; Odontohypophosphatasia, 146300; Hypophosphatasia, adult, 146300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALX3	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALX4	Parietal foramina;Parietal foramina 2, (AD), 609597;Frontonasal dysplasia 2, (AR), 613451		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ANO5	Gnathodiaphyseal dysplasia, 166260; Muscular dystrophy, limb-girdle, type 2L, 611307; Miyoshi muscular dystrophy 3, 613319		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ANTXR1	{Hemangioma, capillary infantile, susceptibility to}, 602089; GAPO syndrome, 230740		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARL14EP	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ASCC3	Intellectual developmental disorder, autosomal recessive 81, OMIM:620700		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
B3GALT6	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640; Ehlers-Danlos syndrome, progeroid type, 2, 615349		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
B4GALT1	Congenital disorder of glycosylation, type IId, OMIM:607091		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
B9D1	Meckel syndrome 9, OMIM:614209;Meckel syndrome 9, MONDO:0013630;Joubert syndrome 27, OMIM:617120;Joubert syndrome 27, MONDO:0014927		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BCORL1	Shukla-Vernon syndrome, 301029		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BSND	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
C8orf37	Retinitis pigmentosa 64, 614500;Cone-rod dystrophy 16, 614500		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNB4	Episodic ataxia, type 5 613855; {Epilepsy, idiopathic generalized, susceptibility to, 9} 607682; {Epilepsy, juvenile myoclonic, susceptibility to, 6} 607682		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CASR	Hypocalciuric hypercalcemia, type I, 145980		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CD96	C syndrome, 211750		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CDC42BPB	CDC42BPB-related Neurodevelopmental Disorder;Central hypotonia;Global developmental delay;Intellectual disability;Seizures;Autistic behavior;Behavioral abnormality		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CDKN1C	IMAGE syndrome, 614732;ntrauterine Growth Restriction, Metaphyseal Dysplasia, Adrenal Hypoplasia Congenita, and Genital Anomalies		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CEP63	?Seckel syndrome 6 614728; Developmental dyslexia		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHL1	verbal function and developmental delay		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHRM1	Neurodevelopmental delay;intellectual disability, MONDO:0001071;autism		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHST14	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CLCN2	Leukoencephalopathy with ataxia, OMIM:615651		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CNPY3	Epileptic encephalopathy, early infantile, 60 (MIM 617929)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COQ9	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRBN	Mental retardation, autosomal recessive 2 607417		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRELD1	Jeffries-Lakhani neurodevelopmental syndrome, OMIM:620771		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, OMIM:612199		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CYP27A1	Cerebrotendinous xanthomatosis 213700		Other - please specify in evaluation comments	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DCC	Gaze palsy, familial horizontal, with progressive scoliosis, 2, OMIM:617542		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DDOST	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IR; CDG1R		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DDX53			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DOCK8	Intellectual developmental disorder, autosomal dominant 2, OMIM:614113		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DPM3	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1O		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DPYSL2	intellectual disability, MONDO:0001071;Aplasia/Hypoplasia of the corpus callosum, HP:0007370		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DROSHA	Global developmental delay;Intellectual disability;Seizures;Cerebral white matter atrophy;Abnormality of the corpus callosum;Abnormality of movement;Stereotypic behavior;Abnormality of head or neck;Short foot		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EEF1B2	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EFNB1	Craniofrontonasal dysplasia, OMIM:304110		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EMG1	Bowen-Conradi syndrome, 211180		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EPB41L1	?Mental retardation, autosomal dominant 11 614257		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EPHA7	Global developmental delay;Intellectual disability, MONDO:0001071;Delayed speech and language development;Behavioral abnormality		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FAAH2			X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FANCA	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FANCC	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FANCD2	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FANCE	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FANCF	Fanconi anemia, complementation group F, 603467		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FANCG	Fanconi anemia, complementation group G, 614082		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FANCI	Fanconi anemia, complementation group I, 609053		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FDFT1	Squalene synthase deficiency, 618156		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FGF14	Spinocerebellar ataxia 27 609307		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FGFR2	Antley-Bixler syndrome;Apert syndrome;Crouzon syndrome;Beare-Stevenson Cutis Gyrata syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FRA10AC1	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, OMIM:620113		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FRAS1	Fraser syndrome 1 219000		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43

FREM2	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FRY	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FTO	Growth retardation, developmental delay, facial dysmorphism 612938		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GABBR1	Neurodevelopmental disorder with language delay and variable cognitive abnormalities, OMIM:620502		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GATA6	Atrioventricular septal defect 5, 614474; Atrial septal defect 9, 614475; Pancreatic agenesis and congenital heart defects, 600001; Persistent truncus arteriosus, 217095; Tetralogy of Fallot, 187500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GBA	Gaucher disease, type I, 230800;Gaucher disease, type II, 230900;Gaucher disease, type III, 231000		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GEMIN4	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, OMIM:617913		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GJB3	Erythrokeratodermia variabilis et progressiva, 133200;Deafness, autosomal dominant 2B, 612644;Deafness, autosomal recessive;Deafness, autosomal dominant, with peripheral neuropathy;Deafness, digenic, GJB2/GJB3, 220290		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLI3	Greig cephalopolysyndactyly syndrome, OMIM:175700;Pallister-Hall syndrome, OMIM:146510		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLS	Global developmental delay, progressive ataxia, and elevated glutamine, OMIM:618412;Developmental and epileptic encephalopathy 71, OMIM:618328;?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, OMIM:618339		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GNAI2	Syndromic intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GNE	Sialuria, MIM#269921		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GPSM2	Chudley-McCullough syndrome, 604213		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GSS	Glutathione synthetase deficiency;Hemolytic anemia due to glutathione synthetase deficiency 231900		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HADHB	Trifunctional protein deficiency, MIM#609015		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HARS	multisystem ataxic syndrome;mild-severe intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HAX1	Neutropenia, severe congenital 3, autosomal recessive, 610738		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HNF1B	Renal cysts and diabetes syndrome, 137920; Diabetes mellitus, noninsulin-dependent, 125853; {Renal cell carcinoma}, 144700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HSPG2	Schwartz-Jampel syndrome, type 1, OMIM:255800;Schwartz-Jampel syndrome, MONDO:0009717		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HTT	Lopes-Maciel-Rodan syndrome, OMIM:617435;LOMARS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IFT43	Cranioectodermal dysplasia 3, 614099		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
INPP4A	Intellectual disability;Seizures		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ITGA7	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
JMJD1C	Intellectual disability;Autism		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNA1	Episodic ataxia/myokymia syndrome, OMIM:160120		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNC3	Spinocerebellar ataxia 13, OMIM:605259;MONDO:0011529		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KIF5B	kyphomelic dysplasia, MONDO:0008881;intellectual disability, MONDO:0001071		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LAMB2	Pierson syndrome, MIM#609049		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LARS2	Perrault syndrome 4, OMIM:615300;Hydrops, lactic acidosis, and sideroblastic anemia, OMIM:617021;Leukodystrophy		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LMBRD1	Gene2Phenotype confirmed gene with ID HPO;Methylmalonic aciduria and homocystinuria, cblF type, 277380		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LMNB2	Microcephaly 27, primary, autosomal dominant, OMIM:619180		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LRP5	Osteoporosis-pseudoglioma syndrome, 259770		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LZTFL1	Bardet-Biedl syndrome 17, MIM#615994		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAPK10	Lennox-Gastaut syndrome;LGS;Epileptic Encephalopathy;Epileptic Encephalopathy Lennox-Gastaut type		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MIR17HG	Feingold syndrome 2, 614326;FS2;Brachydactyly with short stature and microcephaly;Intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MPV17	Gene2Phenotype confirmed gene with ID HPO;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NAGS	N-acetylglutamate synthase deficiency, 237310		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NBAS	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NBN	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NCAPD2	Microcephaly 21, primary, autosomal recessive, OMIM #617983		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFAF1	Mitochondrial complex I deficiency, nuclear type 11, MIM#618234		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFAF2	Leigh syndrome 256000;Mitochondrial complex I deficiency 252010		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFAF5	Mitochondrial complex 1 deficiency, 618238		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NHP2	Dyskeratosis congenita, autosomal recessive 2, 613987;H yeraal-Hreidarsson syndrome		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NPHP3	Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540;Meckel syndrome 7, 267010		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency, 266120		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NUP62	Striatonigral degeneration, infantile, 271930;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NYX	Night blindness, congenital stationary (complete), 1A, X-linked, 310500		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PAX1	Otofaciocervical syndrome 2, 615560		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PDE10A	Dyskinesia, limb and orofacial, infantile-onset, MIM#616921		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PDP1	Pyruvate dehydrogenase phosphatase deficiency, MIM#608782		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PMPCA	Spinocerebellar ataxia, autosomal recessive 2 (MIM 213200)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency, 610090;Neonatal epileptic encephalopathy;Global developmental delay		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POU1F1	Pituitary hormone deficiency, combined or isolated, 1, OMIM:613038		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRODH	Hyperprolinemia, type I, OMIM:239500;{Schizophrenia, susceptibility to, OMIM:4}, 600850		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRRT2	Epilepsy;mental retardation;movement disorders;paroxysmal disorder;Autosomal recessive mental retardation		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PSMB8	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, OMIM:256040		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PTH1R	Metaphyseal chondrodysplasia, Murk Jansen type, 156400; Chondrodysplasia, Blomstrand type, 215045; Eiken syndrome, 600002; Failure of tooth eruption, primary, 125350		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PTHLH	Humoral hypercalcemia of malignancy;Brachydactyly, type E2, 613382		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAD51	Fanconi anemia, complementation group R, OMIM:617244		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAX	Microphthalmia, isolated 3, OMIM:611038;isolated microphthalmia 3, MONDO:0012604		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RMRP	Cartilage-hair hypoplasia, 250250;Metaphyseal dysplasia without; hypotrichosis, 250460;Anauxetic dysplasia, 607095		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RNU4ATAC	Lowry-Wood syndrome, OMIM:226960;Microcephalic osteodysplastic primordial dwarfism, type I, OMIM:210710;Roifman syndrome, OMIM:616651		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RYR2			MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SACS	Spastic ataxia, Charlevoix-Saguenay type, 270550; Autosomal recessive spastic ataxia of Charlevoix-Saguenay, ARSACS; Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SALL1	Townes-Brocks branchiootorenal-like syndrome, 107480; Townes-Brocks syndrome 1, 107480; TBS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCN1B	Developmental and epileptic encephalopathy 52, OMIM:617350;Developmental and epileptic encephalopathy, 52, MONDO:0033361		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome, 300434; Intellectual disability		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC25A38	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC26A2	Diastrophic dysplasia, 222600;Atelosteogenesis II, 256050;Achondrogenesis Ib, 600972;Epiphyseal dysplasia, multiple, 4, 226900;Diastrophic dysplasia, broad bone-platyspondylic variant, 222600;De la; Chapelle dysplasia, 256050		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC27A4	Ichthyosis prematurity syndrome, 608649		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC2A2	{Diabetes mellitus, noninsulin-dependent}, 135853; Fanconi-Bickel; syndrome, 227810		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC35D1	Schneckenbecken dysplasia, 269250		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC39A13	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC4A1	Ovalocytosis; Spherocytosis, type 4, 612653; [Malaria, resistance to], 611162; Renal tubular acidosis, distal, AD, 179800; Renal tubular acidosis, distal, AR, 611590; [Blood group, Diego], 110500; [Blood group, Waldner], 112010; [Blood group, Wright], 112050; [Blood group, Froese], 601551; [Blood group, Swann], 601550		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC4A10	Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities, OMIM:620746		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC4A11	Corneal endothelial dystrophy 2, autosomal recessive, 217700;Corneal endothelial dystrophy and perceptive deafness, 217400;Corneal dystrophy, Fuchs endothelial, 4, 613268		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC5A5	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC5A7	Myasthenic syndrome, congenital, 20, presynaptic,CMS20, 617143		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMAD3	Loeys-Dietz syndrome, type 3, 613795		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency, OMIM:300123;Intellectual disability, X-linked, with panhypopituitarism, MONDO:0010252		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SOX9	Campomelic dysplasia with autosomal sex reversal, OMIM:114290;Acampomelic campomelic dysplasia, OMIM:114290;Campomelic dysplasia, OMIM:114290;campomelic dysplasia, MONDO:0007251		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SRGAP3	3p- syndrome, MIM:613792 (includes intellectual disability)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), OMIM:612073		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TAB2	Congenital heart defects, nonsyndromic, 2, OMIM:614980		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TAF1C	Global developmental delay;Intellectual disability;Spasticity;Strabismus;Seizures;Abnormality of nervous system morphology		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBX1	22q11.2 deletion syndrome, Orphanet:567 (includes developmental delay); DiGeorge syndrome, 188400 (includes mild to moderate learning difficulties); Velocardiofacial syndrome, 192430 (includes learning disability and mental retardation)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TERT	Dyskeratosis congenita, autosomal recessive 4, OMIM:613989		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TGFB1	Inflammatory bowel disease, immunodeficiency, and encephalopathy, OMIM:618213		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
THRB	compromised intellectual development		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TNR	Spastic para- or tetraparesis;Axial muscular hypotonia;Intellectual disability;Transient opisthotonus		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRAPPC10	Neurodevelopmental disorder with microcephaly, short stature, and speech delay, OMIM:620027		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRPC5	TRPC5-related neurodevelopmental disorder		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335; mildly delayed development		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TWIST2	Barber-Say syndrome, 209885 (includes mental retardation in some patients); Focal facial dermal dysplasia 3, Setleis type, 227260 (can include intellectual disability and developmental delay)		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UBE3C	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, OMIM:620270		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UPB1	Beta-ureidopropionase deficiency, 613161 (can include mental retardation, developmental delay)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
VIPAS39	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WDR11	Intellectual disability, MONDO:0001071;Microcephaly, MONDO:0001149;Short stature,HP:0004322		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
XPA	mental retardation; progressive intellectual impariment		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
YARS	Charcot-Marie-Tooth disease, dominant intermediate C 608323;Intellectual disability;deafness;nystagmus;liver dysfunction		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZC3H14	Mental retardation, autosomal recessive 56, 617125; intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZFP57	Diabetes mellitus, transient neonatal, 1, 601410		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABCB11	Cholestasis, progressive familial intrahepatic 2, 601847;Cholestasis, benign recurrent intrahepatic, 2, 605479		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABCB7	ANEMIA, SIDEROBLASTIC, WITH ATAXIA		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABCC6	Pseudoxanthoma elasticum, 264800;Pseudoxanthoma elasticum, forme fruste, 177850;Arterial calcification, generalized, of infancy, 2, 614473		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABCC8				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABCG5				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACAN	Spondyloepiphyseal dysplasia, Kimberley type, 608361;Spondyloepimetaphyseal dysplasia, aggrecan type, 612813;Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ACSF3	Combined malonic and methylmalonic aciduria, 614265		Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ADCY5	Dyskinesia, familial, with facial myokymia, 606703		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ADRA2B	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AFG3L2	Optic atrophy 12, OMIM:618977;Spinocerebellar ataxia 28, OMIM:610246;Spastic ataxia 5, autosomal recessive, OMIM:614487		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AFP				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AGK	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AGL	Glycogen storage disease IIIa, 232400; Glycogen storage disease; IIIb, 232400		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AGPAT2	Lipodystrophy, congenital generalized, type 1, 608594		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AGT				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AGTR2	Mental retardation, X-linked 88, 300852;X-LINKED MENTAL RETARDATION 88		Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AKR1C2	46XY sex reversal 8 614279		Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALDOB	Fructose intolerance, 229600		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALG2	?Congenital disorder of glycosylation, type Ii 607906; Myasthenic syndrome, congenital, 14, with tubular aggregates 616228		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALS2	Amyotrophic lateral sclerosis 2, juvenile, OMIM:205100;Primary lateral sclerosis, juvenile, OMIM:606353;Spastic paralysis, infantile onset ascending, OMIM:607225		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ALX1	?Frontonasal dysplasia 3 613456		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ANKH	Craniometaphyseal dysplasia, 123000Chondrocalcinosis 2, 118600;CRANIOMETAPHYSEAL DYSPLASIA JACKSON TYPE (CMDJ)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ANO10	Spinocerebellar ataxia, autosomal recessive 10 613728		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AP5Z1	Spastic paraplegia 48, autosomal recessive, OMIM:613647		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920;ATAXIA WITH OCULOMOTOR APRAXIA 1		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AQP7				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AR	Androgen insensitivity, OMIM:300068;Androgen insensitivity, partial, with or without breast cancer, OMIM:312300;Hypospadias 1, X-linked, OMIM:300633;Spinal and bulbar muscular atrophy of Kennedy, OMIM:313200		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARHGAP31	Adams-Oliver syndrome 1 100300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARHGAP6			Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARHGEF6	Intellectual developmental disorder, X-linked 46, OMIM:300436		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ARSF			Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ASCL1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ASMT			Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ASMTL			Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATCAY	Ataxia, cerebellar, Cayman type 601238		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATL1	Spastic paraplegia 3A, autosomal dominant, 182600;Hereditary spastic paraplegia;Intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP2A2	Darier disease 124200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP6V1B1	Renal tubular acidosis with deafness, 267300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP7B	Wilson disease 277900		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATP8B1	Cholestasis, progressive familial intrahepatic 1, 211600; Cholestasis, benign recurrent intrahepatic, 243300; Cholestasis, intrahepatic, of pregnancy, 1, 147480		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATXN1	Spinocerebellar ataxia 1, OMIM:164400		Other	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATXN10	Spinocerebellar ataxia 10, OMIM:603516		Other	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATXN2	Spinocerebellar ataxia 2, OMIM:183090;{Amyotrophic lateral sclerosis, susceptibility to, 13}, OMIM:183090;{Parkinson disease, late-onset, susceptibility to}, OMIM:168600		Other	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATXN3	Machado-Joseph disease, OMIM:109150		Other	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATXN3L			Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ATXN7	Spinocerebellar ataxia 7, OMIM:164500		Other	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AVP				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AVPR2			X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BEAN1	Spinocerebellar ataxia 31 117210		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BFSP2	Cataract 12, multiple types, 611597		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BHLHA9	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BIN1	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BMP15	Ovarian dysgenesis 2 300510; Premature ovarian failure 4 300510		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BMPER	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BMPR1B	Acromesomelic dysplasia, Demirhan type, OMIM:609441;Brachydactyly, type A1, D, OMIM:616849;Brachydactyly, type A2, OMIM:112600		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BRCA1	{Breast-ovarian cancer, familial, 1} 604370 autosomal dominant; {Pancreatic cancer, susceptibility to, 4} 614320 autosomal dominant; intellectual disability recessive		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BRCA2	Fanconi anemia, complementation group D1, OMIM:605724		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BRIP1	Fanconi anemia, complementation group J, OMIM:609054		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
BTK	Agammaglobulinemia and isolated hormone deficiency 307200; Agammaglobulinemia, X-linked 1 300755		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
C19orf12	?Spastic paraplegia 43, autosomal recessive 615043; Neurodegeneration with brain iron accumulation 4 614298		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
C2orf71	Retinitis pigmentosa 54, 613428		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
C9orf72	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, OMIM:105550		Other	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNA1F	Aland Island eye disease 300600 XL; Cone-rod dystrophy, X-linked, 3 300476 XLR; Night blindness, congenital stationary (incomplete), 2A, X-linked 300071		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNA1H	Hyperaldosteronism, familial, type IV 617027; {Epilepsy, childhood absence, susceptibility to, 6} 611942; {Epilepsy, idiopathic generalized, susceptibility to, 6} 611942		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNA1S	Hypokalemic periodic paralysis, type 1, 170400; {Malignant; hyperthermia susceptibility 5}, 601887; {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNA2D3				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CACNG2	?Mental retardation, autosomal dominant 10 614256		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CANT1				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CAPN10	{Diabetes mellitus, noninsulin-dependent 1} 601283		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43

CCDC103	Ciliary dyskinesia, primary, 17, 614679		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CCDC39	CILIARY DYSKINESIA, PRIMARY, 14		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CCDC40	Ciliary dyskinesia, primary, 15, 613808		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CCDC8	3-M syndrome, 614205;3M syndrome;Three M syndrome 3		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CCNA2	Autosomal recessive mental retardation		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CCT5	autosomal recessive mutilating sensory neuropathy with spastic paraplegia		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CDH15	Mental retardation, autosomal dominant 3, OMIM:612580		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CDH23	Usher syndrome, type 1D, 601067;Deafness, autosomal recessive; 12, 601386;Usher syndrome, type 1D/F digenic, 601067		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CDH3	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553;Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CDK5R1				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CDT1	Meier-Gorlin syndrome 4, OMIM:613804		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CFP	Properdin deficiency, X-linked 312060		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHM	Choroideremia, 303100		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHRDL1	Megalocornea 1, X-linked 309300		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHRNA2	Epilepsy, nocturnal frontal lobe, type 4 610353		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHRNA4	Epilepsy, nocturnal frontal lobe, 1 600513 AD; {Nicotine addiction, susceptibility to} 188890		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHRNB2	Epilepsy, nocturnal frontal lobe, 3 605375		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHRNG	Myasthenia gravis, neonatal transient; Escobar syndrome, 265000; Multiple pterygium syndrome, lethal type, 253290		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHSY1	Temtamy preaxial brachydactyly syndrome, 605282		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CHUK	Cocoon syndrome, 613630		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CIB2	Deafness, autosomal recessive 48, 609439; Usher syndrome, type IJ, 614869		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CISD2	WOLFRAM SYNDROME TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CLCN5	Dent disease		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CLCN7	Osteopetrosis, autosomal recessive 4, OMIM:611490;Osteopetrosis, autosomal dominant 2, OMIM:166600		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CLCNKA	Bartter syndrome, type 4b, digenic, OMIM:613090;Bartter disease type 4B, MONDO:0000909		Other	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CLCNKB	Bartter syndrome, type 3, OMIM:607364;Bartter disease type 3, MONDO:0011822;Bartter syndrome, type 4b, digenic, OMIM:613090;Bartter disease type 4B, MONDO:0000909		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CLIC2	Intellectual developmental disorder, X-linked syndromic 32, OMIM:300886		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CNKSR1	AUTOSOMAL RECESSIVE MENTAL RETARDATION (G2P)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CNTN4				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COA5	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3 616500;MITOCHONDRIAL COMPLEX IV DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL10A1	Metaphyseal chondrodysplasia, Schmid type, 156500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL11A1	Stickler syndrome, type II, 604841;Marshall syndrome, 154780;{Lumbar disc herniation, susceptibility to}, 603932;Fibrochondrogenesis, 228520		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL11A2	Gene2Phenotype confirmed gene with ID HPO		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL18A1	Knobloch syndrome, type 1, 267750		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL1A1	Osteogenesis imperfecta, type I, 166200; OI type II, 166210; OI type III, 259420; OI type IV, 166220; Ehlers-Danlos syndrome, type I, 130000; Ehlers-Danlos syndrome, type VIIA, 130060; {Osteoporosis}, 166710; Caffey disease, 114000; [Bone mineral density variation QTL], 166710		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL1A2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL25A1				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL2A1	Stickler syndrome, type I, 108300; Kniest dysplasia, 156550; Achondrogenesis, type II or hypochondrogenesis, 200610; SED congenita, 183900; SMED Strudwick type, 184250; Epiphyseal dysplasia, multiple, with myopia and deafness, 132450; Spondyloperipheral dysplasia, 271700; SED, Namaqualand type; Osteoarthritis with mild chondrodysplasia, 604864; Vitreoretinopathy w		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL4A3	Alport syndrome, autosomal recessive, 203780;Hematuria, benign familial, 141200;Alport syndrome, autosomal dominant, 104200		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL4A4	Alport syndrome 2, autosomal recessive, OMIM:203780;Hematuria,familial benign, OMIM:141200		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL4A6	?Deafness, X-linked 6 300914		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL6A1	Bethlem myopathy 1, OMIM:158810;Ullrich congenital muscular dystrophy 1, OMIM:254090		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL6A3	Bethlem myopathy, OMIM:158810;Ullrich congenital muscular dystrophy, OMIM:254090		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL9A1	Stickler syndrome, type IV, OMIM:614134;Epiphyseal dysplasia, multiple, 6, OMIM:614135		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL9A2	Stickler syndrome, type V, OMIM:614284;Epiphyseal dysplasia, multiple, 2, OMIM:600204		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy, OMIM:600969		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COMP	Pseudoachondroplasia, 177170;Epiphyseal dysplasia, multiple 1, 132400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COQ2	Coenzyme Q10 deficiency, primary, 1 607426;{Multiple system atrophy, susceptibility to} 146500		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COQ5	AUTOSOMAL RECESSIVE MENTAL RETARDATION;cerebellar ataxia and static encephalomyopathy due to COQ5 C-methyltransferase deficiency		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COX14	?Mitochondrial complex IV deficiency 220110		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
COX6B1	Mitochondrial complex IV deficiency, nuclear type 7, OMIM:619051		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CP	[Hypoceruloplasminemia, hereditary], 604290; Cerebellar ataxia, 604290; Hemosiderosis, systemic, due to aceruloplasminemia, 604290		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CPA6	Epilepsy, familial temporal lobe, 5		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRB1	Retinitis pigmentosa-12, autosomal recessive, 600105; Leber congenital amaurosis 8, 613835; Pigmented paravenous chorioretinal atrophy, 172870		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRX	Cone-rod retinal dystrophy-2, 120970;Leber congenital; amaurosis 7, 613829		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRYAA	Cataract 9, multiple types, 604219		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRYBA1	Cataract 10, multiple types, 600881		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRYBA4	Cataract 23 610425		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRYBB1	Cataract 17, multiple types, 611544		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRYBB2	Cataract 3, multiple types, 601547		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRYBB3	Cataract 22, OMIM:609741		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRYGC	Cataract 2, multiple types 604307		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CRYGD	Cataract 4, multiple types, 115700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CSF2RA	Surfactant metabolism dysfunction, pulmonary, 4 300770		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CTGF			Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CTNS	Cystinosis, nephropathic, 219800; Cystinosis, ocular; nonnephropathic, 219750; Cystinosis, late-onset juvenile or adolescent nephropathic, 219900; Cystinosis, atypical nephropathic, 219800;		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CTSK	Pycnodysostosis, 265800		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CTTNBP2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CUL7	3-M syndrome 1 273750		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CYP1B1	Glaucoma 3A, primary open angle, congenital, juvenile, or adult; onset, 231300;Peters anomaly, 604229		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
CYP7B1	Spastic paraplegia 5A, autosomal recessive , OMIM:270800		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DCTN1	Neuropathy, distal hereditary motor, type VIIB, 607641; {Amyotrophic lateral sclerosis, susceptibility to}, 105400; Perry syndrome, 168605		Other - please specify in evaluation comments	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DDB2	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DDR2	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DDX58			MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DECR1	2,4-DIENOYL-COA REDUCTASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DHODH	Miller syndrome OMIM: 126064		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DIAPH2			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DIP2B	Mental retardation, FRA12A type, 136630;MENTAL RETARDATION, FRA12A TYPE		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DLGAP2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DLL3	Spondylocostal dysostosis 1, autosomal recessive, 277300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DMP1	Hypophosphatemic rickets, AR, 241520		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DMPK	Myotonic dystrophy 1, OMIM:160900		Other	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DNAAF3	Ciliary dyskinesia, primary, 2, 606763		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DNM2	Charcot Marie Tooth disease, axonal, type 2M, 606482;Charcot Marie Tooth disease, dominant intermediate B, 606482		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant;Neuropathy, hereditary sensory, type IE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DPP6	Intellectual developmental disorder, autosomal dominant 33, OMIM:616311		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DRD2	Dystonia, myoclonic, 159900		Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DSCAM				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DSPP	Dentinogenesis imperfecta, Shields type II, 125490;Deafness, autosomal dominant 36, with dentinogenesis, 605594;Dentinogenesis imperfecta, Shields type III, 125500;Dentin dysplasia, type II, 125420		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DST	Neuropathy, hereditary sensory and autonomic, type VI, 614653Epidermolysis bullosa simplex, sutosomal recessive 2, 615425			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly, OMIM:613091		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EDA	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100;Tooth; agenesis, selective, X-linked 1, 313500		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EDNRA	Migraine, resistance to, 157300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EDNRB	ABCD syndrome, 600501;ALBINISM, BLACK LOCK, CELL MIGRATION DISORDER OF THE NEUROCYTES OF THE GUT, AND DEAFNESS		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EFHC1	{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770; {Epilepsy, juvenile absence, susceptibility to, 1}, 607631		Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EGR2	Gene2Phenotype confirmed gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EIF4G1	Parkinson disease 18, 614251		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ELK1			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ELN	Supravalvar aortic stenosis, 185500;Cutis laxa, AD, 123700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EN2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ENPP1	{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853;{Obesity, susceptibility to}, 601665;Arterial calcification, generalized, of infancy, 1, 208000;Hypophosphatemic rickets, autosomal recessive, 2, 613312;Cole disease, 615522		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EOMES	POLYMICROGYRIA AND CORPUS CALLOSUM AGENESIS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora) 254780		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ERCC4	Xeroderma pigmentosum, group F, 278760; XFE progeroid syndrome, 610965; Fanconi anemia, complementation group Q, 615272; Xeroderma pigmentosum, type F/Cockayne syndrome, 278760		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EVC	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EVC2	Ellis-van Creveld syndrome, 225500		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EXT1	Gene2Phenotype confirmed gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
EYA1	Branchiootorenal syndrome 1, with or without cataracts, 113650; Anterior segment anomalies with or without cataract, 113650; Branchiootic syndrome 1, 602588; ?Otofaciocervical syndrome, 166780		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
F5				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FA2H	Spastic paraplegia 35, autosomal recessive, 612319;spastic paraplegia with ID;cognitive defects;Seizures		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FAH	TYROSINEMIA TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FAM161A	Retinitis pigmentosa 28, 606068		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FAM20A	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FAM58A	STAR syndrome 300707		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FANCB	Fanconi anaemia complementation group B presenting as X linked VACTERL with hydrocephalus syndrome		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FASN	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FBLN5				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FBN1	Marfan syndrome 154700;MASS syndrome 604308;Weill-Marchesani syndrome 2, dominant 608328;Marfan lipodystrophy syndrome 616914		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FBN2	Gene2Phenotype confirmed gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FBP1	Fructose-1,6-bisphosphatase deficiency, 229700		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FBXO7	Parkinson disease 15, autosomal recessive 260300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FBXW4	SPLIT-HAND/FOOT MALFORMATION TYPE 3			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FGD4	Charcot Marie Tooth disease, type 4H, 609311		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FGF10	Aplasia of lacrimal and salivary glands, 180920;LADD syndrome, 149730		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FGFR1	Hartsfield syndrome, OMIM:615465;Pfeiffer syndrome, OMIM:101600;Encephalocraniocutaneous lipomatosis, somatic mosaic, OMIM:613001		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FGFR3	CATSHL syndrome 610474;Hypochondroplasia 146000;SADDAN 616482;Muenke syndrome 602849;Thanatophoric dysplasia, type I 187600		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FHL1	Scapuloperoneal myopathy, X-linked dominant, 300695; Myopathy, X-linked, with postural muscle atrophy, 300696; Myopathy, reducing body, X-linked, severe early-onset, 300717; Myopathy, reducing body, X-linked, childhood-onset, 300718; Emery-Dreifuss muscular dystrophy 6, X-linked, 300696		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FKBP14	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FKBP6	Williams-Beuren syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FKBPL				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FLNA	Heterotopia, periventricular, 1, OMIM:300049;Otopalatodigital syndrome, type II, OMIM:304120;?FG syndrome 2, OMIM:300321		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FLNB	Spondylocarpotarsal synostosis syndrome, 272460; Larsen syndrome, 150250; Atelosteogenesis, type I, 108720; Atelosteogenesis, type III, 108721; Boomerang dysplasia, 112310		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FLT4	Lymphedema, hereditary, IA, 153100;Hemangioma, capillary; infantile, somatic, 602089		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FLVCR1	ATAXIA, POSTERIOR COLUMN, WITH RETINITIS PIGMENTOSA		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOXC1	Iridogoniodysgenesis, type 1, 601631;Rieger or Axenfeld; anomalies, 602482;Axenfeld-Rieger syndrome, type 3, 602482;Iris hypoplasia and glaucoma, 601631		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOXC2	Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis; syndrome with renal disease and diabetes mellitus, 153400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOXE1	Bamforth-Lazarus syndrome, 241850		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOXE3	Anterior segment mesenchymal dysgenesis, 107250;Aphakia, congenital primary, 610256		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked 304790		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FREM1	Bifid nose with or without anorectal and renal anomalies 608980;Manitoba oculotrichoanal syndrome 248450;Trigonocephaly 2 614485		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FRMD7	Nystagmus 1, congenital, X-linked 310700;Nystagmus, infantile periodic alternating, X-linked 310700		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FTL	Neurodegeneration with brain iron accumulation 3 606159;Hyperferritinemia-cataract syndrome;L-ferritin deficiency, dominant and recessive		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FXN	Friedreich ataxia, OMIM:229300;Friedreich ataxia with retained reflexes, OMIM:229300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FYCO1	Cataract 18, autosomal recessive, 610019		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FZD3				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FZD6	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
G6PC3				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GAA	Glycogen storage disease II, 232300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GABRG3				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GALK1	Galactokinase deficiency with cataracts, 230200		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GALNS	MUCOPOLYSACCHARIDOSIS TYPE 4A (MPS4A)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GATA2	Immunodeficiency 21, 614172;Emberger syndrome, 614038;{Myelodysplastic syndrome, susceptibility to}, 614286;{Leukemia, acute; myeloid, susceptibility to}, 601626		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GATA4	Atrial septal defect 2, 607941;Ventricular septal defect 1, 614429;Atrioventricular septal defect 4, 614430;?Testicular anomalies with or without congenital heart disease, 615542;Tetralogy of; Fallot, 187500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GBE1				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GCK				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GDAP1	Charcot-Marie-Tooth disease, axonal, type 2K, OMIM:607831;Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, OMIM:607706;Charcot-Marie-Tooth disease, recessive intermediate, A, OMIM:608340;Charcot-Marie-Tooth disease, type 4A, OMIM:214400		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GDF5	Acromesomelic dysplasia, Hunter-Thompson type, 201250;Brachydactyly, type C, 113100;Chondrodysplasia, Grebe type, 200700;Du Pan syndrome, 228900;Brachydactyly, type A2, 112600;Symphalangism, proximal, 1B, 615298;Multiple synostoses syndrome 2, 610017;{Osteoarthritis-5}, 612400;Brachydactyly, type A1, C, 615072		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GDF6	Klippel-Feil syndrome 1, autosomal dominant, 118100;Microphthalmia, isolated 4, 613094;Microphthalmia with coloboma 6, digenic, 613703;Leber congenital amaurosis 17, 615360		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GHR	Laron dwarfism, OMIM:262500;Growth hormone insensitivity, partial, OMIM:604271;Increased responsiveness to growth hormone, OMIM:604271		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GIGYF2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GJA1	Gene2Phenotype confirmed gene with ID HPO		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GJA3	Cataract 14, multiple types, 601885		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GJA8	Cataract 1, multiple types, 116200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GJB2	Deafness, autosomal recessive 1A, 220290;Deafness, autosomal; dominant 3A, 601544;Vohwinkel syndrome, 124500;Keratoderma, palmoplantar, with deafness, 148350;Keratitis-ichthyosis-deafness; syndrome, 148210;Hystrix-like ichthyosis with deafness, 602540;Bart-Pumphrey syndrome, 149200		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLE1	Lethal congenital contracture syndrome 1, 253310; Arthrogryposis, lethal, with anterior horn cell disease, 611890		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLMN	Glomuvenous malformations, 138000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLRA1	Hyperekplexia;developmental delay;infantile spasms and generalized tonic-clonic seizures		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLUD1	Gene2Phenotype confirmed gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GON4L	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43

GORAB	Geroderma osteodysplasticum		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GOSR2	Epilepsy, progressive myoclonic 6, 614018		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GPHN	Molybdenum cofactor deficiency C, OMIM:615501		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GPR179	Night blindness, congenital stationary (complete), 1E, autosomal; recessive, 614565		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GRIP1				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GRM6	Night blindness, congenital stationary (complete), 1B, autosomal; recessive, 257270		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GRN	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485; Aphasia, primary progressive, 607485; Ceroid lipofuscinosis, neuronal, 11, 614706		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GSPT2	XL INTELLECTUAL DISABILITY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GUCY2C	Diarrhea 6, 614616;Meconium ileus, 614665		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GYS2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HADH	Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HARS2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HIST3H3	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HMGCS2	HMG-CoA synthase-2 deficiency, 605911		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HNF4A	MODY, type I, 125850; {Diabetes mellitus, noninsulin-dependent}, 125853; Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HOXA13	Hand-foot-uterus syndrome, 140000;Guttmacher syndrome, 176305		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HOXD10				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HOXD13	Synpolydactyly, type II, 186000;Brachydactyly, type E, 113300;Brachydactyly, type D, 113200;Synpolydactyly with foot anomalies, 186000;Syndactyly, type V, 186300;Brachydactyly-syndactyly syndrome, 610713;?VACTERL association, 192350		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HPGD	Cranioosteoarthropathy, 259100;Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100;Digital clubbing, isolated congenital, 119900		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HPS1	Hermansky-Pudlak syndrome 1, 203300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HPSE2	Urofacial syndrome 1, 236730		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HR	Alopecia universalis, 203655;Atrichia with papular lesions, 209500;Hypotrichosis 4, 146550		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HSD3B7	Bile acid synthesis defect, congenital, 1, 607765		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HSF4	Cataract 5, multiple types, OMIM:116800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HYAL1	Mucopolysaccharidosis type IX, 601492		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HYDIN	CILIARY DYSKINESIA, PRIMARY, 5; CILD5		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
HYLS1	HYDROLETHALUS SYNDROME TYPE 1 (HLS1)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IFITM5	Osteogenesis imperfecta, type V, 610967		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IFNAR2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IFT122	Cranioectodermal dysplasia 1, 218330		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IFT80	NA		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IGF2	Beckwith-Wiedemann Syndrome; Chromosome 11p15.5-Related Russell-Silver Syndrome		Other - please specify in evaluation comments	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IGHMBP2	Neuronopathy, distal hereditary motor, type VI, 604320;Charcot-Marie-Tooth disease, axonal, type 2S, 616155		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IHH	Acrocapitofemoral dysplasia, OMIM:607778;Brachydactyly, type A1, OMIM:112500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IL11RA	Crouzon-like craniosynostosis; Autosomal Recessive Craniosynostosis		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IMPAD1	Chondrodysplasia with joint dislocations, GRAPP type, 614078		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
INF2	Glomerulosclerosis, focal segmental, 5, 613237Charcot-Marie-Tooth disease, dominant intermediate E, 614455			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
INSR				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IRAK1	Lubs X-Linked Mental Retardation Syndrome; MRXSL		Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
IRF6	van der Woude syndrome, 119300;Popliteal pterygium syndrome 1, 119500;Orofacial cleft 6, 608864		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ITCH	Autoimmune disease, multisystem, with facial dysmorphism, 613385		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis;bullosa, congenital, 614748		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ITGA4			Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ITIH6			Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
JAG1	Alagille syndrome 1, OMIM:118450		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
JAK3	SCID, autosomal recessive, T-negative/B-positive type, 600802		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
JPH3	Neurodevelopmental disorder, MONDO:0700092;Paroxysmal dystonia, MONDO:0016058		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KANK1	CEREBRAL PALSY SPASTIC QUADRIPLEGIC TYPE 2 (CPSQ2)		Other - please specify in evaluation comments	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KATNAL2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KBTBD13	Nemaline myopathy 6, autosomal dominant, 609273		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCND3	Spinocerebellar ataxia 19, 607346		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNE1	Jervell and Lange-Nielsen syndrome 2, OMIM:612347;Long QT syndrome 5, OMIM:613695		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KCNQ1	Long QT syndrome 1, 192500;Jervell and Lange-Nielsen syndrome, 220400;Atrial fibrillation, familial, 3, 607554;Short QT syndrome 2, 609621;{Long QT syndrome 1, acquired, susceptibility to}, 192500		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KIF1B	Hypotonia;coloboma, MONDO:0001476;hypoplasia of the corpus callosum;severe neurodevelopmental delay		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KIF21A				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KIT	Piebaldism, 172800;Gastrointestinal stromal tumor, familial, 606764;Mast cell disease, 154800;Leukemia, acute myeloid, 601626;Germ cell tumors, 273300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KLF1	Blood group--Lutheran inhibitor, 111150;[Hereditary; persistence of fetal hemoglobin], 613566;Dyserythropoietic anemia, congenital, type IV, 613673		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
KRIT1	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 1 (CCM1)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LBR	?Reynolds syndrome		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LDB3	Myopathy, myofibrillar, 4, 609452; Cardiomyopathy, dilated 1C, 601493; Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LEMD3	Osteopoikilosis, 166700;Buschke-Ollendorff syndrome, 166700;Melorheostosis with osteopoikilosis, 155950		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LFNG	?Spondylocostal dysostosis 3, autosomal recessive, 609813		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LGI1	Epilepsy, familial temporal lobe, 1 600512;AUTOSOMAL DOMINANT PARTIAL EPILEPSY WITH AUDITORY FEATURES		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LHX3	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LHX4	Pituitary hormone deficiency, combined, 4, 262700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LIMK1	Williams-Beuren syndrome			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LITAF	Charcot-Marie-Tooth disease, type 1C, 601098		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LMNA	Emery-Dreifuss muscular dystrophy 2, AD, 181350; Cardiomyopathy, dilated, 1A, 115200; Lipodystrophy, familial partial, 2, 151660; Emery-Dreifuss muscular dystrophy 3, AR, 181350; Charcot-Marie-Tooth disease, type 2B1, 605588; Muscular dystrophy, congenital, 613205; Muscular dystrophy, limb-girdle, type 1B, 159001; Mandibuloacral dysplasia, 248370; Hutchinson-Gilford		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LMX1B	Nail-patella syndrome, 161200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LOXHD1	Deafness, autosomal recessive 77, 613079		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LRAT	LEBER CONGENITAL AMAUROSIS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LRP1				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LRP4	Cenani-Lenz syndactyly syndrome, 212780;Sclerosteosis 2, 614305		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LRRC6	Ciliary dyskinesia, primary, 19, 614935		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LRRK2	Parkinson disease 8, 607060		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LTBP2	Glaucoma 3, primary congenital, D, 613086;Microspherophakia; and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750;Weill-Marchesani syndrome 3, recessive, 614819		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LTBP3	Tooth agenesis, selective, 6, 613097		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
LYST	Gene2Phenotype confirmed gene with ID HPO;Chediak-Higashi syndrome, 214500		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAFB	Duane retraction syndrome 3 (617041);Multicentric carpotarsal osteolysis syndrome (166300)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAGEE2			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAGI2	Epileptic encephalopathy;Infantile spasms		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAOB			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAP3K1	46XY sex reversal 6, 613762		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAP3K15			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAP7D3	Intellectual disability		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MAPT	Dementia, frontotemporal, with or without parkinsonism, 600274; Pick disease, 172700; Supranuclear palsy, progressive, 601104; Supranuclear palsy, progressive atypical, 260540; {Parkinson disease, susceptibility to}, 168600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MARS2	?Combined oxidative phosphorylation deficiency 25 (616430) (global developmental delay)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MATN3	Epiphyseal dysplasia, multiple, 5, 607078;{Osteoarthritis; susceptibility 2}, 140600;Spondyloepimetaphyseal dysplasia, 608728		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MCEE	Methylmalonyl-CoA epimerase deficiency		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MEGF10	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MESP2	Spondylocostal dysostosis 2, autosomal recessive, 608681		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MET				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MFRP	NA		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MGP	Keutel syndrome 245150		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MITF	Waardenburg syndrome, type 2A, 193510; Waardenburg; syndrome/ocular albinism, digenic, 103470; Tietz albinism-deafness syndrome, 103500; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MLH1	Mismatch repair cancer syndrome		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MMP13	Spondyloepimetaphyseal dysplasia, Missouri type, 602111;Metaphyseal anadysplasia 1, 602111		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MNX1	Gene2Phenotype confirmed gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MPDZ	Hydrocephalus, nonsyndromic, autosomal recessive 2		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MPI	Congenital disorder of glycosylation, type Ib, OMIM:602579;MPI-CDG, MONDO:0011257		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MPZ	Associated with Charcot-Marie-Tooth disease, dominant intermediate D (607791);Charcot-Marie-Tooth disease, type 1B		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MRAP				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MSX1	Ectodermal dysplasia 3, Witkop type, OMIM:189500;Orofacial cleft 5, OMIM:608874;Tooth agenesis, selective, 1, with or without orofacial cleft, OMIM:106600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MSX2	Craniosynostosis, type 2, 604757;Parietal foramina 1, 168500;Parietal foramina with cleidocranial dysplasia, 168550		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MTM1	Myopathy, centronuclear, X-linked, OMIM:310400			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MTMR14	{Centronuclear myopathy, autosomal, modifier of}, 160150;Autosomal dominant centronuclear myopathy		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MTMR2	Charcot-Marie-Tooth disease, type 4B, 601382		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MTPAP	?Spastic ataxia 4, autosomal recessive		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MTTP	Abetalipoproteinemia, 200100		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MYBPC1	Arthrogryposis, distal, type 1B 614335 AD;Lethal congenital contracture syndrome 4 614915 AR		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MYH3	Gene2Phenotype confirmed gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MYH6	Cardiomyopathy, familial hypertrophic, 14, 613251;Atrial septal; defect 3, 614089;Cardiomyopathy, dilated, 1EE, 613252;{Sick sinus syndrome 3}, 614090		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MYH8	Carney complex variant, 608837;Trismus-pseudocamptodactyly; syndrome, 158300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MYH9	May-Hegglin anomaly, 155100;Fechtner syndrome, 153640;Sebastian syndrome, 605249;Deafness, autosomal dominant 17, 603622;Epstein syndrome, 153650;Macrothrombocytopenia and progressive; sensorineural deafness, 600208		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MYO5B	Diarrhea 2, with microvillus atrophy, OMIM:251850		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MYO7A	Gene2Phenotype both DD and IF gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
MYT1	Intellectual disability, MONDO:0001071		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDN	Smith-Magenis-like syndrome		MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDRG1	Charcot-Marie-Tooth disease, type 4D, 601455		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFA10	Leigh syndrome, 256000;Leigh disease with leukodystrophy;Nuclear Gene-Encoded Leigh syndrome		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFA11	Mitochondrial complex I deficiency 252010			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFA12	Mitochondrial complex I deficiency, nuclear type 23, OMIM:618244		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFA9	Leigh syndrome due to mitochondrial complex I deficiency, 256000;Leigh disease with leukodystrophy;Nuclear Gene-Encoded Leigh syndrome		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFAF3	Mitochondrial complex I deficiency		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFS2	Mitochondrial complex I deficiency		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NEB	Nemaline myopathy 2, autosomal recessive, 256030		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NEFL	Charcot Marie Tooth disease, type 2E, 607684;Charcot Marie Tooth disease, type 1F, 607734		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NEK1	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NGF				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, OMIM:611291		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora), 254780		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NIPA1	Spastic paraplegia 6, autosomal dominant, 600363;Non Imprinted In Prader-Willi/Angelman Syndrome 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NKX2-5	Atrial septal defect 7, with or without AV conduction defects, 108900;Tetrology of Fallot, 187500;Hypothyroidism, congenital nongoitrous, 5, 225250;Ventricular septal defect 3, 614432;Hypoplastic left heart syndrome 2, 614435;Conotruncal heart malformations, variable, 217095		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NLGN4X	SUSCEPTIBILITY TO AUTISM X-LINKED TYPE 2 (AUTSX2)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NLRP3	CINCA syndrome, OMIM:607115		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NMNAT1	Leber congenital amaurosis 9, 608553		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NODAL	Heterotaxy, visceral, 5, 270100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NOG	Symphalangism, proximal, 185800;Multiple synostoses syndrome 1, 186500;Tarsal-carpal coalition syndrome, 186570;Stapes ankylosis with broad thumb and toes, 184460;Brachydactyly, type B2, 611377		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NOP56	Spinocerebellar ataxia 36, OMIM:614153		Other	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NOTCH2	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NPHP4	Nephronophthisis 4, 606966;Senior-Loken syndrome 4, 606996		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NPHS1	Nephrotic syndrome, type 1, 256300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NPHS2	Nephrotic syndrome, type 2, 600995		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NPR2	Acromesomelic dysplasia, Maroteaux type, 602875;Epiphyseal chondrodysplasia, Miura type, 615923		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NPR3				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NR1I3	EHMT1-like Intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NR5A1	46XY sex reversal 3, 612965;Premature ovarian failure 7, 612964;Adrenocortical insufficiency;Spermatogenic failure 8, 613957		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NRXN2	autism spectrum disorder;intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NRXN3	Autism spectrum disorder		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NTNG1				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
NXF5	Intellectual Disability		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
OBSL1	3-M Syndrome 2, 612921		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ORC1	Meier-Gorlin syndrome 1, OMIM:224690		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ORC4	Meier-Gorlin syndrome 2, OMIM:613800		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ORC6	Meier-Gorlin syndrome 3, OMIM:613803		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
OXCT1	Succinyl-CoA: 3-Oxoacid CoA Transferase (SCOT) Deficiency;SUCCINYL-COA-3-KETOACID-COA TRANSFERASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
P2RY4			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PALB2	Fanconi anemia, complementation group N, 610832;{Breast cancer, susceptibility to}, 114480;{Pancreatic cancer, susceptibility to, 3}, 613348		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PANK2	Neurodegeneration with brain iron accumulation 1, 234200;HARP syndrome, 607236		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PAPSS2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PARK7	Parkinson disease 7, autosomal recessive early-onset, 606324		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PARP1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PAX2	Gene2Phenotype confirmed gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PAX3	Waardenburg syndrome, type 1, 193500;Waardenburg syndrome, type; 3, 148820;Craniofacial-deafness-hand syndrome, 122880;Rhabdomyosarcoma 2, alveolar, 268220		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PAX6	Aniridia, Cerebellar Ataxia, And Mental Retardation;Aniridia, 106210Peters anomaly, 604229Cataract with late-onset corneal dystrophy, 106210Keratitis, 148190Foveal hyperplasia, 136520Morning glory disc anomaly, 120430Optic nerve hypoplasia, 165550Coloboma, ocular, 120200Coloboma of optic nerve, 120430Gillespie syndrome, 206700;KERATITIS HEREDITARY (KERH		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PAX7	Hypotonia;Axial hypotonia;Ptosis;Scoliosis;Delayed motor milestones;Myopathy, congenital, progressive, with scoliosis, 618578		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PAX9	Tooth agenesis, selective, 3, 604625		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PCBD1	HYPERPHENYLALANINEMIA, BH4-DEFICIENT, D		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PDCD10	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 3 (CCM3)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43

PDE6G	Retinitis pigmentosa 57, 613582		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PDGFB	Meningioma, SIS-related, 607174; Dermatofibrosarcoma protuberans, 607907; Basal ganglia calcification, idiopathic, 5, 615483		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PDYN	Spinocerebellar ataxia 23, 610245		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PECR	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PGM1	Congenital disorder of glycosylation, type It, 614921		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PGRMC1			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PHKA1			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PHKA2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PHKG2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PHOX2B	CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL, WITH OR WITHOUT HIRSCHSPRUNG DISEASE		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PIEZO2	Gene2Phenotype confirmed gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PIK3C3				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PIK3R1	?Agammaglobulinemia 7, autosomal recessive, 615214; SHORT; syndrome, 269880; Immunodeficiency 36, 616005		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PINK1	Parkinson disease 6, early onset, 605909		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PITX2	Axenfeld-Rieger syndrome, type 1, 180500;Iridogoniodysgenesis, type 2, 137600;Ring dermoid of cornea, 180550;Peters anomaly, 604229		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PITX3	Anterior segment mesenchymal dysgenesis, 107250; Cataract 11, multiple types, 610623; Cataract 11, syndromic, 610623		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PKD1L1	Laterality defects		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PKHD1	Polycystic kidney and hepatic disease, 263200		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PLCE1	Nephrotic syndrome, type 3, 610725		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PLEC	Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, OMIM:616487;Epidermolysis bullosa simplex 5A, Ogna type, OMIM:131950;Epidermolysis bullosa simplex 5B, with muscular dystrophy, OMIM:226670;Epidermolysis bullosa simplex 5C, with pyloric atresia, OMIM:612138;Muscular dystrophy, limb-girdle, autosomal recessive 17, OMIM:613723		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PLOD1	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PLOD2	Bruck syndrome 2, 609220		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PLOD3	Lysyl hydroxylase 3 deficiency, 612394		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PMP22	Charcot-Marie-Tooth disease, type 1A, 118220; Dejerine-Sottas; disease, 145900; Neuropathy, recurrent, with pressure palsies, 162500; Charcot-Marie-Tooth disease, type 1E, 118300; Roussy-Levy syndrome, 180800; Neuropathy, inflammatory demyelinating, 139393		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PMS2	Mismatch repair cancer syndrome, 276300;Colorectal cancer, hereditary nonpolyposis, type 4, 614337		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PNKD	Paroxysmal nonkinesigenic dyskinesia, 118800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POLD1	{Colorectal cancer, susceptibility to, 10}, 612591;Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
POLR1D	Treacher Collins syndrome 2, OMIM:613717		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PPOX	Porphyria variegata, 176200			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PPP2R2B	Spinocerebellar ataxia 12, OMIM:604326		Other	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PREPL	Hypotonia-cystinuria syndrome, 606407;HCS;2p21 deletion syndrome;Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRICKLE1	Epilepsy, progressive myoclonic 1B, OMIM:612437		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRKAR1A	Acrodysostosis 1, with or without hormone resistance, OMIM:101800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRKCG	Spinocerebellar ataxia 14, 605361		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRKRA	Dystonia 16, 612067		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PROP1	Pituitary hormone deficiency, combined, 2, 262600		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRSS56	Microphthalmia, isolated 6, 613517		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PRX	Charcot-Marie-Tooth disease, type 4F, OMIM:614895;Dejerine-Sottas disease, OMIM:145900		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PSAT1	Phosphoserine aminotransferase deficiency, OMIM:610992;Neu-Laxova syndrome 2, OMIM:616038		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PSEN1	Alzheimer disease, type 3, 607822; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822; Dementia, frontotemporal, 600274; Pick disease, 172700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PTPN21				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
PYGL	Glycogen storage disease VI, 232700		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
QKI	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAB27A	Griscelli syndrome, type 2, 607624			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAB40AL	Mental retardation, X-linked, syndromic, Martin-Probst type, 300519		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RABL6	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAD50	Nijmegen breakage syndrome-like disorder, OMIM:613078		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAD51C	FANCONI ANEMIA, COMPLEMENTATION GROUP 0		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RALGDS	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RANBP2	{Encephalopathy, acute, infection-induced, 3, susceptibility to}608033		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RAPSN	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RASA1	Parkes Weber syndrome, 608355;Capillary; malformation-arteriovenous malformation, 608354;Basal cell carcinoma, somatic, 605462		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RBFOX1				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RBM28	Alopecia, neurologic defects, and endocrinopathy syndrome,612079; Intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RBM8A	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RECQL4	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
REEP1	Spastic paraplegia 31, autosomal dominant, 610250; ?Neuronopathy, distal hereditary motor, type VB, 614751		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RET	Gene2Phenotype both DD and IF gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RFX6	Mitchell-Riley syndrome, 615710		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RGS7	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RIMS1				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RIPK4	Popliteal pterygium syndrome 2, lethal type, 263650		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RNF135	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RNF168	RIDDLE syndrome, 611943;Radiosensitivity-immunodeficiency-dysmorphic features-learning difficulties syndrome		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ROBO3	Gaze palsy, horizontal, with progressive scoliosis, 607313		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ROR2	Brachydactyly, type B1, OMIM:113000 (AD);Robinow syndrome, autosomal recessive, OMIM:268310 (AR)		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RPE65	Leber congenital amaurosis 2, 204100;Retinitis pigmentosa 20, 613794		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RPGR			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RPGRIP1	Leber congenital amaurosis 6, 613826;Cone-rod dystrophy 13, 608194		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RPS19	Diamond-Blackfan anemia 1, 105650		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RSPO4	Anonychia congenita, 206800		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RTN2	Spastic paraplegia 12, autosomal dominant, 604805		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RUNX2	Cleidocranial dysplasia, 119600;Cleidocranial dysplasia, forme; fruste, with brachydactyly, 119600;Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600;Metaphyseal dysplasia with maxillary; hypoplasia with or without brachydactyly, 156510		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RYR1	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
RYR3	EPILEPTIC ENCEPHALOPATHY		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SALL4	Duane-radial ray syndrome, 607323;IVIC syndrome, 147750		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SBDS	Shwachman-Bodian-Diamond syndrome, 260400		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SBF2	Charcot-Marie-Tooth disease, type 4B2, 604563		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCARF2	Van den Ende-Gupta syndrome, 600920		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCN4A	Hyperkalemic periodic paralysis, type 2, 170500; Paramyotonia; congenita, 168300; Myotonia congenita, atypical, acetazolamide-responsive, 608390; Myasthenic syndrome, acetazolamide-responsive, 614198; Hypokalemic periodic paralysis, type 2, 613345		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCN9A	Erythermalgia, primary, 133020; Paroxysmal extreme pain disorder, 167400, Insensitivity to pain, congenital, 243000; Febrile seizures, familial, 3B, 613863; Epilepsy, generalized, with febrile seizures plus, type 7, 613863; Small fiber neuropathy, 133020; {Dravet syndrome, modifier of}, 607208; HSAN2D, autosomal recessive, 243000		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCO1	Mitochondrial complex IV deficiency, nuclear type 4, OMIM:619048		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SCRIB	8Q24.3 DELETION-LIKE		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SEC23B	Dyserythropoietic anemia, congenital, type II, OMIM:224100		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SEMA3E	Severe Intellectual Disability with Cognitive Regression		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SETDB2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SETX	Ataxia-ocular apraxia-2, 606002; Amyotrophic lateral sclerosis; 4, juvenile, 602433		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SF3B4	Gene2Phenotype confirmed gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SGCA				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SGCE	Dystonia-11, myoclonic, 159900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SH3PXD2B	Frank-ter Haar syndrome, 249420		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SH3TC2	Charcot-Marie-Tooth disease, type 4C, 601596; Mononeuropathy of the median nerve, mild, 613353		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SHOX	Langer mesomelic dysplasia, OMIM:249700 (PR);Leri-Weill dyschondrosteosis, OMIM:127300 (PD);Short stature, idiopathic familial, OMIM:300582		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SIGMAR1	Amyotrophic lateral sclerosis 16, juvenile, 614373		Other - please specify in evaluation comments	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SIX1	Brachiootic syndrome 3, 608389;Deafness, autosomal dominant 23, 605192		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SIX5	Branchiootorenal syndrome 2, 610896		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SKIV2L	Trichohepatoenteric syndrome 2, 614602		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC20A2	Basal ganglia calcification, idiopathic, 1; 13600;Idiopathic basal ganglia calcification, adult onset; Basal ganglia calcification, idiopathic, childhood onset		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC22A5	Carnitine deficiency, systemic primary, OMIM:212140;systemic primary carnitine deficiency disease, MONDO:0008919		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC25A13				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC25A19	Microcephaly, Amish type, 607196		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC26A9				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC2A10	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC31A1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC52A3	Brown-Vialetto-Van Laere syndrome 1, 211530		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC5A2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC6A4				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC6A5	hyperekplexia (MIM:614618) and intellectual disability		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC7A7				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SLC9A9	{?Autism susceptibility 16}, 613410		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SMARCAL1	Schimke immunoosseous dysplasia, OMIM:242900		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SNCA	Parkinson disease 4, 605543; Dementia, Lewy body, 127750; Parkinson disease 1, 168601		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SNX3	microcephaly, microphthalmia, ectrodactyly, prognathism (MMEP); mental retardation		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SOBP	syndromic and nonsyndromic ID with psychosis; Mental retardation, anterior maxillary protrusion, and strabismus, 613671; MRAMS		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SOX17	Vesicoureteral reflux 3, 613674		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPAST	Spastic paraplegia 4, autosomal dominant, 182601		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPG21	Mast syndrome, 248900		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPG7	Spastic paraplegia 7, autosomal recessive, OMIM:607259;hereditary spastic paraplegia 7, MONDO:0011803		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPRY3			Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPTLC1				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SPTLC2	Neuropathy, hereditary sensory and autonomic, type IC, 613640		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SREBF2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SRPX2	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, OMIM:300643		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SRY	46XY SEX REVERSAL 1		Other - please specifiy in evaluation comments	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
STAR	Lipoid adrenal hyperplasia, 201710		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
STAT1	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892;Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796;Immunodeficiency 31C, autosomal dominant, 614162		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
STAT5B	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, OMIM:245590;Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, OMIM:618985		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
STS	Gene2Phenotype confirmed gene with ID HPO		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
STX11				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SYNE1	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998; Spinocerebellar ataxia, autosomal recessive 8, 610743; intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SYNE2	autism spectrum disorder, developmental delay and intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SYT14	psychomotor retardation			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
SYTL5			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TACO1	Mitochondrial complex IV deficiency, 220110 (includes mild to moderate developmental delay and intellectual disability)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TAF7L			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TARDBP	Amyotrophic lateral sclerosis 10, with or without FTD, 612069; Frontotemporal lobar degeneration, TARDBP-related, 612069		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBP	Spinocerebellar ataxia 17, OMIM:607136		Other	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBX15	Cousin Syndrome; Craniofacial Dysmorphism, Hypoplasia of Scapula and Pelvis, and Short Stature		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBX20	Atrial septal defect 4, 611363		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBX22	Cleft palate with ankyloglossia, 303400;?Abruzzo-Erickson; syndrome, 302905		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBX3	Ulnar-mammary syndrome, 181450		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBX4	Small patella syndrome, 147891		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBX5	Holt-Oram syndrome, 142900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TBXAS1	Ghosal hematodiaphyseal syndrome, OMIM:231095		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TCOF1	Gene2Phenotype confirmed gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TCTN1	Joubert syndrome 13, 614173; intellectual disability		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TECR	non-syndromic mental retardation			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TEK	Venous malformations, multiple cutaneous and mucosal, 600195		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TFAP2A	Branchiooculofacial syndrome, 113620		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TFAP2B	Char syndrome, 169100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TGFB2	Loeys-Dietz syndrome, type 4, 614816		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TGFB3	Arrhythmogenic right ventricular dysplasia 1, 107970;?Rienhoff syndrome, 615582		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TGFBR1	Loeys-Dietz syndrome 1, 609192 (includes uncommon developmental delay and mental retardation)			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TGFBR2	Loeys-Dietz syndrome 2, 610168			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TGM6	Spinocerebellar ataxia 35, 613908		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
THAP1	DYSTONIA 6, TORSION		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TIMM8A	Deafness, X-linked 1, progressiveMohr-Tranebjaerg syndrome, 304700Jensen syndrome, 311150;JENSEN SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TINF2	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TLR8			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TMEM135	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TMLHE	{Autism, susceptibility to, X-linked 6}, 300872 (includes Intellectual disability)		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TMPRSS6	Iron-refractory iron deficiency anemia, 206200		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TP63	ADULT syndrome, OMIM:103285;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, OMIM:604292;Hay-Wells syndrome, OMIM:106260;Limb-mammary syndrome, OMIM:603543;Orofacial cleft 8, OMIM:618149;Rapp-Hodgkin syndrome, OMIM:129400;Split-hand/foot malformation 4, OMIM:605289		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TPH2				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TPK1				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRAPPC2	NA		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRHR				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRIM32	Bardet-Biedl syndrome 11, 615988		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRIM37	MULIBREY NANISM;MUL;Muscle-liver-brain-eye nanism		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRIP11	Achondrogenesis, type IA, 200600		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRPM1	Night blindness, congenital stationary (complete), 1C, autosomal; recessive, 613216		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TRPS1	Trichorhinophalangeal syndrome, type I, 190350;Trichorhinophalangeal syndrome, type III, 190351		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43

TRPV4	Brachyolmia type 3, 113500;Spondylometaphyseal dysplasia, Kozlowski type, 184252;Metatropic dysplasia, 156530;Hereditary motor and sensory neuropathy, type IIc, 606071;Scapuloperoneal spinal; muscular atrophy, 181405;[Sodium serum level QTL 1], 613508;Parastremmatic dwarfism, 168400;SED, Maroteaux type, 184095;Spinal muscular atrophy, distal, congenital nonpro		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TSHR	Hypothyroidism, congenital, nongoitrous, 1 275200; Thyroid; adenoma, hyperfunctioning, somatic; Hyperthyroidism, nonautoimmune, 609152; Thyroid carcinoma with thyrotoxicosis; Hyperthyroidism, familial; gestational, 603373		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TTBK2	Spinocerebellar ataxia 11, 604432		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TTN				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TTPA	Ataxia with isolated vitamin E deficiency, 277460		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TTR				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TUBA8	Polymicrogyria with optic nerve hypoplasia, 613180;POLYMICROGYRIA WITH OPTIC NERVE HYPOPLASIA (PMGONH)		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TUFM	Combined oxidative phosphorylation deficiency 4, 610678		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TYR	Albinism, oculocutaneous, type IA, OMIM:203100;Albinism, oculocutaneous, type IB, OMIM:606952;Waardenburg syndrome/albinism, digenic, OMIM:103470		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
TYRP1	Albinism, oculocutaneous, type III, 203290;[Skin/hair/eye; pigmentation, variation in, 11 (Melanesian blond hair)], 612271		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UGT1A1	NA		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UQCRB	Mitochondrial complex III deficiency, nuclear type 3, OMIM:615158		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UROS	Porphyria, congenital erythropoietic, 263700		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
USB1	Poikiloderma with neutropenia		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
UVSSA	UV-sensitive syndrome 3, 614640		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
VAMP7			Unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
VDR	Gene2Phenotype confirmed gene with ID HPO		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
VIP	Asperger syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
VPS35	Parkinson disease 17, 614203		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
VSX2	Microphthalmia with coloboma 3, 610092;Microphthalmia, isolated 2, 610093		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WDR13	X-linked intellectual disability; XLID		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WDR19	Nephronophthisis 13, 614377;Cranioectodermal dysplasia 4, 614378;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WDR35	Cranioectodermal dysplasia 2, 613610;Short-rib thoracic; dysplasia 7 with or without polydactyly, 614091		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WFS1	Wolfram syndrome 1, 222300;Wolfram-like syndrome, autosomal dominant, 614296		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WNT10B	Split-hand/foot malformation 6, 225300		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WNT3	Tetra-amelia, autosomal recessive, 273395		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WNT4	MULLERIAN APLASIA AND HYPERANDROGENISM		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WNT5A	Gene2Phenotype confirmed gene with ID HPO		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WNT7A	Ulna and fibula, absence of, with severe limb deficiency, 276820;Fuhrmann syndrome, 228930		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WRAP53	Dyskeratosis congenita, autosomal recessive 3, 613988		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WRN				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
WT1	Wilms tumor, type 1, 194070;Denys-Drash syndrome, 194080;Nephrotic syndrome, type 4, 256370;Frasier syndrome, 136680;Meacham syndrome, 608978;Mesothelioma, somatic, 156240		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
XIAP			X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
XIST				Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
XK	X-linked mental retardation		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
XPC	Xeroderma pigmentosum, group C, 278720		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
XPNPEP3	Nephronophthisis-like nephropathy 1 OMIM:613159;nephronophthisis-like nephropathy 1 MONDO:0013163		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
YWHAE	neurodevelopmental disorder, MONDO:0700092		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZBTB16	Skeletal defects, genital hypoplasia, and mental retardation, 612447		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZBTB40	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZCCHC12	nonsyndromic X-linked mental retardation; XLMR		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZCCHC8	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZDHHC15	cerebral palsy;intellectual disability;autism spectrum disorder;epilepsy		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZIC3	Heterotaxy, visceral, 1, X-linked 306955;Congenital heart defects, nonsyndromic, 1, X-linked, 306955;VACTERL association, X-linked, 314390		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy, 608612; Restrictive dermopathy, lethal, 275210		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZNF41	Mental retardation, X-linked 89, 300848;Mental Retardation, X-linked		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZNF592	Spinocerebellar ataxia, autosomal recessive 5, 606937			Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZNF674	Mental Retardation, X-linked;Mental retardation, X-linked 92, 300851		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
ZNF81	Mental Retardation, X-linked;Mental retardation, X-linked 45, 300498		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
DMPK	Myotonic dystrophy 1, OMIM:160900		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
FMR1	Fragile X syndrome, OMIM:300624;Fragile X tremor/ataxia syndrome, OMIM:300623		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AFF3	neurodevelopmental disorder unspecified		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
GLS	Global developmental delay, progressive ataxia, and elevated glutamine, OMIM:618412		BIALLELIC, autosomal or pseudoautosomal	Intellectual disability	Neurodevelopmental disorders	Neurology and neurodevelopmental disorders	8.43
AAAS	ACHALASIA-ADDISONIANISM-ALACRIMA SYNDROME 231550		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AARS	EARLY-ONSET EPILEPTIC ENCEPHALOPATHY WITH PERSISTENT MYELINATION DEFECT		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AASS	Hyperlysinemia, OMIM:238700;Hyperlysinemia (disease), MONDO:0009388		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ABAT	ABAT-related GABA-transaminase Deficiency		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ABCB11	ABCB11-RELATED INTRAHEPATIC CHOLESTASIS 601847		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ABCB7	ANEMIA, SIDEROBLASTIC, WITH ATAXIA 301310		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
ABCC6	ARTERIAL CALCIFICATION, GENERALIZED, OF INFANCY, 2 614473		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ABCC9	CANTU SYNDROME HYPERTRICHOTIC OSTEOCHONDRODYSPLASIA 239850		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ABCD1	ADRENOLEUKODYSTROPHY, X-LINKED 300100		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
ABCD4	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, CBLJ TYPE 614857		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ABHD5	CHANARIN-DORFMAN SYNDROME 275630		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ACAD9	ACYL-COA DEHYDROGENASE FAMILY MEMBER TYPE 9 DEFICIENCY 611126		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ACADM	MEDIUM CHAIN ACYL-COENZYME A DEHYDROGENASE DEFICIENCY 201450		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ACADS	SHORT CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY 201470		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ACADVL	VERY LONG CHAIN ACYL-COENZYME A DEHYDROGENASE DEFICIENCY 201475		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ACAN	SPONDYLOEPIMETAPHYSEAL DYSPLASIA AGGRECAN TYPE 612813;SPONDYLOEPIPHYSEAL DYSPLASIA TYPE KIMBERLEY 608361		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
ACAT1	ALPHA-METHYLACETOACETIC ACIDURIA 203750		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ACBD5	ACBD5 deficiency, OMIM:618863		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ACO2	INFANTILE CEREBELLAR-RETINAL DEGENERATION 614559		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ACOX1	ADRENOLEUKODYSTROPHY PSEUDONEONATAL 264470		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ACP5	SPONDYLOENCHONDRODYSPLASIA WITH IMMUNE DYSREGULATION 607944		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ACSL4	MENTAL RETARDATION X-LINKED TYPE 63 300387;ALPORT SYNDROME WITH MENTAL RETARDATION MIDFACE HYPOPLASIA AND ELLIPTOCYTOSIS 300194		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
ACTA1	NEMALINE MYOPATHY 3, 161800		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ACTA2	AORTIC ANEURYSM, FAMILIAL THORACIC 6 611788;MOYAMOYA DISEASE 5 614042		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ACTB	BARAITSER-WINTER SYNDROME;ACTB Haploinsufficiency syndtome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ACTC1	ACTC1-related distal arthrogryposis with congenital heart disease		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ACTG1	BARAITSER-WINTER SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ACVR1	FIBRODYSPLASIA OSSIFICANS PROGRESSIVA 135100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ACY1	AMINOACYLASE-1 DEFICIENCY 609924		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ADA	ADENOSINE DEAMINASE DEFICIENCY 102700		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ADAMTS18	KNOBLOCH SYNDROME 2 608454		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ADAMTSL2	GELEOPHYSIC DYSPLASIA 1		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ADAR	AICARDI-GOUTIERES SYNDROME ASSOCIATED WITH A TYPE I INTERFERON SIGNATURE 615010;DYSCHROMATOSIS SYMMETRICA HEREDITARIA 1 127400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
ADCY5	ADCY5-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ADNP	MENTAL RETARDATION, AUTOSOMAL DOMINANT, 28 615873		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ADSL	ADENYLOSUCCINASE DEFICIENCY 103050		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AFF2	Intellectual developmental disorder, X-linked 109, OMIM:309548;FRAXE intellectual disability, MONDO:0010659		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
AFF3	Skeletal dysplasia with severe neurological disease		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
AGA	ASPARTYLGLUCOSAMINURIA 208400		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AGK	SENGERS SYNDROME 212350		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AGL	GLYCOGEN STORAGE DISEASE TYPE III 232400		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AGO1	AGO1-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
AGPS	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA TYPE 3 600121		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AGXT	HYPEROXALURIA, PRIMARY, TYPE 1 259900		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AHI1	JOUBERT SYNDROME 614615		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AIFM1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 6 300816;COWCHOCK SYNDROME 310490		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
AIMP1	LEUKODYSTROPHY, HYPOMYELINATING, 3 260600		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AIPL1	LEBER CONGENITAL AMAUROSIS 4 604393		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AIRE	AUTOIMMUNE POLYENDOCRINOPATHY SYNDROME TYPE 1 240300		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AK2	RETICULAR DYSGENESIS 267500		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AKR1D1	BILE ACID SYNTHESIS DEFECT, CONGENITAL, 2 235555		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AKT1	PROTEUS SYNDROME 176920		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
AKT2	AKT2-related hypoinsulinemic hypoglycemia and hemihypertrophy, OMIM:240900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
AKT3	HEMIMEGALENCEPHALY AKT3 603387		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ALDH18A1	SPASTIC PARAPLEGIA 9, AUTOSOMAL DOMINANT 601162;CUTIS LAXA, AUTOSOMAL DOMINANT 3 616603;MENTAL RETARDATION-JOINT HYPERMOBILITY-SKIN LAXITY WITH OR WITHOUT METABOLIC ABNORMALITIES 612652		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
ALDH1A2	ALDH1A2-related diaphragmatic hernia and pulmonary hypoplasia		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALDH3A2	SJOEGREN-LARSSON SYNDROME 270200		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALDH4A1	HYPERPROLINEMIA TYPE 2 239510		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALDH5A1	SUCCINATE SEMIALDEHYDE DEHYDROGENASE DEFICIENCY 271980		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALDH7A1	PYRIDOXINE-DEPENDENT EPILEPSY 266100		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALDOA	GLYCOGEN STORAGE DISEASE XII 611881		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALG1	ALG1-CDG 300141		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALG11	ALG11-CDG 319490		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALG12	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1G 607143		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALG13	EPILEPTIC ENCEPHALOPATHY;CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IS 300884;EPILEPTIC ENCEPHALOPATHIES.		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
ALG2	ALG2-CDG 237149		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALG3	ALG3-CDG 237128		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALG6	ALG6-CDG 237124		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALG8	Congenital disorder of glycosylation, type Ih OMIM:608104;ALG8-CDG MONDO:0011969		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALG9	ALG9-CDG 300153		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALMS1	ALSTROM SYNDROME 203800		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALPL	HYPOPHOSPHATASIA 241500		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALS2	ALS2-RELATED DISORDERS 240656		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALX1	FRONTONASAL DYSPLASIA TYPE 3 136760		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALX3	FRONTONASAL DYSPLASIA TYPE 1 136760		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ALX4	PARIETAL FORAMINA 2 609597;FRONTONASAL DYSPLASIA 2 613451		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
AMER1	OSTEOPATHIA STRIATA WITH CRANIAL SCLEROSIS 300373		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
AMT	GLYCINE ENCEPHALOPATHY 605899		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ANAPC1	Rothmund-Thomson Syndrome Type 1		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ANKH	CHONDROCALCINOSIS 2 118600;CRANIOMETAPHYSEAL DYSPLASIA JACKSON TYPE 123000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ANKRD11	KBG SYNDROME 148050		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ANKRD26	THROMBOCYTOPENIA 2 188000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ANO5	LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 2L, OMIM:611307;GNATHODIAPHYSEAL DYSPLASIA, OMIM:166260		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ANTXR1	GAPO syndrome, OMIM:230740		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AP1S2	MENTAL RETARDATION X-LINKED TYPE 59 300630		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
AP3B2	Epileptic Encephalopathy with Optic Atrophy		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AP4B1	CEREBRAL PALSY SPASTIC QUADRIPLEGIC TYPE 5 614066		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AP4E1	CEREBRAL PALSY SPASTIC QUADRIPLEGIC TYPE 4 613744		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AP4M1	CEREBRAL PALSY SPASTIC QUADRIPLEGIC TYPE 3 612936		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AP4S1	CEREBRAL PALSY SPASTIC QUADRIPLEGIC TYPE 6 614067		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
APTX	ATAXIA WITH OCULOMOTOR APRAXIA 1 208920		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
AR	SPINAL AND BULBAR MUSCULAR ATROPHY, OMIM:313200;ANDROGEN INSENSITIVITY SYNDROME, OMIM:300068		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
ARFGEF2	PERIVENTRICULAR HETEROTOPIA WITH MICROCEPHALY 608097		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ARG1	ARGININEMIA 207800		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ARHGAP31	ADAMS-OLIVER SYNDROME 1 100300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ARHGEF9	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 8 300607		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
ARID1A	COFFIN-SIRIS SYNDROME 135900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ARID1B	MENTAL RETARDATION, AUTOSOMAL DOMINANT 12 135900;COFFIN SIRIS SYNDROME 135900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ARL6	RETINITIS PIGMENTOSA TYPE 55 613575;BARDET-BIEDL SYNDROME TYPE 3 209900		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ARSA	ARYLSULFATASE A DEFICIENCY 250100		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ARSB	MUCOPOLYSACCHARIDOSIS TYPE 6 253200		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ARSE	CHONDRODYSPLASIA PUNCTATA 1, X-LINKED 302950		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
ARX	MENTAL RETARDATION X-LINKED ARX-RELATED 300419;AGENESIS OF THE CORPUS CALLOSUM WITH ABNORMAL GENITALIA 300004;LISSENCEPHALY X-LINKED TYPE 2 300215;EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 1 308350;PARTINGTON SYNDROME 309510		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
ASAH1	FARBER LIPOGRANULOMATOSIS 228000;SPINAL MUSCULAR ATROPHY ASSOCIATED WITH PROGRESSIVE MYOCLONIC EPILEPSY 159950		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ASL	ARGININOSUCCINATE LYASE DEFICIENCY 207900		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ASNS	Asparagine synthetase deficiency, OMIM:615574		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ASPA	CANAVAN DISEASE 271900		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ASPM	PRIMARY AUTOSOMAL RECESSIVE MICROCEPHALY 279936		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ASS1	CITRULLINEMIA TYPE I 615700		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ASXL1	BOHRING-OPITZ SYNDROME 605039		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ATG7	ATG7-related intellectual disability and ataxia, OMIM:619422;developmental delay;ataxia		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ATIC	AICA-RIBOSURIA 608688		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ATL1	ATL1-associated hereditary spastic paraplegia		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ATM	ATAXIA-TELANGIECTASIA 208900		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ATN1	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, OMIM:618494		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ATP13A2	PARKINSON DISEASE 9 606693		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ATP1A2	ATP1A2-related epileptic encephalopathy;MIGRAINE, FAMILIAL HEMIPLEGIC, ATP1A2-related;Autosomal recessive ATP1A2-related neuronal migration disorder with epilepsy		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
ATP1A3	RAPID-ONSET DYSTONIA-PARKINSONISM 128235;ALTERNATING HEMIPLEGIA OF CHILDHOOD 104290		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ATP6V0A1	ATP6V0A1-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ATP6V1B1	DISTAL RENAL TUBULAR ACIDOSIS WITH DEAFNESS 267300		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ATP7A	SPINAL MUSCULAR ATROPHY, DISTAL, X-LINKED 3 300489;OCCIPITAL HORN SYNDROME 304150;MENKES DISEASE 309400		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1

ATP8B1	ATP8B1-RELATED INTRAHEPATIC CHOLESTASIS 211600		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
ATR	SECKEL SYNDROME TYPE 1 210600		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
ATRX	MENTAL RETARDATION SYNDROMIC X-LINKED WITH HYPOTONIC FACIES SYNDROME TYPE 1 309580;ALPHA-THALASSEMIA MENTAL RETARDATION SYNDROME X-LINKED NON-DELETION TYPE 301040		X-LINKED; hemizygous mutation in males; biallelic mutations in females	DDG2P			5.1
ALH1	1-METHYLGLUTACONIC ACIDURIA TYPE 1 250950		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
AUTS2	SYNDROMIC INTELLECTUAL DISABILITY 612150		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
B3GALT6	SPONDYLOEPIMETAPHYSEAL DYSPLASIA WITH JOINT LAXITY TYPE 1, 271640;EHLERS-DANLOS SYNDROME 130070		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
B4GALT1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IId, OMIM:607091		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
B4GALT7	EHLERS-DANLOS SYNDROME PROGEROID TYPE 130070		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
B9D1	MECKEL SYNDROME 9 614209		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BANF1	NESTOR-GUILLERMO PROGERIA SYNDROME 614008		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BAP1	BAP1-associated neurodevelopmental syndrome		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
BBS1	Bardet-Biedl syndrome 1 OMIM 209900,Bardet-Biedl syndrome 1 MONDO:0008854		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BBS10	BARDET-BIEDL SYNDROME TYPE 10 209900		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BBS12	BARDET-BIEDL SYNDROME TYPE 12 209900		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BBS2	BARDET-BIEDL SYNDROME TYPE 2 209900		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BBS4	BARDET-BIEDL SYNDROME TYPE 4 209900		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BBS5	BARDET-BIEDL SYNDROME TYPE 5 209900		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BBS7	BARDET-BIEDL SYNDROME TYPE 7 209900		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BBS9	BARDET-BIEDL SYNDROME TYPE 9 209900		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BCAP31	DEAFNESS, DYSTONIA, AND CENTRAL HYPOMYELINATION WITH DISORGANIZATION OF THE GOLGI APPARATUS		X-LINKED; hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
BCKDHA	MAPLE SYRUP URINE DISEASE 248600		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BCKDHB	MAPLE SYRUP URINE DISEASE 248600		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BCL11A	INTELLECTUAL DISABILITY		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
BCOR	MICROPHTHALMIA SYNDROMIC TYPE 2 300166		X-LINKED; hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
BCS1L	GRACILE SYNDROME 603358		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BFSP2	CATARACT AUTOSOMAL DOMINANT BFSP2-RELATED 611597		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
BHLHA9	SPLIT HAND AND FOOT MALFORMATION 225600;MESOAXIAL SYNOSTOTIC SYNDACTYLY WITH PHALANGEAL REDUCTION, MALIK-PERCIN TYPE 69432		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
BIN1	CENTRONUCLEAR MYOPATHY 2 255200		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BLM	BLOOM SYNDROME 210900		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BLOC1S6	HERMANSKY-PUDLAK SYNDROME 9 614171		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BMP2	Short stature, palatal anomalies, congenital heart disease, and skeletal malformations		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
BMP4	MICROPHTHALMIA, SYNDROMIC 6 607932;OROFACIAL CLEFT 11 600625		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
BMPER	DIAPHANOSPONDYLODYSOSTOSIS 608022		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BMPR1B	BRACHYDACTYLY TYPE A2 112600		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BNC2	Conjunctival Lower Urinary Tract Obstruction		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
BOLA3	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 2 614299		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BRAF	LEOPARD SYNDROME TYPE 3 613707;NOONAN SYNDROME TYPE 7 613706;CARDIOFACIOCUTANEOUS SYNDROME 115150		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
BRAT1	LETHAL NEONATAL RIGIDITY AND SEIZURE SYNDROME 614498		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BRCA1	INTELLECTUAL DISABILITY 614578		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BRCA2	FANCONI ANEMIA COMPLEMENTATION GROUP D TYPE 1 605724		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BRIP1	FANCONI ANEMIA, COMPLEMENTATION GROUP J 609054		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BRSK2	Neurodevelopmental Disorder		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
BRWD3	MENTAL RETARDATION X-LINKED TYPE 93 300659		X-LINKED; hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
BSND	BARTTER SYNDROME TYPE 4A 602522		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BTD	BIOTINIDASE DEFICIENCY 253260		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
BUB1B	MOSAIC VARIEGATED ANEUPLOIDY SYNDROME 1 257300		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
C12orf57	COLOBOMA, HYPOPLASTIC CORPUS CALLOSUM AND INTELLECTUAL DISABILITY 218340;TEMTAMY SYNDROME		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
C12orf65	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 7 613559		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
C2orf71	RETINITIS PIGMENTOSA 54 613428		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
C5orf42	JOUBERT SYNDROME 614615		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
C8orf37	CONE-ROD DYSTROPHY 16 614500		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CA2	OSTEOPETROSIS AUTOSOMAL RECESSIVE TYPE 3 259730		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CA8	CEREBELLAR ATAXIA MENTAL RETARDATION AND DYSEQUILIBRIUM SYNDROME TYPE 3 613227		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CACNA1A	EPILEPTIC ENCEPHALOPATHY		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CACNA1C	Timothy syndrome, OMIM:601005;Timothy syndrome, MONDO:0010979;CACNA1C-related disorder		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CACNA1D	PRIMARY ALDOSTERONISM, SEIZURES, AND NEUROLOGIC ABNORMALITIES 615474;SINOATRIAL NODE DYSFUNCTION AND DEAFNESS 614896		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
CACNA1E	Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesia		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CACNA1G	AUTOSOMAL RECESSIVE MENTAL RETARDATION CACNA1G-related developmental disorder (monoallelic)		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CAMTA1	CEREBELLAR ATAXIA, NONPROGRESSIVE, WITH MENTAL RETARDATION 614756		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CANT1	Desbuquois dysplasia 1, OMIM:251450		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CASK	MENTAL RETARDATION X-LINKED CASK-RELATED 300749;EG SYNDROME TYPE 4 300422;MRX WITH/WITHOUT NYSTAGMUS 300749		X-LINKED; hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
CASP2	intellectual developmental disorder, autosomal recessive 80, with variant tessencephaly, OMIM:620653		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CBFB	CBFB-related cleidocranial dysplasia		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CBL	NOONAN SYNDROME-LIKE DISORDER WITH OR WITHOUT JUVENILE MYELOMONOCYTIC LEUKEMIA 613563		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CBS	CYSTATHIONINE BETA-SYNTHASE DEFICIENCY 236200		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CC2D1A	MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 3 608443		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CC2D2A	JOUBERT SYNDROME 9 612285;COACH SYNDROME 216360;MECKEL SYNDROME, TYPE 6 612284		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CCBE1	HENNEKAM LYMPHANGIECTASIA-LYMPHEDEMA SYNDROME 235510		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CCDC103	PRIMARY CILIARY DYSKINESIA		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CCDC22	SYNDROMIC X-LINKED INTELLECTUAL DISABILITY		X-LINKED; hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
CCDC39	CILIARY DYSKINESIA, PRIMARY, 14 613807		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CCDC40	CILIARY DYSKINESIA, PRIMARY, 15 613808		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CCDC8	THREE M SYNDROME 3 614205		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CCDC88C	HYDROCEPHALUS, NONSYNDROMIC, AUTOSOMAL RECESSIVE 236600		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CD151	NEPHROPATHY WITH PRETIBIAL EPIDERMOLYSIS BULLOSA AND DEAFNESS 609057		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CDC6	MEIER-GORLIN SYNDROME 5 613805		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CDH1	Blepharo-cheilo-dontic syndrome		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CDH23	USHER SYNDROME TYPE 1D 601067;DEAFNESS AUTOSOMAL RECESSIVE TYPE 12 601386		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CDH3	LEMI SYNDROME 225280;HYPOTRICHOSIS, CONGENITAL, WITH JUVENILE MACULAR DYSTROPHY 601553		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CDK5RAP2	PRIMARY, AUTOSOMAL RECESSIVE MICROCEPHALY 279936		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CDKL5	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE TYPE 2 300672		X-LINKED; hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
CDKN1C	BECKWITH-WIEDEMANN SYNDROME 130650;IMAGe Syndrome		MONOALLELIC; autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)	DDG2P			5.1
CDON	HOLOPROSENCEPHALY 11 614226		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CDT1	MEIER-GORLIN SYNDROME 4 613804		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CENPJ	SECKEL SYNDROME TYPE 4 613676;MICROCEPHALY PRIMARY TYPE 6 608393		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CEP135	PRIMARY MICROCEPHALY AND DISTURBED CENTROSOMAL FUNCTION 614673		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CEP152	MICROCEPHALY PRIMARY TYPE 4 604321;SECKEL SYNDROME TYPE 5 613823		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CEP290	BARDET-BIEDL SYNDROME TYPE 14 209900;LEBER CONGENITAL AMAUROSIS TYPE 10 611755;JOUBERT SYNDROME TYPE 5 610188;SENIOR-LOKEN SYNDROME TYPE 6 610189;MECKEL SYNDROME TYPE 4 611134		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CEP41	JOUBERT SYNDROME 15 614464		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CEP57	MOSAIC VARIEGATED ANEUPLOIDY SYNDROME 2 614114		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CEP63	SECKEL SYNDROME 6 614728		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CEP89L	CEP89L-associated posterior predominant lissencephaly, OMIM:618873		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CFC1	CFC1-RELATED CONOTRUNCAL HEART MALFORMATIONS 319372		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CFL2	NEMALINE MYOPATHY 7 610687		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CHD2	EPILEPTIC ENCEPHALOPATHY		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CHD3	Macrocephaly and impaired speech and language		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CHD7	CHARGE SYNDROME 214800;IDIOPATHIC HYPOGONADOTROPIC HYPOGONADISM 146110;KALLMANN SYNDROME TYPE 5 612370		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CHD8	AUTISM 209850		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CHM	CHOROIDEREMIA 303100		X-LINKED; hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
CHRDL1	X-LINKED MEGALOCORNEA, X-LINKED 309300		X-LINKED; hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
CHRNA1	MULTIPLE PTERYGIUM SYNDROME LETHAL TYPE 253290		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CHRNA3	CHRNA3-related congenital anomalies of the kidney and urinary tract		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CHRNA4	NOCTURNAL FRONTAL LOBE EPILEPSY, TYPE 1 600513		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CHRNB1	CHRNB1-related congenital myasthenia, biallelic, OMIM:616314;CHRNB1-related congenital myasthenia, monoallelic, OMIM:616313		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
CHRNB2	CHRNB2-RELATED NOCTURNAL FRONTAL LOBE EPILEPSY, AUTOSOMAL DOMINANT 605375;NOCTURNAL FRONTAL LOBE EPILEPSY, AUTOSOMAL DOMINANT 117426		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CHRNG	MULTIPLE PTERYGIUM SYNDROME ESCOBAR VARIANT 265000		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CHST14	EHLERS-DANLOS SYNDROME MUSCULOCONTRACTURAL TYPE 601776		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CHST3	SPONDYLOEPIPHYSEAL DYSPLASIA WITH CONGENITAL JOINT DISLOCATIONS 143095		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CHSY1	TEMTAMY PREAXIAL BRACHYDACTYLY SYNDROME 605282		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CHUK	COCOON SYNDROME 613630		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CIB2	NONSYNDROMIC DEAFNESS DFNB48 609439;USHER SYNDROME TYPE 1J 614869		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CIC	CAPICUA, DROSOPHILA, HOMOLOG OF 612082		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CISD2	WOLFRAM SYNDROME TYPE 2 604928		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CLCN7	OLCN7-RELATED OSTEOPETROSIS 611490		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CLCNKB	BARTTER SYNDROME TYPE 4B 613090		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CLDN19	HYPOMAGNESEMIA 5, RENAL, WITH OCULAR INVOLVEMENT 248190		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CLMP	CONGENITAL SHORT BOWEL SYNDROME 615237		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CLN3	NEURONAL CEROID LIPOFUSCINOSIS TYPE 3 204200		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CLN5	Ceroid lipofuscinosis, neuronal, 5 OMIM:256731;neuronal ceroid lipofuscinosis 5 MONDO:0009745		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CLN6	CEROID LIPOFUSCINOSIS, NEURONAL, 6, OMIM:601780;CEROID LIPOFUSCINOSIS, NEURONAL, KUFS TYPE, ADULT ONSET, OMIM:204300		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CLN8	NEURONAL CEROID LIPOFUSCINOSIS TYPE 8 NORTHERN EPILEPSY VARIANT 610003;NEURONAL CEROID LIPOFUSCINOSIS TYPE 8 600143		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CNKSR2	INTELLECTUAL DISABILITY WITH EPILEPSY		X-LINKED; hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
CNNM2	CNNM2-related neurodevelopmental disorder with hypomagnesemia autosomal recessive form		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
CNOT3	CNOT3 syndrome		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CNPY3	EARLY ONSET EPILEPTIC ENCEPHALOPATHY		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CNTNAP2	CORTICAL DYSPLASIA-FOCAL EPILEPSY SYNDROME 610042		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COG1	COG1-CDG 300197		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COG4	COG4-CDG 613493;Saul-Wilson syndrome 618150		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
COG5	COG5-CDG 613494		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COG7	COG7-CDG 300171		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COG8	COG8-CDG 300264		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COL10A1	SCHMID TYPE METAPHYSEAL CHONDRODYSPLASIA 156500		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
COL11A1	STICKLER SYNDROME, TYPE II 604841;FIBROCHONDROGENESIS 228520		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
COL11A2	WEISSENBACHER-ZWEYMUELLER SYNDROME 184840;AUTOSOMAL RECESSIVE OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA 215150;DEAFNESS AUTOSOMAL DOMINANT TYPE 13 601868;STICKLER SYNDROME TYPE 3 184840;DEAFNESS AUTOSOMAL RECESSIVE TYPE 53 609706		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
COL18A1	Knobloch syndrome, type 1, OMIM:267750		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COL1A1	CAFFEY DISEASE 114000;OSTEOGENESIS IMPERFECTA TYPE IA 166210;OSTEOGENESIS IMPERFECTA TYPE II 259420;COL1A1/2-RELATED OSTEOGENESIS IMPERFECTA 166210;EHLERS-DANLOS SYNDROME, CLASSIC TYPE, COL1A1-RELATED 130060;OSTEOGENESIS IMPERFECTA TYPE I 166200;EHLERS-DANLOS SYNDROME TYPE VIIA 319158		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
COL25A1	CONGENITAL FATIGUING EXTRAOCULAR MUSCLES, CONGENITAL, 5 616219		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COL2A1	ACHONDROGENESIS TYPE 2 200610;PLATYSPONDYLIC LETHAL SKELETAL DYSPLASIA TORRANCE TYPE 151210;SPONDYLOEPIPHYSEAL DYSPLASIA 271700;PRIMARY AVASCULAR NECROSIS OF FEMORAL HEAD 608805;SPONDYLOEPIMETAPHYSEAL DYSPLASIA STRUDWICK TYPE 184250;SPONDYLOEPIPHYSEAL DYSPLASIA,CONGENITA 183900;RHEU		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
COL4A1	PORENCEPHALY 1 175780		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
COL4A2	PORENCEPHALY 2 614483		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
COL4A3	ALPORT SYNDROME AUTOSOMAL RECESSIVE, OMIM:203780;ALPORT SYNDROME AUTOSOMAL DOMINANT, OMIM:104200		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
COL4A4	ALPORT SYNDROME AUTOSOMAL RECESSIVE 203780		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COL4A5	COL4A5 associated myopathy		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
COL6A2	COL6A2-related Ullrich congenital muscular dystrophy (monoallelic), OMIM:254090;COL6A2-related Ullrich congenital muscular dystrophy (biallelic), OMIM:254090		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
COL6A3	DYSTONIA 27 616641;ULLRICH CONGENITAL MUSCULAR DYSTROPHY 1 254090		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COL9A1	STICKLER SYNDROME TYPE 6 614134;MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 6 614135		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
COL9A2	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 2 600204;STICKLER SYNDROME, TYPE V 614284		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
COL9A3	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 3 600969;Stickler syndrome		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
COLEC11	3MC SYNDROME 2 265050		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COQ2	COENZYME Q10 DEFICIENCY 607426		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COQ4	COENZYME Q10 DEFICIENCY, PRIMARY, 7 616276		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COQ9	COENZYME Q10 DEFICIENCY 607426		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COX10	LEIGH SYNDROME 256000;MITOCHONDRIAL COMPLEX IV DEFICIENCY 220110		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COX15	LEIGH SYNDROME 256000;MITOCHONDRIAL COMPLEX IV DEFICIENCY 220110		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
COX6B1	MITOCHONDRIAL COMPLEX IV DEFICIENCY 220110		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CPS1	CARBAMOYL PHOSPHATE SYNTHETASE 1 DEFICIENCY 237300		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CRB1	RETINITIS PIGMENTOSA 12, AUTOSOMAL RECESSIVE 600105;LEBER CONGENITAL AMAUROSIS 8 613835		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CREBBP	RUBINSTEIN-TAYBI SYNDROME TYPE 1 180849;CREBBP intellectual disability without typical RTS features		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CRELD1	HETEROTAXY SYNDROME 207574		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CRX	CRX-RELATED LEBER CONGENITAL AMAUROSIS;LEBER CONGENITAL AMAUROSIS 7 613829		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CRYAA	CATARACT, NUCLEAR 123580;CATARACT, AUTOSOMAL RECESSIVE CONGENITAL 1 604219		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
CRYBA1	CATARACT CONGENITAL ZONULAR WITH SUTURAL OPACITIES 600881		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CRYBB1	CATARACT 17, MULTIPLE TYPES, MONOALLELIC;CATARACT 17, MULTIPLE TYPES;CATARACT 17, MULTIPLE TYPES, BIALLELIC		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
CRYBB2	CATARACT, CONGENITAL, CERULEAN TYPE, 2 601547;CATARACT, COPPOCK-LIKE 604307		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CRYBB3	CATARACT, CONGENITAL, NUCLEAR, AUTOSOMAL RECESSIVE 2 609741		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CRYGC	CATARACT AUTOSOMAL DOMINANT 604219;CATARACT COPPOCK-LIKE 604307		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CSF1R	BRAIN ABNORMALITIES, NEURODEGENERATION, AND DYSOSTEOSCLEROSIS 618476		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CST3	EXFOLIATIVE ICHTHYOSIS; AUTOSOMAL RECESSIVE; ICHTHYOSIS BULLOSA OF SIEMENS-LIKE 607936		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CSTB	UNVERRICHT-LUNDBORG DISEASE 254800		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CTC1	CEREBRORETINAL MICROANGIOPATHY WITH CALCIFICATIONS AND CYSTS 612199		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CTDP1	CONGENITAL CATARACTS FACIAL DYSMORPHISM AND NEUROPATHY SYNDROME 604168		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CTNNB1	MENTAL RETARDATION, AUTOSOMAL DOMINANT 19 615075		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CTNND1	Blepharo-cheilo-dontic syndrome		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CTNS	CYSTINOSIS ADULT NON-NEPHROPATHIC TYPE 219750;CYSTINOSIS LATE-ONSET JUVENILE OR ADOLESCENT NEPHROPATHIC TYPE 219900;CYSTINOSIS NEPHROPATHIC TYPE 219800		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CTSA	GALACTOSIALIDOSIS 256540		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
CTSD	NEURONAL CEROID LIPOFUSCINOSIS TYPE 10 610127		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1

CTSK	PYCNODYSOSTOSIS 265800		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
CUL3	CUL3-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CUL4B	MENTAL RETARDATION SYNDROMIC X-LINKED CABEZAS TYPE 300354		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
CUL7	3-M SYNDROME 1 273750		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
CYB5R3	METHEMOGLOBINEMIA DUE TO DEFICIENCY OF METHEMOGLOBIN REDUCTASE 319395		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
CYP1B1	PRIMARY CONGENITAL GLAUCOMA TYPE 3A 231300		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
CYP27A1	Cerebrotendinous xanthomatosis		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DAG1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C7 613818		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DARS2	LEUKOENCEPHALOPATHY WITH BRAINSTEM AND SPINAL CORD INVOLVEMENT AND LACTATE ELEVATION 611105		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DBT	MAPLE SYRUP URINE DISEASEQ 248600		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DCC	Midline-bridging neuronal commissure disruption, horizontal gaze palsy, scoliosis, and intellectual disability		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DCDC2	RENAL-HEPATIC CILIOPATHY 616217		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DCX	SUBCORTICAL BAND HETEROTOPIA X-LINKED 300067;LISSENCEPHALY X-LINKED TYPE 1 300067		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
DDB2	XERODERMA PIGMENTOSUM, GROUP E, DDB-NEGATIVE SUBTYPE 278740		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DDC	AROMATIC L-AMINO-ACID DECARBOXYLASE DEFICIENCY 608643		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DDOST	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IR 614507		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DDR2	SPONDYLOEPIMETAPHYSEAL DYSPLASIA SHORT LIMB-HAND TYPE 271665		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DDX11	WARSAW BREAKAGE SYNDROME 613398		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DEAF1	Autism, intellectual disability, basal ganglia dysfunction and epilepsy;MENTAL RETARDATION, AUTOSOMAL DOMINANT 24 615828		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
DHCR24	DESMOSTEROLOSIS 238860		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DHCR7	SMITH-LEMLI-OPITZ SYNDROME 270400		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DHDDS	Epilepsy and intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
DHFR	MEGALOBLASTIC ANEMIA DUE TO DIHYDROFOLATE REDUCTASE DEFICIENCY 613839		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DHODH	POSTAXIAL ACROFACIAL DYSOSTOSIS 263750		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DIS3L2	PERLMAN SYNDROME 267000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DKC1	DYSKERATOSIS CONGENITA, X-LINKED, 305000;DKC1-RELATED DYSKERATOSIS CONGENITA 314912		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
DLAT	PYRUVATE DEHYDROGENASE E2 DEFICIENCY 245348		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DLD	DIHYDROLIPOAMIDE DEHYDROGENASE (E3) DEFICIENCY 248600;LEIGH SYNDROME 256000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DLG3	MENTAL RETARDATION X-LINKED TYPE 90 300189		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
DLL3	SPONDYLOCOSTAL DYSOSTOSIS TYPE 1 277300		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DMD	DUCHENNE MUSCULAR DYSTROPHY 310200;BECKER MUSCULAR DYSTROPHY 300376;CARDIOMYOPATHY DILATED X-LINKED TYPE 3B 302045		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
DMP1	HYPOPHOSPHATEMIC RICKETS, AR 241520		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DNAAF3	PRIMARY CILIARY DYSKINEASIA 606763		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DNAH5	CILIARY DYSKINESIA, PRIMARY, 3		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DNAH9	Motile Cilia Defects and Situs Inversus		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DNM1	EPILEPTIC ENCEPHALOPATHY		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
DNM1L	DNM1L-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
DNMT3A	Tatton-Brown Rahman syndrome (OVERGROWTH SYNDROME WITH INTELLECTUAL DISABILITY), 615879;Microcephalic primordial dwarfism		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
DNMT3B	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME 1 242860		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DOCK6	ADAMS-OLIVER SYNDROME 2 614219		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DOCK8	HYPERIMMUNOGLOBULIN E RECURRENT INFECTION SYNDROME AUTOSOMAL RECESSIVE 243700		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DOLK	CONGENITAL DISORDERS OF GLYCOSYLATION 612379		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DPAGT1	MYASTHENIC SYNDROME, CONGENITAL, WITH TUBULAR AGGREGATES 2 614750;DPAGT1-CDG 300129		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DPM1	CONGENITAL DISORDERS OF GLYCOSYLATION 612379		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DPM3	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1O 612937		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DSG1	SEVERE DERMATITIS, MULTIPLE ALLERGIES AND METABOLIC WASTING, 615508		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DSP	DSP-related developmental disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
DSPP	DENTINOGENESIS IMPERFECTA, SHIELDS TYPE II 125490;DEAFNESS AUTOSOMAL DOMINANT TYPE 39 WITH DENTINOGENESIS IMPERFECTA 1 605594		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
DYM	DYGGVE-MELCHIOR-CLAUSEN SYNDROME 223800;SMITH-MCCORT DYSPLASIA 607326		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DYNC1H1	SPINAL MUSCULAR ATROPHY, LOWER EXTREMITY-PREDOMINANT, AD 158600;SEVERE ID WITH NEURONAL MIGRATION DISORDER 600112		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
DYNC2H1	ASPHYXIATING THORACIC DYSTROPHY TYPE 3 613091;SHORT RIB-POLYDACTYLY SYNDROME TYPE 3 263510		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DYRK1A	MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 7 614104		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
EBP	CHONDRODYSPLASIA PUNCTATA 2, X-LINKED 302960		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
ECM1	LIPOID PROTEINOSIS OF URBACH AND WIETHE, OMIM:247100		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EDA	TOOTH AGENESIS SELECTIVE X-LINKED TYPE 1 313500;ECTODERMAL DYSPLASIA TYPE 1 305100		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
EDAR	ECTODERMAL DYSPLASIA 10A, HYPOHIDROTIC/HAIR/NAIL TYPE, AUTOSOMAL DOMINANT;Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
EDN1	AURICULOCONDYLAR SYNDROME 602483		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
EDNRA	MANDIBULOFACIAL DYSOSTOSIS WITH ALOPECIA 616367		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
EDNRB	ABCD SYNDROME, OMIM:600501		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
EFEMP2	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE IB, OMIM:614437		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EFNB1	CRANIOFRONTONASAL SYNDROME, OMIM:304110		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
EFTUD2	MANDIBULOFACIAL DYSOSTOSIS WITH MICROCEPHALY 610536		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
EGR2	NEUROPATHY, CONGENITAL HYPOMYELINATING, 1 605253		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EHMT1	Kleefstra syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
EIF2AK3	WOLCOTT-RALLISON SYNDROME 226980		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EIF2B4	EIF2B4-RELATED LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, OMIM:603896		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EIF2B5	EIF2B5-RELATED LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, OMIM:603896		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ELAC2	INFANTILE HYPERTROPHIC CARDIOMYOPATHY, LACTIC ACIDOSIS, AND ISOLATED COMPLEX I DEFICIENCY 252010		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ELN	ELN-RELATED CUTIS LAXA 314088;SUPRAVALVAR AORTIC STENOSIS 185500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ELOVL4	ICHTHYOSIS, SPASTIC QUADRIPLEGIA, AND MENTAL RETARDATION 614457		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EMG1	Bowen-Conradi syndrome		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ENPP1	HYPOPHOSPHATEMIC RICKETS, AUTOSOMAL RECESSIVE, 2 613312;ARTERIAL CALCIFICATION, GENERALIZED, OF INFANCY, 1 208000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EP300	RUBINSTEIN-TAYBI SYNDROME TYPE 2 613684		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
EPCAM	DIARRHEA 5, WITH TUFTING ENTEROPATHY, CONGENITAL, OMIM:613217		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ERBB3	Hirschprung disease with intestinal pseudo-obstruction;LETHAL CONGENITAL CONTRACTURE SYNDROME TYPE 2 607598		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ERCC1	FANCONI ANEMIA 229154;CEREBROOCULOFACIOSKELETAL SYNDROME 4 610758		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ERCC2	XERODERMA PIGMENTOSUM COMPLEMENTATION GROUP D 278730;CEREBRO-OCULO-FACIO-SKELETAL SYNDROME TYPE 2 610756;TRICHOTHIODYSTROPHY PHOTOSENSITIVE 601675		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ERCC3	XERODERMA PIGMENTOSUM COMPLEMENTATION GROUP B 610651;TRICHOTHIODYSTROPHY PHOTOSENSITIVE 601675		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ERCC4	PRIMORDIAL DWARFISM 615272;XFE PROGEROID SYNDROME 610965;XERODERMA PIGMENTOSUM, GROUP F 278760;FANCONI ANEMIA, COMPLEMENTATION GROUP Q 615272		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ERCC5	XERODERMA PIGMENTOSUM COMPLEMENTATION GROUP G 278780		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ERCC6	DE SANCTIS-CACCHIONE SYNDROME 278800;UV-SENSITIVE SYNDROME 609413;CEREBRO-OCULO-FACIO-SKELETAL SYNDROME TYPE 1 214150;COCKAYNE SYNDROME TYPE B 133540		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ERCC8	COCKAYNE SYNDROME TYPE A 216400		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ESAM	ESAM-related neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ESCO2	SC PHOCOMELIA SYNDROME 269000;ROBERTS SYNDROME 268300		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ETFA	Glutaric acidemia IIA, OMIM:231680;multiple acyl-CoA dehydrogenase deficiency, MONDO:0009282		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ETFB	Glutaric acidemia IIB, OMIM:231680;multiple acyl-CoA dehydrogenase deficiency, MONDO:0009282		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ETFDH	GLUTARIC ACIDURIA TYPE 2C 231680		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ETHE1	ETHYLMALONIC ENCEPHALOPATHY 602473		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EVC	ELLIS-VAN CREVELD SYNDROME 225500;ACROFACIAL DYSOSTOSIS WEYERS TYPE 193530		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EVC2	ACROFACIAL DYSOSTOSIS WEYERS TYPE 193530;ELLIS-VAN CREVELD SYNDROME 225500		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EXOSC3	PONTOCEREBELLAR HYPOPLASIA TYPE 1 607596		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EXT1	HEREDITARY MULTIPLE EXOSTOSES TYPE 1 133700;TRICHO-RHINO-PHALANGEAL SYNDROME TYPE 2 150230		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
EXT2	EXOSTOSES, MULTIPLE, TYPE 2 133701		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
EXTL3	Neuro immuno skeletal Dysplasia Syndrome		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EYA1	BRANCHIOOTORENAL SYNDROME TYPE 1, OMIM:113650		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
EZH2	WEAVER SYNDROME 2 614421		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FAH	TYROSINEMIA TYPE 1 276700		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FAM126A	LEUKODYSTROPHY HYPOMYELINATING TYPE 5 610532		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FAM161A	RETINITIS PIGMENTOSA 28 606068		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FAM20A	AMELOGENESIS IMPERFECTA AND GINGIVAL FIBROMATOSIS SYNDROME 614253		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FAM20C	RAINE SYNDROME 259775		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FAM58A	STAR SYNDROME 300707		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
FANCA	FANCONI ANEMIA, COMPLEMENTATION GROUP A 227650		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FANCB	FANCB-RELATED FANCONI ANEMIA 229139		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
FANCC	FANCONI ANEMIA, COMPLEMENTATION GROUP C 227645		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FANCD2	FANCONI ANEMIA, COMPLEMENTATION GROUP D2 227646		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FANCE	FANCONI ANEMIA, COMPLEMENTATION GROUP E 600901		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FANCF	FANCONI ANEMIA, COMPLEMENTATION GROUP F 603467		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FANCG	FANCONI ANEMIA, COMPLEMENTATION GROUP G 614082		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FANCI	FANCI-RELATED FANCONI ANEMIA 234476;FANCONI ANEMIA 229154		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FANCL	FANCL-RELATED FANCONI ANEMIA 234480;FANCONI ANEMIA 229154		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FANCM	FANCM-RELATED FANCONI ANEMIA 287713;FANCONI ANEMIA 229154		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FARS2	Neurometabolic disorder due to FARS2 deficiency		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FBN1	WEILL-MARCHESANI SYNDROME AUTOSOMAL DOMINANT, OMIM:608328;MARFAN SYNDROME, OMIM:154700;SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME, OMIM:182212;Marfan Syndrome, biallelic, OMIM:154700		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
FBN2	Contractural arachnodactyly, congenital OMIM:121050;congenital contractural arachnodactyly MONDO:0007363		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FBP1	FRUCTOSE 1,6 BISPHOSPHATASE DEFICIENCY 229700		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FBXW11	SYNDROMIC INTELLECTUAL DISABILITY 612100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FBXW7	FBXW7-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FGD1	AARSKOG-SCOTT SYNDROME 305400		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
FGF10	LADD SYNDROME 149730		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FGF14	FGF14-related episodic ataxia		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FGF3	DEAFNESS WITH LABYRINTHINE APLASIA, MICROTIA AND MICRODONTIA 610706		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FGF9	MULTIPLE SYNOSTOSES SYNDROME TYPE 3 612961		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FGFR1	Hypogonadotropic hypogonadism 2 with or without anosmia, OMIM:147950;Encephalocraniocutaneous lipomatosis, OMIM:613001;PFEIFFER SYNDROME, OMIM:101600;OSTEOGLOPHONIC DYSPLASIA, OMIM:166250;Hartsfield syndrome, OMIM:615465		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FGFR2	BEARE-STEVENSON CUTIS GYRATA SYNDROME 123790;ANTLEY-BIXLER SYNDROME 207410;FAMILIAL SCAPHOCEPHALY SYNDROME 609579;JACKSON-WEISS SYNDROME 123150;APERT SYNDROME 101200;CROUZON SYNDROME 123500;LACRIMO-AURICULO-DENTO-DIGITAL SYNDROME 149730;ACROCEPHALOSYNDACTYLY TYPE V 101600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FGFR3	THANATOPHORIC DYSPLASIA TYPE 2 187601;THANATOPHORIC DYSPLASIA TYPE 1 187600;MUENKE SYNDROME 602849;ACHONDROPLASIA 100800;CROUZON SYNDROME WITH ACANTHOSIS NIGRICANS 612247;CAMPTODACTYLY TALL STATURE AND HEARING LOSS SYNDROME 610474;LACRIMO-AURICULO-DENTO-DIGITAL SYNDROME 149730;HYPOCH		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FH	FUMARASE DEFICIENCY 606812		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FHL1	EMERY-DREIFUSS MUSCULAR DYSTROPHY 6, X-LINKED 300696		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
FIG4	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4J 611228;CLEIDOCRANIAL DYSPLASIA WITH MICROGNATHIA, ABSENT THUMBS, AND DISTAL APHALANGIA YUNIS-VARON SYNDROME 216340		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FKBP10	BRUCK SYNDROME TYPE 1		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FKBP14	EHLERS-DANLOS SYNDROME WITH PROGRESSIVE KYPHOSCOLIOSIS, MYOPATHY, AND HEARING LOSS		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FKRP	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH OR WITHOUT MENTAL RETARDATION TYPE B5 606612;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH BRAIN AND EYE ANOMALIES TYPE A5 613153;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C5 607155		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FKTN	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH BRAIN AND EYE ANOMALIES TYPE A4 253800;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C4 611588;CARDIOMYOPATHY DILATED TYPE 1X 611615;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITHOUT MENTAL RETARDATION TYPE B4		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FLNA	PERIVENTRICULAR NODULAR HETEROTOPIA 1 300049;FRONTOMETAPHYSEAL DYSPLASIA 305620;FG SYNDROME TYPE 2 300321;X-LINKED CONGENITAL IDIOPATHIC INTESTINAL PSEUDOOBSTRUCTION 300048;MELNICK-NEEDLES SYNDROME 309350;Childhood Interstitial Lung Disease;EPILEPTIC ENCEPHALOPATHY;OTOPALATODIGITAL SYNDROME TYPI		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
FLNB	AUTOSOMAL DOMINANT LARSEN SYNDROME 150250;ATELOSTEOGENESIS TYPE 3 108721;ATELOSTEOGENESIS TYPE 1 108720;BOOMERANG DYSPLASIA 112310;SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME 272460		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
FLT4	MILROY DISEASE 153100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FLVCR1	ATAXIA, POSTERIOR COLUMN, WITH RETINITIS PIGMENTOSA 609033		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FLVCR2	PROLIFERATIVE VASCULOPATHY AND HYDRAENCEPHALY-HYDROCEPHALY SYNDROME 225790		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FMN2	NONSYNDROMIC AUTOSOMAL-RECESSIVE INTELLECTUAL DISABILITY		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FMR1	FRAGILE X TREMOR/ATAXIA SYNDROME, OMIM:300623;PREMATURE OVARIAN FAILURE SYNDROME TYPE 1, OMIM:311360;FRAGILE X SYNDROME, OMIM:300624		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
FN1	Spondylometaphyseal Dysplasia with Corner Fractures 184255		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FOLR1	NEURODEGENERATION DUE TO CEREBRAL FOLATE TRANSPORT DEFICIENCY 613068		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FOXC1	IRIDOGONIODYSGENESIS ANOMALY 601631;AXENFELD-RIEGER SYNDROME TYPE 3 602482;PETERS ANOMALY 604229		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FOXC2	LYMPHEDEMA-DISTICHIASIS SYNDROME 153400;HEREDITARY LYMPHEDEMA II 241432		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FOXE1	BAMFORTH-LAZARUS SYNDROME 241850		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FOXE3	CONGENITAL PRIMARY APHAKIA 610256;ANTERIOR SEGMENT MESENCHYMAL DYSGENESIS 107250		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
FOXF1	ALVEOLAR CAPILLARY DYSPLASIA WITH MISALIGNMENT OF PULMONARY VEINS 265380		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FOXG1	CONGENITAL VARIANT OF RETT SYNDROME 613454		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FOXL2	BLEPHAROPHIMOSIS, PTOSIS, AND EPICANTHUS INVERSUS SYNDROME 110100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FOXN1	ALOPECIA AND T-CELL IMMUNODEFICIENCY 601705		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FOXP1	MENTAL RETARDATION WITH LANGUAGE IMPAIRMENT AND AUTISTIC FEATURES 613670		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FOXP2	SPEECH-LANGUAGE DISORDER 1 602081		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FOXP3	IPEX SYNDROME 304790		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
FOXRED1	MITOCHONDRIAL COMPLEX I DEFICIENCY 252010		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FRA10AC1	FRA10AC1-related neurodevelopmental disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FRAS1	FRASER SYNDROME 219000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FREM1	MANITOBA OCULOTRICHOANAL SYNDROME 248450		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FREM2	FRASER SYNDROME 219000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FRMPD4	Intellectual Disability		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
FTCD	GLUTAMATE FORMIMINOTRANSFERASE DEFICIENCY 229100		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FTL	HEREDITARY HYPERFERRITINEMIA-CATARACT SYNDROME 600886		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FTSJ1	MENTAL RETARDATION X-LINKED TYPE 44 309549		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
FUCA1	FUCOSIDOSIS 230000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FUT8	Congenital Disorder of Glycosylation with Defective Fucosylation		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FYCO1	CATARACT, AUTOSOMAL RECESSIVE CONGENITAL 2 610019		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FZD6	NAIL DISORDER NON-SYNDROMIC CONGENITAL TYPE 10 614157		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GAA	GLYCOGEN STORAGE DISEASE TYPE II 232300		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GABBR1	GABBR1-associated neurodevelopmental disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GABRA1	JUVENILE MYOCLONIC EPILEPSY 611136;EPILEPTIC ENCEPHALOPATHY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GABRB3	EPILEPTIC ENCEPHALOPATHIES;CHILDHOOD ABSENCE EPILEPSY TYPE 5 612269		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GABRG2	EPILEPSY, GENERALIZED, WITH FEBRILE SEIZURES PLUS, TYPE 3 611277;GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 3 611277		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GALC	KRABBE DISEASE 245200		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GALE	EPIMERASE-DEFICIENCY GALACTOSEMIA 230350		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GALK1	GALACTOSEMIA II 230200		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GALNS	MUCOPOLYSACCHARIDOSIS TYPE 4A 253000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GALT	GALACTOSEMIA 230400		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1

GAMT	GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY 612736		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GAN	Giant axonal neuropathy 1		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GAS2L2	Impaired Cilia Orientation and Mucociliary Clearance		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GATA2	EMBERGER SYNDROME 614038		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GATA3	HYPOPARATHYROIDISM, SENSORINEURAL DEAFNESS, AND RENAL DISEASE, OMIM:146255		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GATA4	ATRIAL SEPTAL DEFECT TYPE 2 607941		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GATA6	PANCREATIC AGENESIS, DIAPHRAGMATIC HERNIA AND CONGENITAL HEART DEFECTS 600001;ATRIOVENTRICULAR SEPTAL DEFECT 5 614474;ATRIAL SEPTAL DEFECT 9 614475		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GATM	ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY 612718		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GBA	GAUCHER DISEASE TYPE 3 231000;GAUCHER DISEASE PERINATAL LETHAL 608013;GAUCHER DISEASE 230800;GAUCHER DISEASE TYPE 1 230800;GAUCHER DISEASE TYPE 2 230900;GAUCHER DISEASE TYPE 3C 231005		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GBE1	GBE1-associated Glycogen storage disease IV, OMIM:232500		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GCDH	GLUTARICACIDEMIA TYPE 1 231670		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GCH1	DYSTONIA TYPE 5 128230;GTP CYCLOHYDROLASE 1 DEFICIENCY 233910		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
GCSH	GLYCINE ENCEPHALOPATHY 605899		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GDF1	CONOTRUNCAL HEART MALFORMATIONS 217095;TETRALOGY OF FALLOT 187500;TRANSPOSITION OF THE GREAT ARTERIES DEXTRO-LOOPED TYPE 3 613854		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GDF5	BRACHYDACTYLY TYPE C 113100;ACROMESOMELIC CHONDRODYSPLASIA GREBE TYPE 200700;ACROMESOMELIC CHONDRODYSPLASIA HUNTER-THOMPSON TYPE 201250;DU PAN SYNDROME 228900;SYMPHALANGISM PROXIMAL SYNDROME 185800;BRACHYDACTYLY TYPE A1 112500;BRACHYDACTYLY TYPE A2 112600;MULTIPLE SYNOSTOSES SYN		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
GDF6	KLIPPEL-FEIL SYNDROME TYPE 1 118100;MICROPHTHALMIA ISOLATED TYPE 4 613094		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GEMIN4	NEURODEVELOPMENTAL DISORDER WITH MICROCEPHALY, CATARACTS, AND RENAL ABNORMALITIES, OMIM:617913		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GFAP	ALEXANDER DISEASE 203450		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GFM1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 609060		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GHR	PITUITARY DWARFISM II 262500		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GJA1	HALLERMANN-STREIFF SYNDROME, OMIM:234100;SYNDACTYLY TYPE 3, OMIM:186100;AUTOSOMAL DOMINANT OCULODENTODIGITAL DYSPLASIA, OMIM:164200;HYPOPLASTIC LEFT HEART SYNDROME, OMIM:241550;AUTOSOMAL RECESSIVE OCULODENTODIGITAL DYSPLASIA, OMIM:257850		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
GJA3	CATARACT ZONULAR PULVERULENT CATARACT TYPE 3 601885		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GJA8	CATARACT ZONULAR PULVERULENT TYPE 1 116200;CATARACT-MICROCORNEA SYNDROME 116150		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GJB3	DEAFNESS AUTOSOMAL DOMINANT TYPE 2B, OMIM:612644;ERYTHROKERATODERMIA VARIABILIS ET PROGRESSIVA, OMIM:133200;DEAFNESS, AUTOSOMAL RECESSIVE		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
GJC2	SPASTIC PARAPLEGIA, 44 613206;LEUKODYSTROPHY, HYPOMYELINATING, 2 608804;LYMPHEDEMA, HEREDITARY, IC 613480		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
GK	GLYCEROL KINASE DEFICIENCY 307030		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
GLB1	GM1-GANGLIOSIDOSIS TYPE 1 230500;MUCOPOLYSACCHARIDOSIS TYPE 4B 253010;GM1-GANGLIOSIDOSIS TYPE 2 230600;GM1-GANGLIOSIDOSIS TYPE 3 230650		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GLDC	GLDC-RELATED GLYCINE ENCEPHALOPATHY 605899		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GLE1	ARTHROGRYPOSIS, LETHAL, WITH ANTERIOR HORN CELL DISEASE 611890		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GLI2	GLI2-RELATED HOLOPROSENCEPHALY 261768		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GLI3	PREAXIAL POLYDACTYLY TYPE IV 269157;GREIG CEPHALOPOLYSYNDACTYLY SYNDROME 175700;POSTAXIAL POLYDACTYLY TYPE A 174200;PALLISTER-HALL SYNDROME 146510		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GLIS2	NEPHRONOPHTHISIS 7 611498		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GLIS3	DIABETES MELLITUS NEONATAL WITH CONGENITAL HYPOTHYROIDISM 610199		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GLMN	GLOMUVENOUS MALFORMATIONS 138000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GLRA1	GLRA1-related hyperexplexia, biallelic, OMIM:149400;GLRA1-related hyperexplexia, monoallelic, OMIM:149400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
GLRB	GLRB-related hyperexplexia, biallelic, OMIM:614619;GLRB-related hyperexplexia, monoallelic		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GLUD1	HYPERINSULINISM-HYPERAMMONEMIA SYNDROME 606762		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GLUL	CONGENITAL SYSTEMIC GLUTAMINE DEFICIENCY 610015		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GM2A	GM2-GANGLIOSIDOSIS TYPE AB 272750		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GNA14	Congenital vascular tumours		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GNAQ	Congenital Hemangioma		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GNAS	ALBRIGHT HEREDITARY OSTEODYSTROPHY, OMIM:103580;MCCUNE-ALBRIGHT SYNDROME, OMIM:174800;PSEUDOHYPOPARATHYROIDISM TYPE 1B, OMIM:603233		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GNB5	Sinus Bradycardia and Cognitive Disability		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GNPAT	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA TYPE 2 222765		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GNPTAB	MUCOLIPIDOSIS TYPE II 252500;MUCOLIPIDOSIS TYPE III COMPLEMENTATION GROUP A 252600		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GNPTG	MUCOLIPIDOSIS TYPE III COMPLEMENTATION GROUP C 252605		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GNS	MUCOPOLYSACCHARIDOSIS TYPE 3D 252940		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GORAB	Geroderma osteodysplasticum		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GPC3	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 1 312870		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
GPC4	KEIPERT SYNDROME 301026		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
GPC6	OMODYSPLASIA TYPE 1 (OMOD1) [258315		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GPSM2	CHUDLEY-MCCULLOUGH SYNDROME		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GPX4	SPONDYLOMETAPHYSEAL DYSPLASIA, SEDAGHATIAN TYPE 250220		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GRHL2	ECTODERMAL DYSPLASIA/SHORT STATURE SYNDROME 616029		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GRIA3	MENTAL RETARDATION X-LINKED TYPE 94 300699		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
GRIK2	MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 6 611092		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant OMIM:614254;intellectual disability, autosomal dominant 8 MONDO:0013655		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
GRIN2A	LANDAU-KLEFFNER SYNDROME 245570;EPILEPSY WITH NEURODEVELOPMENTAL DEFECTS 613971		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GRIN2B	EPILEPTIC ENCEPHALOPATHY;AUTISM 209850;MENTAL RETARDATION, AUTOSOMAL DOMINANT 6 613970		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GRM1	CONGENITAL CEREBELLAR ATAXIA 614831		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GRM6	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1B 257270		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GRM7	GRM7-related neurodevelopmental disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GSPT2	XL INTELLECTUAL DISABILITY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
GTF2H5	TRICHOTHIODYSTROPHY PHOTOSENSITIVE 601675		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GUCY2C	MECONIUM ILEUS;FAMILIAL DIARRHEA DIARRHEA 6 614616		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
GUSB	MUCOPOLYSACCHARIDOSIS TYPE 7 253220		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HACE1	HACE1 related disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HADH	3-HYDROXYACYL-COENZYME A DEHYDROGENASE DEFICIENCY 231530		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HADHA	LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY 609016		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HAX1	NEUTROPENIA, SEVERE CONGENITAL 3, AUTOSOMAL RECESSIVE 610738		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HCCS	MICROPHTHALMIA SYNDROMIC TYPE 7 309801		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
HCN1	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 24 615871		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
HDAC4	BRACHYDACTYLY-MENTAL RETARDATION SYNDROME 600430		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
HDAC8	CORNELIA DE LANGE-LIKE SYNDROME HDAC8 XLR;Cornelia de Lange Syndrome HDAC8 X-linked dominant		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
HERC2	HERC2-related neurodevelopmental disorder, OMIM:615516		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HESX1	SEPTOOPTIC DYSPLASIA 256657;HESX1-RELATED COMBINED PITUITARY HORMONE DEFICIENCY 319358		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
HEXA	GM2-GANGLIOSIDOSIS TYPE 1 272800		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HEXB	GM2-GANGLIOSIDOSIS TYPE 2 268800		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HGSNAT	MUCOPOLYSACCHARIDOSIS TYPE 3C 252930		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HIBCH	HIBCH DEFICIENCY 250620		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HK1	HK1-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
HLCS	HOLOCARBOXYLASE SYNTHETASE DEFICIENCY 609018		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HMGCL	3-HYDROXY-3-METHYLGLUTARYL-COENZYME A LYASE DEFICIENCY 246450		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HMGCR	HMGCR-related limb-girdle muscular dystrophy		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HMGCS2	3-HYDROXY-3-METHYLGLUTARYL-COA SYNTHASE 2 DEFICIENCY 605911		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HMX1	OCULOAURICULAR SYNDROME 612109		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HNF1B	RENAL CYSTS AND DIABETES SYNDROME 137920		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
HNF4A	HNF4A-RELATED MATURITY-ONSET DIABETES OF THE YOUNG TYPE 1 125850;ATYPICAL DOMINANT FANCONI SYNDROME WITH MODY 315353		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
HNRNPU	EPILEPTIC ENCEPHALOPATHY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
HOXA1	BOSLEY-SALIH-ALORAINY SYNDROME 601536;ATHABASKAN BRAINSTEM DYSGENESIS SYNDROME 601536		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HOXA13	HAND-FOOT-GENITAL SYNDROME 140000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
HOXD13	BRACHYDACTYLY-SYNDACTYLY SYNDROME 610713;VACTERL ASSOCIATION 192350;BRACHYDACTYLY TYPE E 113300;SYNPOLYDACTYLY 1 186000;SYNDACTYLY TYPE 5 186300;BRACHYDACTYLY TYPE D 113200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
HPD	TYROSINEMIA TYPE 3 276710;HAWKINSINURIA 140350		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
HPGD	CRANIOOSTEOARTHROPATHY 259100		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HPRT1	LESCH-NYHAN SYNDROME 300322;GOUT HPRT-RELATED 300323		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
HPS1	HERMANSKY-PUDLAK SYNDROME 203300		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HPSE2	UROFACIAL SYNDROME 236730		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HR	ALOPECIA UNIVERSALIS 146550;ATRICHIA WITH PAPULAR LESIONS 209500		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HRAS	CONGENITAL MYOPATHY WITH EXCESS OF MUSCLE SPINDLES 218040;COSTELLO SYNDROME 218040		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
HSD17B10	MENTAL RETARDATION SYNDROMIC X-LINKED TYPE 10 300220;2-METHYL-3-HYDROXYBUTYRYL-COA DEHYDROGENASE DEFICIENCY 300438		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
HSD17B4	D-BIFUNCTIONAL PROTEIN DEFICIENCY 261515;PERRAULT SYNDROME		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HSD3B7	BILE ACID SYNTHESIS DEFECT, CONGENITAL, 1 607765		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HSF4	CATARACT ZONULAR HSF4-RELATED 116800;CATARACT MARNER TYPE 116800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
HSPD1	LEUKODYSTROPHY HYPOMYELINATING TYPE 4, OMIM:612233		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HSPG2	SCHWARTZ-JAMPEL SYNDROME 255800;DYSSEGMENTAL DYSPLASIA SILVERMAN-HANDMAKER TYPE 224410		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HUWE1	MENTAL RETARDATION SYNDROMIC X-LINKED TURNER TYPE 300706		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
HYAL1	MUCOPOLYSACCHARIDOSIS TYPE 9 601492		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HYDIN	CILIARY DYSKINESIA, PRIMARY, 5 608647		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HYLS1	HYDROLETHALUS SYNDROME TYPE 1 236680		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IDS	MUCOPOLYSACCHARIDOSIS TYPE 2 309900		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
IDUA	MUCOPOLYSACCHARIDOSIS TYPE 1H/S 607015;MUCOPOLYSACCHARIDOSIS TYPE 1S 607016;MUCOPOLYSACCHARIDOSIS TYPE 1H 607014		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IER3IP1	IER3IP1-related microcephaly with simplified gyral pattern, epilepsy, and neonatal diabetes		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IFIH1	AICARDI-GOUTIERES SYNDROME 7, OMIM:615846;SINGLETON-MERTEN SYNDROME, OMIM:182250		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
IFITM5	OSTEOGENESIS IMPERFECTA TYPE V 610967		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
IFT122	CRANIOECTODERMAL DYSPLASIA 218330		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IFT140	MAINZER-SALDINO SYNDROME 266920		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IFT43	CRANIOECTODERMAL DYSPLASIA TYPE 3 614099		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IFT80	ASPHYXIATING THORACIC DYSTROPHY 2 611263		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IGF1	Insulin-like growth factor I deficiency, OMIM:608747		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IGF1R	INSULIN-LIKE GROWTH FACTOR I, RESISTANCE TO 270450		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
IGF2	BECKWITH-WIEDEMANN SYNDROME 130650;CHROMOSOME 11P15.5-RELATED RUSSELL-SILVER SYNDROME 180860		MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)	DDG2P			5.1
IGFBP7	RETINAL ARTERIAL MACROANEURYSM WITH SUPRAVALVULAR PULMONIC STENOSIS 614224		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IGHMBP2	SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS 1 604320		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IHH	ACROCAPITOFEMORAL DYSPLASIA 607778;BRACHYDACTYLY, TYPE A1 112500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
IKBKG	SUSCEPTIBILITY TO X-LINKED FAMILIAL ATYPICAL MICOBACTERIOSIS TYPE 1 300636;IMMUNODEFICIENCY NEMO-RELATED WITHOUT ANHIDROTIC ECTODERMAL DYSPLASIA 300584;ECTODERMAL DYSPLASIA ANHIDROTIC WITH IMMUNODEFICIENCY-OSTEOPETROSIS-LYMPHEDEMA 300301;ECTODERMAL DYSPLASIA ANHIDROTIC WITH IMMUNODEFICI		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
IL11RA	Autosomal Recessive Craniosynostosis		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IL1RAPL1	MENTAL RETARDATION X-LINKED TYPE 21 300143		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
IMPAD1	CHONDRODYSPLASIA WITH JOINT DISLOCATIONS, GRAPP TYPE 614078		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IQSEC2	MENTAL RETARDATION X-LINKED TYPE 1 309530		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
IRF6	VAN DER WOUDE SYNDROME 119300;POPLITEAL PTERYGIUM SYNDROME 119500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
IRX5	HYPERTELORISM, SEVERE, WITH MIDFACE PROMINENCE, MYOPIA, MENTAL RETARDATION, AND BONE FRAGILITY 611174		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ISPD	WALKER WARBURG SYNDROME 614643		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ITCH	AUTOIMMUNE DISEASE, SYNDROMIC MULTISYSTEM 613385		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ITGA3	INTERSTITIAL LUNG DISEASE, NEPHROTIC SYNDROME, AND EPIDERMOLYSIS BULLOSA, CONGENITAL 614748		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ITGA7	CONGENITAL MUSCULAR DYSTROPHY 607855		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ITPR1	Gillespie Syndrome, OMIM:206700;Gillespie Syndrome, monoallelic, OMIM:206700;SPINOCEREBELLAR ATAXIA 29, CONGENITAL NONPROGRESSIVE, OMIM:117360		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
IVD	ISOVALERIC ACIDEMIA 243500		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
JAG1	ALAGILLE SYNDROME 279357		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
JAG2	JAG2-related muscular dystrophy		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
JAK3	SEVERE COMBINED IMMUNE DEFICIENCY, AUTOSOMAL RECESSIVE, T CELL-NEGATIVE, B CELL -POSITIVE, NK CELL-NEGATIVE, JAK3-RELATED 600802		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
JAM3	HEMORRHAGIC DESTRUCTION OF THE BRAIN, SUBEPENDYMAL CALCIFICATION, AND CATARACTS 613730		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
KANSL1	CHROMOSOME 17Q21.31 MICRODELETION SYNDROME 610443		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KARS	DEAFNESS, AUTOSOMAL RECESSIVE 89, OMIM:613916;KARS1-related leukoencephalopathy with or without deafness, OMIM:619147		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
KAT6B	BLEPHAROPHIMOSIS/INTELLECTUAL DISABILITY PHENOTYPE WHICH IS NOONAN-LIKE;GENITOPATELLAR SYNDROME 606170		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KBTBD13	NEMALINE MYOPATHY 6 609273		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KCNC3	SPINOCEREBELLAR ATAXIA TYPE 13 605259		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KCND2	KCND2-related neurodevelopmental disorder with or without seizures		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KCND3	KCND3-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KCNE1	Jervell and Lange-Nielsen syndrome 2, OMIM:612347;Long QT syndrome 5, OMIM:613695		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
KCNJ10	SEIZURES-SENSORINEURAL DEAFNESS-ATAXIA-MENTAL RETARDATION-ELECTROLYTE IMBALANCE 612780		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
KCNJ11	DIABETES MELLITUS, KCNJ11-RELATED TRANSIENT NEONATAL 261090;FAMILIAL HYPERINSULINISM 3272		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
KCNJ6	KEPPEN-LUBINSKY SYNDROME 614098		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KCNJ8	Cantu syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KCNMA1	GENERALIZED EPILEPSY AND PAROXYSMAL DYSKINESIA 609446		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
KCNN3	ZIMMERMANN-LABAND SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KCNQ1	JERVELL AND LANGE-NIELSEN SYNDROME TYPE 1 220400		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
KCNQ2	EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 7 613720;BENIGN NEONATAL EPILEPSY TYPE 1 121200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KCNQ3	KCNQ3 syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KCTD7	NEURONAL CEROID LIPOFUSCINOSIS;PROGRESSIVE MYOCLONIC EPILEPSY TYPE 3 611726		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
KDM5C	MENTAL RETARDATION SYNDROMIC X-LINKED JARID1C-RELATED 300534		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
KDM6A	KABUKI SYNDROME 2 300867		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
KDM6B	AUTOSOMAL RECESSIVE MENTAL RETARDATION;KDM6B-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KIF11	AUTOSOMAL-DOMINANT MICROCEPHALY ASSOCIATED WITH LYMPHEDEMA AND/OR CHORIORETINOPATHY 152950		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KIF1A	NEUROPATHY, HEREDITARY SENSORY, TYPE IIC, 614213;NESCAV SYNDROME, 614255		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
KIF22	SPONDYLOEPIMETAPHYSEAL DYSPLASIA WITH JOINT LAXITY, TYPE 2 603546		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KIF5A	KIF5A-associated severe neonatal myoclonus, OMIM:617235		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KIF5B	KIF5B-related disease		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KIF7	ACROCALLOSAL SYNDROME 200990;AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
KIT	HUMAN PIEBALDISM 172800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KLF1	ANEMIA, DYSERYTHROPOIETIC CONGENITAL, TYPE IV 613673		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KLF7	KLF7-related developmental disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KLHL7	Cold-induced sweating syndrome type 1 (CISS1-like Phenotype Associated with Early-Onset Retinitis Pigmentosa;Crisponi/CISS1-like phenotype associated with early-onset retinitis pigmentosa		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
KMT2C	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KMT2D	KABUKI SYNDROME 147920		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KMT2E	INTELLECTUAL DISABILITY;Neurodevelopmental disorder and Epilepsy 618512		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KRAS	CARDIOFACIOCUTANEOUS SYNDROME 115150;NOONAN SYNDROME TYPE 3 609942		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1

KRIT1	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 1, OMIM:116860		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KRT74	HYPOTRICHOSIS SIMPLEX OF THE SCALP 2 613981		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
L1CAM	MENTAL RETARDATION-APHASIA-SHUFFLING GAIT-ADDUCTED THUMBS SYNDROME 303350;SPASTIC PARAPLEGIA X-LINKED TYPE 1 303350;HYDROCEPHALUS DUE TO STENOSIS OF THE AQUEDUCT OF SYLVIUS 307000;PARTIAL AGENESIS OF THE CORPUS CALLOSUM 304100		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
L2HGDH	L-2-HYDROXYGLUTARIC ACIDURIA 236792		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LAMA1	CEREBELLAR DYSPLASIA WITH CYSTS WITH OR WITHOUT RETINAL DYSTROPHY 615960;AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LAMA2	CONGENITAL MUSCULAR DYSTROPHY 607855		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LAMB1	COBBLESTONE BRAIN MALFORMATION WITHOUT MUSCULAR OR OCULAR ABNORMALITIES 615191		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LAMC3	OCCIPITAL CORTICAL MALFORMATIONS 614115		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LAMP2	DANON DISEASE 300257		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
LARS2	PERRAULT SYNDROME		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LBR	HYDROPS-ECTOPIC CALCIFICATION-MOTH-EATEN SKELETAL DYSPLASIA 215140		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LEMD3	MELORHEOSTOSIS 155950;BUSCHKE-OLLENDORFF SYNDROME 166700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
LETM1	LETM1-related neurodevelopmental disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LFNG	SPONDYLOCOSTAL DYSOSTOSIS TYPE 3 609813		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LHX3	PITUITARY HORMONE DEFICIENCY COMBINED TYPE 3 221750		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LHX4	LHX4-RELATED COMBINED PITUITARY HORMONE DEFICIENCY 290135		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
LIAS	Neonatal-onset epilepsy, defective mitochondrial energy metabolism, and glycine elevation		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LIFR	Stuve-Wiedeman syndrome, OMIM:601559		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LIG4	SEVERE COMBINED IMMUNODEFICIENCY AUTOSOMAL RECESSIVE T-CELL-NEGATIVE/B-CELL-NEGATIVE/NK-CELL-POSITIVE WITH SENSITIVITY TO IONIZING RADIATION 602450;LIG4 SYNDROME 606593		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LIPN	ICHTHYOSIS, LAMELLAR, 4 613943		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LMBRD1	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA TYPE CBLF 277380		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LMNA	EMERY-DREIFUSS MUSCULAR DYSTROPHY TYPE 2, OMIM:181350;FAMILIAL PARTIAL LIPODYSTROPHY TYPE 2, OMIM:151660;CHARCOT-MARIE-TOOTH DISEASE TYPE 2B1, OMIM:605588;HUTCHINSON-GILFORD PROGERIA SYNDROME, OMIM:176670		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
LMNB1	LMNB1-associated developmental disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
LMNB2	LMNB2-related Primary Microcephaly		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
LMX1B	NAIL-PATELLA SYNDROME 161200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
LRAT	LEBER CONGENITAL AMAUROSIS 608553		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LRP2	DONNAI-BARROW SYNDROME, OMIM:222448;INTELLECTUAL DISABILITY, OMIM:616579		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LRP4	CENANI-LENZ SYNDACTYLY SYNDROME 212780		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LRP5	VITREORETINOPATHY EXUDATIVE TYPE 4 601813;OSTEOPOROSIS-PSEUDOGLIOMA SYNDROME 259770;HIGH BONE MASS TRAIT 601884;OSTEOPETROSIS AUTOSOMAL DOMINANT TYPE 1 607634;ENDOSTEAL HYPEROSTOSIS WORTH TYPE 144750		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
LRPPRC	LEIGH SYNDROME, FRENCH-CANADIAN TYPE 220111		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LRRC6	PRIMARY CILIARY DISKINESIA 614935		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LTBP1	LTBP1-related cutis laxa and craniosynostosis		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LTBP2	MICROSPHEROPHAKIA 251750;PRIMARY CONGENITAL GLAUCOMA TYPE 3D 613086		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LTBP3	PLATYSPONDYLY WITH AMELOGENESIS IMPERFECTA 601216		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
LYST	CHEDIAK-HIGASHI SYNDROME 214500		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MADD	MADD-related developmental disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MAF	Ayme-Gripp syndrome: CATARACT, DEAFNESS, INTELLECTUAL DISABILITY, SEIZURES, AND A DOWN SYNDROME-LIKE FACIES;CATARACT PULVERULENT JUVENILE-ONSET MAF-RELATED 610202;CATARACT CONGENITAL CERULEAN TYPE 4 610202		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MAFB	MULTICENTRIC CARPOTARSAL OSTEOLYSIS SYNDROME, OMIM:166300;Duane Syndrome, Aberrant Extraocular Muscle Innervation, and Inner-Ear Defects		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MAGEL2	Schaaf-Yang syndrome, OMIM:615547		MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)	DDG2P			5.1
MAMLD1	X-LINKED HYPOSPADIAS TYPE 2 300758		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
MAN1B1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MAN2B1	LYSOSOMAL ALPHA-MANNOSIDOSIS 248500		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MANBA	LYSOSOMAL BETA-MANNOSIDOSIS 248510		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MAOA	BRUNNER SYNDROME 300615		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
MAP2K1	CARDIOFACIOCUTANEOUS SYNDROME 115150		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MAP2K2	CARDIOFACIOCUTANEOUS SYNDROME 115150		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MAP3K1	46XY SEX REVERSAL 6 613762		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MASP1	3MC SYNDROME 1 257920		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MAT1A	METHIONINE ADENOSYLTRANSFERASE DEFICIENCY 250850		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MATN3	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 5 607078		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MBD5	EHMT1-LIKE INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MC2R	GLUCOCORTICOID DEFICIENCY 1 202200		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MCCC1	3-METHYLCROTONYL-COA CARBOXYLASE DEFICIENCY 210200		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MCCC2	3-METHYLCROTONYL-COA CARBOXYLASE 2 DEFICIENCY 210210		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MCEE	METHYLMALONYL-COA EPIMERASE DEFICIENCY 251120		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MCOLN1	MUCOLIPIDOSIS IV 252650		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MCPH1	MICROCEPHALY PRIMARY TYPE 1 251200		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MECP2	MENTAL RETARDATION SYNDROMIC X-LINKED LUBS TYPE 300260;MENTAL RETARDATION SYNDROMIC X-LINKED TYPE 13 300055;CHROMOSOME XQ28 DUPLICATION SYNDROME 300815;ENCEPHALOPATHY NEONATAL SEVERE DUE TO MECP2 MUTATIONS 300673;RETT SYNDROME (RTT)[312750		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
MED12	OPITZ-KAVEGGIA SYNDROME 305450;LUJAN-FRYNS SYNDROME 309520		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
MED13	MED13 - Neurodevelopment disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MED13L	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MED17	MICROCEPHALY, POSTNATAL PROGRESSIVE, WITH SEIZURES AND BRAIN ATROPHY 613668		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MED25	Basel-Vanagaite-Smirin-Yosef Syndrome, OMIM:616449		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MEF2C	MENTAL RETARDATION-STEREOTYPIC MOVEMENTS-EPILEPSY AND/OR CEREBRAL MALFORMATIONS 613443		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MEGF10	MYOPATHY, EARLY-ONSET, AREFLEXIA, RESPIRATORY DISTRESS, AND DYSPHAGIA 614399		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MEIS2	MEIS2-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MESP2	SPONDYLOCOSTAL DYSOSTOSIS TYPE 2 608681		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MFF	ENCEPHALOPATHY DUE TO DEFECTIVE MITOCHONDRIAL AND PEROXISOMAL FISSION 2, OMIM:617086		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MFN2	MFN2-related developmental disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MFRP	NANOPHTHALMOS 2 609549;MICROPHTHALMIA ISOLATED TYPE 5 611040		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MFSD2A	Microcephaly 15, primary, autosomal recessive, 616486		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MFSD8	MFSD8-RELATED NEURONAL CEROID-LIPOFUSCINOSIS 610951		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MGAT2	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 2A 212066		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MGP	KEUTEL SYNDROME 245150		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MID1	OPITZ G/BBB SYNDROME, X-LINKED 300000		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
MIR17HG	FEINGOLD SYNDROME 614326		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MITF	TIETZ SYNDROME, OMIM:103500;WAARDENBURG SYNDROME TYPE 2A, OMIM:193510;Coloboma, Osteopetrosis, Microphthalmia, Macrocephaly, Albinism, and Deafness, OMIM:617306		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
MKKS	BARDET-BIEDL SYNDROME TYPE 6 209900;MCKUSICK-KAUFMAN SYNDROME 236700		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MKS1	MECKEL SYNDROME TYPE 1 249000;BARDET-BIEDL SYNDROME TYPE 13 209900		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MLC1	LEUKOENCEPHALOPATHY MEGALENCEPHALIC WITH SUBCORTICAL CYSTS 604004		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MLYCD	MALONYL-COA DECARBOXYLASE DEFICIENCY 167676		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MMAA	METHYLMALONIC ACIDURIA TYPE CBLA 251100		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MMAB	METHYLMALONIC ACIDURIA TYPE CBLB 251110		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MMACHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, CBLC TYPE 277400		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MMADHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA TYPE CBLD 277410		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MMP13	METAPHYSEAL ANADYSPLASIA TYPE 1 602111;SPONDYLOEPIMETAPHYSEAL DYSPLASIA MISSOURI TYPE 602111		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
MNX1	CURRARINO SYNDROME 176450		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MOCS1	MOLYBDENUM COFACTOR DEFICIENCY 603707		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MOCS2	MOLYBDENUM COFACTOR DEFICIENCY 603707		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MOGS	CONGENITAL DISORDERS OF GLYCOSYLATION 612379		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MPDU1	CONGENITAL DISORDERS OF GLYCOSYLATION 612379		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MPI	CONGENITAL DISORDERS OF GLYCOSYLATION 612379		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MPLKIP	TRICHOTHIODYSTROPHY NON-PHOTOSENSITIVE TYPE 1 234050		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MPV17	MITOCHONDRIAL DNA DEPLETION SYNDROME 6 256810		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MPZ	NEUROPATHY, CONGENITAL HYPOMYELINATING, 2, OMIM:618184		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MRPS22	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 5 611719		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MSX1	CLEFT LIP +/- CLEFT PALATE 608874		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MSX2	ENLARGED PARIETAL FORAMINA/CRANIUM BIFIDUM 168550;CRANIOSYNOSTOSIS, TYPE 2 604757		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MTFMT	MTFMT-related mitochondrial disease with regression and lactic acidosis		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MTHFR	METHYLENETETRAHYDROFOLATE REDUCTASE DEFICIENCY 236250		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MTM1	MYOTUBULAR MYOPATHY, X-LINKED 310400		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
MTO1	INFANTILE HYPERTROPHIC CARDIOMYOPATHY AND LACTIC ACIDOSIS 614702		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MTR	METHYLCOBALAMIN DEFICIENCY TYPE G 250940		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MTRR	HOMOCYSTINURIA-MEGALOBLASTIC ANEMIA, CBL E TYPE 236270		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MUT	METHYLMALONIC ACIDURIA TYPE MUT 251000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MYBPC1	LETHAL CONGENITAL CONTRACTURE SYNDROME 4, OMIM:614915;MYBPC1-related arthrogryposis and myopathy		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
MYCN	FEINGOLD SYNDROME TYPE 1 164280		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MYH3	DISTAL ARTHROGRYPOSIS TYPE 2A, OMIM:193700;Recessive Spondylocarpotarsal Synostosis Syndrome		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
MYH8	DISTAL ARTHROGRYPOSIS TYPE, OMIM:158300;CARNEY COMPLEX VARIANT, OMIM:608837		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MYH9	DEAFNESS AUTOSOMAL DOMINANT TYPE 17 603622;SEBASTIAN SYNDROME 155100;MACROTHROMBOCYTOPENIA WITH PROGRESSIVE SENSORINEURAL DEAFNESS 155100;EPSTEIN SYNDROME 155100;MAY-HEGGLIN ANOMALY 155100;FECHTNER SYNDROME 155100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MYLK	Megacystis Microcolon Intestinal Hypoperistalsis Syndrome		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MYO18B	KLIPPEL-FEIL SYNDROME 4, AUTOSOMAL RECESSIVE, WITH NEMALINE MYOPATHY AND FACIAL DYSMORPHISM, OMIM:616549		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MYO5A	ELEJALDE SYNDROME 256710;GRISCELLI SYNDROME TYPE 3 609227		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MYO5B	MICROVILLUS INCLUSION DISEASE 251850		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MYO7A	USHER SYNDROME TYPE 1B 276900;DEAFNESS AUTOSOMAL RECESSIVE TYPE 2 600060		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MYOCD	Congenital megabladder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MYPN	Childhood-Onset, Slowly Progressive Nemaline Myopathy		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MYT1L	MYT1L syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NAA10	X-linked anophthalmia syndrome/Lenz;NONPECIFIC SEVERE ID;X-linked anophthalmia syndrome;OGDEN SYNDROME 300855		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
NAGA	SCHINDLER DISEASE 609241;KANZAKI DISEASE 609242		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NAGLU	MUCOPOLYSACCHARIDOSIS TYPE 3B 252920		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NAGS	N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY 237310		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NARS2	NARS2-associated oxidative phosphorylation deficiency		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NBAS	ACUTE LIVER FAILURE (ALF) IN INFANCY AND CHILDHOOD;ACUTE LIVER FAILURE (ALF) IN INFANCY AND CHILDHOOD;SHORT STATURE, OPTIC NERVE ATROPHY, AND PELGER-HUET ANOMALY 616483		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NBEA	NBEA Neurodevelopment disorder with seizures		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NBN	NIJMEGEN BREAKAGE SYNDROME 251260		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDE1	LISSENCEPHALY 4 614019		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDP	NORRIE DISEASE 310600		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
NDST1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFA1	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY 252010		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
NDUFA10	LEIGH SYNDROME DUP 256000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFA12	NDUFA12-associated mtochondrial complex I deficiency, OMIM:618244		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFA6	Early Onset Isolated Mitochondrial Complex I Deficiency		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFA8	NDUFA8-related developmental disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFAF2	LEIGH SYNDROME 256000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFB3	NDUFB3-associated mitochondrial complex I deficiency, OMIM:618246		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFS1	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY 252010;LEIGH SYNDROME 256000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFS4	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY 252010;LEIGH SYNDROME 256000;LEIGH SYNDROME DUP 256000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFS7	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY 252010		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFS8	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY 252010		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFV1	MITOCHONDRIAL COMPLEX I DEFICIENCY 252010		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NDUFV2	Mitochondrial complex I deficiency, nuclear type 7, OMIM:618229		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NEB	AUTOSOMAL RECESSIVE TYPICAL NEMALINE MYOPATHY 256030		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NEDD4L	Periventricular nodular heterotopia with ID, cleft palate and 2.3 toe syndactyly		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NEK1	SHORT RIB-POLYDACTYLY SYNDORME, TYPE II 263520		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NEK8	RENAL-HEPATIC-PANCREATIC DYSPLASIA 2 615415;NEPHRONOPHTHISIS 9 613824		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NEU1	SIALIDOSIS 256550		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NF1	WATSON SYNDROME 193520;NEUROFIBROMATOSIS TYPE 1 162200;NEUROFIBROMATOSIS-NOONAN SYNDROME 601321;FAMILIAL SPINAL NEUROFIBROMATOSIS 162210		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NFE2L2	NFE2L2-related leukoencephalopathy, immune deficiency and hypohomocysteinaemia, OMIM:617744		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NFIA	Macrocephaly with intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NFIX	MARSHALL-SMITH SYNDROME 602535;SOTOS-LIKE SYNDROME 614753		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NFU1	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 1 605711		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NHP2	DYSKERATOSIS CONGENITA, AUTOSOMAL RECESSIVE 2 613987		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NHS	NANCE-HORAN SYNDROME 302350;CATARACT CONGENITAL X-LINKED 302200		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
NIPBL	CORNELIA DE LANGE SYNDROME TYPE 1 122470		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NKX2-1	CHOREOATHETOSIS, HYPOTHYROIDISM, AND NEONATAL RESPIRATORY DISTRESS 610978;BENIGN HEREDITARY CHOREA 118700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NKX2-5	ATRIAL SEPTAL DEFECT WITH ATRIOVENTRICULAR CONDUCTION DEFECTS 108900;TETRALOGY OF FALLOT 187500;CONGENITAL HYPOTHYROIDISM NON-GOITROUS TYPE 5 225250		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NKX3-2	SPONDYLO-MEGAEPIPHYSEAL-METAPHYSEAL DYSPLASIA 613330		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NLGN3	AUTISM SPECTRUM DISORDERS 198890		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
NLGN4X	SUSCEPTIBILITY TO AUTISM X-LINKED TYPE 2 300495		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
NMNAT1	LEBER CONGENITAL AMAUROSIS 608553		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NODAL	HETEROTAXY SYNDROME 207574		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NOG	TARSAL-CARPAL COALITION SYNDROME 186570;MULTIPLE SYNOSTOSES SYNDROME TYPE 1 186500;STAPES ANKYLOSIS WITH BROAD THUMB AND TOES 184460;BRACHYDACTYLY TYPE B2 611377;SYMPHALANGISM PROXIMAL SYNDROME 185800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NOTCH1	LEFT VENTRICULAR OUTFLOW TRACT OBSTRUCTION 109730;ADAMS OLIVER SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NOTCH2	HAJDU-CHENEY SYNDROME 102500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NPC1	NIEMANN-PICK DISEASE, TYPE C1 257220		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NPC2	NIEMANN-PICK DISEASE, TYPE C2 607625		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NPHP1	JOUBERT SYNDROME TYPE 4 609583;NEPHRONOPHTHISIS TYPE 1 256100;SENIOR-LOKEN SYNDROME TYPE 1 266900		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NPHP3	NEPHRONOPHTHISIS TYPE 3 604387;RENAL-HEPATIC-PANCREATIC DYSPLASIA 208540;MECKEL SYNDROME TYPE 7 267010		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NPHP4	NEPHRONOPHTHISIS TYPE 4 606966		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NPHS1	NEPHROTIC SYNDROME TYPE 1 256300		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NPHS2	NEPHROTIC SYNDROME, TYPE 2 600995		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NPM1	Dyskeratosis Congenita		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NPR2	ACROMESOMELIC DYSPLASIA MAROTEAUX TYPE 602875		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NPR3	Enhanced Growth and Connective Tissue Abnormalities		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NR2F1	BOSCH-BOONSTRA OPTIC ATROPHY SYNDROME 615722		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1

NR4A2	NR4A2-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NRAS	NOONAN SYNDROME TYPE 6 613224		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NRXN1	PITT HOPKINS 2;AUTISM 209850		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
NRXN2	AUTISM 209850		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NSD1	WEAVER SYNDROME 277590;BECKWITH-WIEDEMANN SYNDROME 130650;SOTOS SYNDROME 117550		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NSDHL	CK SYNDROME 300831;CONGENITAL HEMIDYSPLASIA WITH ICHTHYOSIFORM ERYTHRODERMA AND LIMB DEFECTS 308050		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
NSUN2	AUTOSOMAL- RECESSIVE INTELLECTUAL DISABILITY MRT5 611091		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NT5C3A	HEMOLYTIC ANEMIA DUE TO UMPH1 DEFICIENCY 266120		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NTRK1	CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS 256800		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NTRK2	Epilepsy and intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NUBPL	MITOCHONDRIAL COMPLEX I DEFICIENCY 252010		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NUP62	INFANTILE STRIATONIGRAL DEGENERATION 271930		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NYX	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1A 310500		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
OBSL1	3-M SYNDROME 2 612921		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
OCLN	OCLN-associated Band-like Calcification with Simplified Gyration and Polymicrogyria		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
OCRL	DENT DISEASE TYPE 2 300555;LOWE OCULOCEREBRORENAL SYNDROME 309000		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
ODC1	ODC1-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
OFD1	SIMPSON-GOLABI-BEHMEL SYNDROME TYPE 2 300209;ORAL-FACIAL-DIGITAL SYNDROME TYPE 1 311200;JOUBERT SYNDROME TYPE 10 300804		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
OPHN1	MENTAL RETARDATION X-LINKED OPHN1-RELATED 300486		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
ORC1	MEIER-GORLIN SYNDROME 1 224690		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ORC4	MEIER-GORLIN SYNDROME 2 613800		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ORC6	MEIER-GORLIN SYNDROME 3 613803		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
OTC	ORNITHINE TRANSCARBAMYLASE DEFICIENCY 311250		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
OTX2	MICROPHTHALMIA SYNDROMIC TYPE 5 610125		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
OXCT1	SUCCINYL-COA-3-KETOACID-COA TRANSFERASE DEFICIENCY 245050		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PAFAH1B1	SUBCORTICAL BAND HETEROTOPIA 607432;LISSENCEPHALY TYPE 1 607432		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PAH	NON-PHENYLKETONURIA HYPERPHENYLALANINEMIA 261600;PHENYLKETONURIA 261600		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PAK3	AGENESIS OF THE CORPUS CALLOSUM;MENTAL RETARDATION X-LINKED TYPE 30 300558		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
PALB2	FANCONI ANEMIA, COMPLEMENTATION GROUP N 610832		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PAPSS2	SPONDYLOEPIMETAPHYSEAL DYSPLASIA PAKISTANI TYPE 612847		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PAX1	OTOFACIOCERVICAL SYNDROME, OMIM:166780		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PAX2	RENAL-COLOBOMA SYNDROME 120330		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PAX3	WAARDENBURG SYNDROME, TYPE 1 193500;CRANIOFACIAL-DEAFNESS-HAND SYNDROME 122880		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
PAX6	COLOBOMA OF OPTIC NERVE 120430;ANIRIDIA 106210;FOVEAL HYPOPLASIA 136520;BILATERAL OPTIC NERVE HYPOPLASIA 165550;KERATITIS HEREDITARY 148190;PETERS ANOMALY 604229;ANIRIDIA CEREBELLAR ATAXIA AND MENTAL DEFICIENCY 206700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PAX8	CONGENITAL HYPOTHYROIDISM NON-GOITROUS TYPE 2 218700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PAX9	TOOTH AGENESIS, SELECTIVE, 3 604625		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PC	PYRUVATE CARBOXYLASE DEFICIENCY 266150		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PCBD1	HYPERPHENYLALANINEMIA, BH4-DEFICIENT, D 264070		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PCCA	PROPIONIC ACIDEMIA 606054		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PCCB	PROPIONIC ACIDEMIA 606054		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PCDH19	EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 9, OMIM:300088		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, OMIM:210720		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PDCD10	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 3 603285		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PDE10A	Childhood-Onset Chorea with Bilateral Striatal Lesions		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PDE4D	ACRODYSOSTOSIS 101800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PDE6G	RETINITIS PIGMENTOSA 57 613582		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PDE6H	RETINAL CONE DYSTROPHY 3 PDE6H 610024;ACHROMATOPSIA		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PDHA1	INTELLECTUAL DISABILTIY 312170;X-LINKED LEIGH SYNDROME 312170;PYRUVATE DEHYDROGENASE E1-ALPHA DEFICIENCY IN FEMALES 312170		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
PDHX	LACTICACIDEMIA DUE TO PDX1 DEFICIENCY 245349		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PDSS1	COENZYME Q10 DEFICIENCY, PRIMARY, 2 614651		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PDSS2	COENZYME Q10 DEFICIENCY, PRIMARY, 3 614652		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEPD	PROLIDASE DEFICIENCY 170100		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX1	PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 1 602136;ADRENOLEUKODYSTROPHY NEONATAL 202370;INFANTILE REFSUM DISEASE 266510		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX10	ZELLWEGER SYNDROME 214100;ADRENOLEUKODYSTROPHY NEONATAL 202370;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 7 602859		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX11B	Peroxisome biogenesis disorder 14B		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX12	ZELLWEGER SYNDROME 214100;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 3 601758		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX13	PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 13 601789;ADRENOLEUKODYSTROPHY NEONATAL 202370		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX14	ZELLWEGER SYNDROME 214100;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP K 601791		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX16	ZELLWEGER SYNDROME 214100;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 9 603360		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX19	ZELLWEGER SYNDROME 214100;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 14 600279		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX2	ZELLWEGER SYNDROME 214100;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 5 170993;INFANTILE REFSUM DISEASE 266510		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX26	ZELLWEGER SYNDROME 214100;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 8 608666;ADRENOLEUKODYSTROPHY NEONATAL 202370;INFANTILE REFSUM DISEASE 266510		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX3	ZELLWEGER SYNDROME 214100;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 12 603164		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX5	ZELLWEGER SYNDROME 214100;ADRENOLEUKODYSTROPHY NEONATAL 202370;INFANTILE REFSUM DISEASE 266510		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX6	Heimler syndrome 2, OMIM:616617;Peroxisome biogenesis disorder 4A (Zellweger), OMIM:614862;Peroxisome biogenesis disorder 4B, OMIM:614863		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PEX7	REFSUM DISEASE 266500;RHIZOMELIC CHONDRODYSPLASIA PUNCTATA TYPE 1 215100;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 11 601757		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PGK1	PHOSPHOGLYCERATE KINASE 1 DEFICIENCY 300653		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
PGM1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IT 614921		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PHF6	BOERJESON-FORSSMAN-LEHMANN SYNDROME 301900		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
PHF8	MENTAL RETARDATION SYNDROMIC X-LINKED SIDERIUS TYPE 300263		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
PHGDH	PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY 601815;NEU-LAXOVA SYNDROME 256520		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PHIP	Developmental delay, ID, obesity and dysmorphic features		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PHOX2B	CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL, WITH OR WITHOUT HIRSCHSPRUNG DISEASE 209880;NEUROBLASTOMA WITH HIRSCHSPRUNG DISEASE 613013		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PIEZO2	ARTHROGRYPOSIS, DISTAL, TYPE 3 114300;Ataxia, dysmetria, contractures & scoliosis with normal cognition but loss of discriminative touch perception		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
PIGA	MULTIPLE CONGENITAL ANOMALIES-HYPOTONIA-SEIZURES SYNDROME 2 300868		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
PIGL	ZUNICH NEUROECTODERMAL SYNDROME 280000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PIGN	MULTIPLE CONGENITAL ANOMALIES-HYPOTONIA-SEIZURES SYNDROME 614080		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PIGO	HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 2 614749		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PIGV	HYPERPHOSPHATASIA WITH MENTAL RETARDATION 239300		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PIK3CA	HEMIMEGALENCEPHALY PIK3CA;CLOVES: CONGENITAL LIPOMATOUS OVERGROWTH, VASCULAR MALFORMATIONS, AND EPIDERMAL NEVI, OMIM:612918;MEGALENCEPHALY-CAPILLARY MALFORMATION-POLYMICROGYRIA SYNDROME, SOMATIC 3, OMIM:602501		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PIK3R1	SHORT SYNDROME 269880;AGAMMAGLOBULINEMIA 7, AUTOSOMAL RECESSIVE 615214		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
PIK3R2	MEGALENCEPHALY-POLYMICROGYRIA-POLYDACTYLY-HYDROCEPHALUS SYNDROME 1 603387		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PIP5K1C	LETHAL CONGENITAL CONTRACTURE SYNDROME TYPE 3 611369		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PITX1	CONGENITAL CLUBFOOT 119800;HOMEOTIC ARM-TO-LEG TRANSFORMATION ASSOCIATED WITH GENOMIC REARRANGEMENTS AT THE PITX1 LOCUS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PITX2	AXENFELD-RIEGER SYNDROME TYPE 1 180500;PETERS ANOMALY 604229;RING DERMOID OF CORNEA 180550;IRIDOGONIODYSGENESIS TYPE 2 137600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PITX3	CATARACT AUTOSOMAL DOMINANT 604219;CATARACT POSTERIOR POLAR TYPE 4 610623;ANTERIOR SEGMENT MESENCHYMAL DYSGENESIS 107250		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PKD1L1	Laterality defects		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PKHD1	POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE 263200		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PLA2G6	INFANTILE NEUROAXONAL DYSTROPHY 1, OMIM:256600		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PLCB1	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 12 613722		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PLCB4	AURICULOCONDYLAR SYNDROME 602483		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PLCE1	NEPHROTIC SYNDROME, TYPE 3 610725		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PLEC	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2Q 613723;EPIDERMOLYSIS BULLOSA SIMPLEX WITH PYLORIC ATRESIA 612138		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PLOD1	EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC FORM 225400		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PLOD2	BRUCK SYNDROME TYPE 2 609220		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PLP1	SPASTIC PARAPLEGIA X-LINKED TYPE 2 312920;LEUKODYSTROPHY HYPOMYELINATING TYPE 1 312080		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
PLXND1	MOEBIUS SYNDROME;TRUNCUS ARTERIOSIS		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PMM2	CONGENITAL DISORDERS OF GLYCOSYLATION 612379		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PMS2	MISMATCH REPAIR CANCER SYNDROME 276300		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PNKP	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 10 613402;ATAXIA-OCULOMOTOR APRAXIA 4 616267		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PNPLA1	CONGENITAL ICHTHYOSIS		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PNPLA2	NEUTRAL LIPID STORAGE DISEASE WITH MYOPATHY 610717		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PNPLA6	PNPLA6-related Disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PNPO	PNPO-related neonatal encephalopathy responsive to pyridoxal-5'-phosphate, OMIM:610090		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
POGZ	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
POLD1	SUBCUTANEOUS LIPODYSTROPHY, DEAFNESS, MANDIBULAR HYPOPLASIA AND MALE HYPOGONADISM		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
POLG	MITOCHONDRIAL DNA DEPLETION SYNDROME 4A 203700		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
POLR1C	TREACHER COLLINS SYNDROME TYPE 3 248390		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
POLR1D	TREACHER COLLINS SYNDROME TYPE 2 613717		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
POLR3A	Autosomal Recessive Wiedemann Rautenstrauch Syndrome;LEUKODYSTROPHY, HYPOMYELINATING, 7, WITH OR WITHOUT OLIGODONTIA AND/OR HYPOGONADOTROPIC HYPOGONADISM 607694		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
POLR3B	Autosomal recessive mental retardation;Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism;POLR3B-related neurodevelopmental disorder		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
POMGNT1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH MENTAL RETARDATION TYPE B3 613151;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C3 613157;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH BRAIN AND EYE ANOMALIES TYPE A3 (MDDGA3 253280		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
POMT1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C1 609308;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH MENTAL RETARDATION TYPE B1 613155;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH BRAIN AND EYE ANOMALIES TYPE A1 236670		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
POMT2	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH BRAIN AND EYE ANOMALIES TYPE A2 613150;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C2 613158;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH MENTAL RETARDATION TYPE B2 613156		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PORCN	FOCAL DERMAL HYPOPLASIA 305600		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
POU1F1	POU1F1-RELATED COMBINED PITUITARY HORMONE DEFICIENCY 613038		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PPM1D	PPM1D syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PPP2R1A	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PPT1	NEURONAL CEROID LIPOFUSCINOSIS TYPE 1 256730		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PQBP1	RENPENNING S(YNDROME 1 309500		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
PREPL	HYPOTONIA-CYSTINURIA SYNDROME 606407		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PRKACA	PRKACA-related Multiple Congenital Malformation Syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PRKAR1A	ACRODYSOSTOSIS 101800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PRMT7	Pseudohypoparathyroidism-like disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PROP1	PROP1-RELATED COMBINED PITUITARY HORMONE DEFICIENCY 262600		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PRPF8	PRPF8-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PRPS1	ARTS SYNDROME 301835;PHOSPHORIBOSYLPYROPHOSPHATE SYNTHETASE SUPERACTIVITY 300661;DEAFNESS X-LINKED TYPE 1 304500;CHARCOT-MARIE-TOOTH DISEASE X-LINKED RECESSIVE TYPE 5 311070		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
PRRT2	AUTOSOMAL RECESSIVE MENTAL RETARDATION;BENIGN FAMILIAL INFANTILE EPILEPSY AND INFANTILE CONVULSIONS WITH CHOREOATHETOSIS SYNDROME 602066		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
PRRX1	AGNATHIA-OTOCEPHALY COMPLEX biallelic;AGNATHIA-OTOCEPHALY COMPLEX monoallelic		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PRSS12	MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 1 249500		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PRSS56	MICROPHTHALMIA ISOLATED TYPE 6 613517		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PSAP	ATYPICAL KRABBE DISEASE 611722		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PSAT1	PHOSPHOSERINE AMINOTRANSFERASE DEFICIENCY 610992;NEU-LAXOVA SYNDROME 256520		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PSMB8	NAKAJO SYNDROME 256040		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PSPH	PHOSPHOSERINE PHOSPHATASE DEFICIENCY 614023;NEU-LAXOVA 256520		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PTCH1	BASAL CELL NEVUS SYNDROME 109400;HOLOPROSENCEPHALY-7 610828		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PTCHD1	AUTISM/ID 300830		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
PTEN	BANNAYAN-ZONANA SYNDROME 153480;COWDEN DISEASE 158350;MACROCEPHALY/AUTISM SYNDROME 605309;VACTERL ASSOCIATION WITH HYDROCEPHALUS 276950;PROTEUS SYNDROME 176920;LHERMITTE-DUCLOS DISEASE 158350		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PTF1A	DIABETES MELLITUS, PERMANENT NEONATAL, WITH CEREBELLAR AGENESIS 609069;PANCREATIC AGENESIS		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PTH	FAMILIAL ISOLATED HYPOPARATHYROIDISM 146200		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PTH1R	JANSEN METAPHYSEAL CHONDRODYSPLASIA 156400;EIKEN SKELETAL DYSPLASIA 600002;PRIMARY FAILURE OF TOOTH ERUPTION 125350;CHONDRODYSPLASIA BLOMSTRAND TYPE 215045		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
PTHLH	BRACHYDACTYLY, TYPE E2 613382;CLUBBING WITH SKELETAL DYSPLASIA INC ACROOSTEOLYSIS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PTPN11	LEOPARD SYNDROME TYPE 1 151100;NOONAN SYNDROME 1 163950		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PTPN14	CHOANAL ATRESIA AND LYMPHEDEMA 613611		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PTS	6-PYRUVOYLTETRAHYDROPTERIN SYNTHASE DEFICIENCY 261640		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PUS1	MYOPATHY, LACTIC ACIDOSIS, AND SIDEROBLASTIC ANEMIA 1, OMIM:600462		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PXDN	CONGENITAL CATARACT, CORNEAL OPACITY, AND DEVELOPMENTAL GLAUCOMA		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PYCR1	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE IIB 612940		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PYGL	GLYCOGEN STORAGE DISEASE TYPE VI 232700		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
QDPR	BH4-DEFICIENT HYPERPHENYLALANINEMIA C 261630		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RAB18	WARBURG MICRO SYNDROME TYPE 3 614222		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RAB23	ACROCEPHALOPOLYSYNDACTYLY TYPE 2 201000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RAB39B	MENTAL RETARDATION X-LINKED TYPE 72 (MRX72) +/- PARKINSONS 300271		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
RAB3GAP1	WARBURG MICRO SYNDROME TYPE 1 600118		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RAB3GAP2	MARTSOLF SYNDROME 212720		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RAC1	Developmental Disorders with Diverse Phenotypes		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RAD21	COHESINOPATHY 614701		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RAD51C	FANCONI ANEMIA, COMPLEMENTATION GROUP 0 613390		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RAF1	NOONAN SYNDROME 5 611553		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RAI1	SMITH-MAGENIS SYNDROME 182290		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RALGAPA1	RALGAPA1-related neurodevelopmental disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RAPSN	CONGENITAL MYASTHENIC SYNDROME WITH ACETYLCHOLINE RECEPTOR DEFICIENCY 608931;FETAL AKINESIA DEFORMATION SEQUENCE 208150		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RARS2	PONTOCEREBELLAR HYPOPLASIA TYPE 6 318922		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RAX	MICROPHTHALMIA ISOLATED TYPE 3 611038		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RBBP8	RBBP8-related microcephaly and intellectual disability, OMIM:251255		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RBM10	TARP SYNDROME 311900		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
RBM8A	THROMBOCYTOPENIA-ABSENT RADIUS SYNDROME 274000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RECQL4	RAPADILINO SYNDROME 266280;BALLER-GEROLD SYNDROME 218600;ROTHMUND-THOMSON SYNDROME 268400		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RELN	LISSENCEPHALY 2 319306		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RET	RENAL AGENESIS 191830;MULTIPLE ENDOCRINE NEOPLASIA IIB 162300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RFT1	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1N 612015		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RFX6	MARTINEZ-FRIAS SYNDROME 601346		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RIN2	MACROCEPHALY, ALOPECIA, CUTIS LAXA, AND SCOLIOSIS TALL FOREHEAD, SPARSE HAIR, SKIN HYPEREXTENSIBILITY, AND SCOLIOSIS		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RIPK4	POPLITEAL PTERYGIUM SYNDROME, LETHAL TYPE 263650		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RMRP	CARTILAGE-HAIR HYPOPLASIA 250250		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RNASEH2A	AICARDI-GOUTIERES SYNDROME 4 610333		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1

RNASEH2B	AICARDI-GOUTIERES SYNDROME 2 610181		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
RNASEH2C	AICARDI-GOUTIERES SYNDROME 3 610329		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
RNASET2	LEUKOENCEPHALOPATHY, CYSTIC, WITHOUT MEGALENCEPHALY 612951		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
RNU4ATAC	MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM, TYPE I 210710		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
ROBO3	FAMILIAL HORIZONTAL GAZE PALSY WITH PROGRESSIVE SCOLIOSIS 607313		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
ROGDI	KOHLSCHTTER-TZ SYNDROME 226750		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
ROR2	Brachydactyly, type B1, OMIM 113000 (AD) Robinow syndrome, autosomal recessive, OMIM 268310 (AR)		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
RORA	INTELLECTUAL DISABILITY		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RPE65	LEBER CONGENITAL AMAUROSIS 608553		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
RPGRIP1	LEBER CONGENITAL AMAUROSIS 6 613826;CONE-ROD DYSTROPHY 13 608194		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
RPGRIP1L	JOUBERT SYNDROME TYPE 7;611560;JOUBERT SYNDROME TYPE 7 611560;COACH SYNDROME 216360		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
RPL10	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC, 35, OMIM:300998		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
RPL11	Diamond-Blackfan anemia with cleft palate and abnormal thumbs		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RPS19	RPS19-RELATED DIAMOND-BLACKFAN ANEMIA 220176		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RPS26	DIAMOND-BLACKFAN ANEMIA 10, OMIM:613309		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RPS6KA3	Coffin-Lowry Syndrome 2 RPS6KA3 XLD;Coffin-Lowry Syndrome 2 RPS6KA3 XLR		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
RRM2B	Mitochondrial depletion syndrome		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
RSPO4	ANONYCHIA CONGENITA 206800		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
RSRC1	INTELLECTUAL DEVELOPMENTAL DISORDER, AUTOSOMAL RECESSIVE 70		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
RUNX2	CLEIDOCRANIAL DYSPLASIA 119600		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RYR1	MINICORE MYOPATHY WITH EXTERNAL OPHTHALMOPLEGIA 255320		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SACS	SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY TYPE 270550		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SALL1	TOWNES-BROCKS SYNDROME 107480		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SALL4	ACRO-RENAL-OCULAR SYNDROME 607323;DUANE-RADIAL RAY SYNDROME 607323		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SAMD9	MIRAGE - myelodysplasia, infection, restriction of growth, adrenal hypoplasia, genital phenotypes, enteropathy		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SAMHD1	AICARDI-GOUTIERES SYNDROME 2 612952		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SARS2	SARS2 related hyperuricemia, pulmonary hypertension, renal failure and alkalosis, OMIM 613845		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SART3	SART3-related neurodevelopmental disorder with 46 XY gonadal dysgenesis (NDYGOS)		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SATB2	NONSPECIFIC SEVERE ID;CLEFT PALATE ISOLATED 119540;SYNDROMAL PIERRE ROBIN SEQUENCE		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SBDS	SHWACHMAN-DIAMOND SYNDROME 607444		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SCARF2	VAN DEN ENDE-GUPTA SYNDROME 600920		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SCN1A	SCN1A-RELATED SEIZURE DISORDERS 607208		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SCN1B	EPILEPSY, GENERALIZED, WITH FEBRILE SEIZURES PLUS, TYPE 1 604233;BRUGADA SYNDROME 5 612838		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SCN2A	NONSPECIFIC SEVERE ID;BENIGN FAMILIAL NEONATAL INFANTILE SEIZURES 248968;INFANTILE EPILEPTIC ENCEPHALOPATHY		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SCN3A	Focal epilepsy		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SCN4A	HYPOKALEMIC PERIODIC PARALYSIS 613345;PARAMYOTONIA CONGENITA OF VON EULENBURG 168300;HYPERKALEMIC PERIODIC PARALYSIS TYPE 1 170500		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SCN8A	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 13, 614558		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SCO1	MITOCHONDRIAL COMPLEX IV DEFICIENCY 220110		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SCO2	FATAL INFANTILE CARDIOENCEPHALOMYOPATHY DUE TO CYTOCHROME C OXIDASE DEFICIENCY 604377		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SCOCAH	SENIOR-LOKEN SYNDROME 7 613615		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SDHA	LEIGH SYNDROME 256000		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SDHAF1	MITOCHONDRIAL COMPLEX II DEFICIENCY 252011		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SEC23B	ANEMIA, DYSERYTHROPOIETIC CONGENITAL, TYPE II 224100		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SEC15BP2	THYROID HORMONE METABOLISM, ABNORMAL 609698		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SEMA3A	SEMA3A-related skeletal dysplasia		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SEPSECS	Pontocerebellar hypoplasia type 2D		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SETBP1	DEVELOPMENTAL AND EXPRESSIVE LANGUAGE DELAY-SCHINZEL-GIEDION MIDFACE RETRACTION SYNDROME 269150		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SETD2	SETD2-associated Overgrowth Syndrome		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SF3B4	ACROFACIAL DYSOSTOSIS 1; NAGER TYPE 154400		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SGSH	MUCOPOLYSACCHARIDOSIS TYPE 3A 252900		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SH3PXD2B	FRANK-TER HAAR SYNDROME 249420		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SHANK2	SUSCEPTIBILITY TO AUTISM TYPE 17 613436		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SHANK3	PHELAN-MCDERMID SYNDROME 606232		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SHH	MICROPHTHALMIA ISOLATED WITH COLOBOMA TYPE 5 611638;TRIPHALANGEAL THUMB-POLYSYNDACTYLY SYNDROME 174500;SOLITARY MEDIAN MAXILLARY CENTRAL INCISOR 147250;HOLOPROSENCEPHALY TYPE 3 236100		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SHOC2	NOONAN-LIKE SYNDROME WITH LOOSE ANAGEN HAIR 607721		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SHOX	LERI-WEILL DYSCHONDROSTEOSIS 127300;LANGER MESOMELIC DYSPLASIA 249700		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form; autosomal or pseudoautosomal	DDG2P			5.1
SHROOM3	NEURAL TUBE DEFECT		MONOALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SIL1	MARINESCO-SJOGREN SYNDROME 248800		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SIM1	Severe obesity with neurobehavioral features		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SIX1	BRANCHIOOTIC SYNDROME TYPE 3, OMIM:608389;Non-syndromic craniosynostosis		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SIX3	HOLOPROSENCEPHALY 609637		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SIX5	BRANCHIOOTORENAL SYNDROME TYPE 2 610896		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SKI	SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME 182212		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2 614602		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC12A6	AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL NEUROPATHY 218000		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC16A2	MCT8 (SLC16A2)-SPECIFIC THYROID HORMONE CELL TRANSPORTER DEFICIENCY 300523		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
SLC17A5	INFANTILE SIALIC ACID STORAGE DISORDER 269920;SALLA DISEASE 604369		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC19A3	THIAMINE METABOLISM DYSFUNCTION SYNDROME 2 607483		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC1A2	EPILEPTIC ENCEPHALOPATHY		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SLC22A5	SYSTEMIC PRIMARY CARNITINE DEFICIENCY 212140		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC25A15	HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME 238970		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC25A19	AMISH LETHAL MICROCEPHALY 248305		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY 212138		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC25A22	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 3 319213		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC25A38	ANEMIA, SIDEROBLASTIC, PYRIDOXINE-REFRACTORY, AUTOSOMAL RECESSIVE 205950		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC25A4	Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number Footaine progeroid syndrome		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SLC26A2	ACHONDROGENESIS TYPE 1B 600972;MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 4 226900;ATELOSTEOGENESIS TYPE 2 256050;DIASTROPHIC DYSPLASIA 222600		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC27A4	ICHTHYOSIS, PREMATURITY, SYNDROME 608649		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC2A1	GLUT1 DEFICIENCY SYNDROME TYPE 2 612126 GLUT1 DEFICIENCY SYNDROME TYPE 1 606777		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SLC2A10	ARTERIAL TORTUOSITY SYNDROME 208050		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC2A2	FANCONI-BICKEL SYNDROME 227810		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC31A1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC33A1	AUTOSOMAL-RECESSIVE DISORDER WITH CONGENITAL CATARACTS, HEARING LOSS, AND LOW SERUM COPPER AND CERULOPLASMIN		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC35A3	CONGENITAL DISORDERS OF GLYCOSYLATION 615274		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC35C1	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 2C 266265		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC35D1	SCHNECKENBECKEN DYSPLASIA 269250		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC37A4	?GLC37A4-related congenital disorder of glycosylation with liver dysfunction;Glycogen storage disease Ib, OMIM 232220		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
SLC39A13	EHLERS-DANLOS SYNDROME-LIKE SPONDYLOCHEIRODYSPLASIA 612350;SPONDYLOEPIMETAPHYSEAL DYSPLASIA WITH ABNORMAL DENTITION 601668		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC46A1	HEREDITARY FOLATE MALABSORPTION 229050		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC4A1	RENAL TUBULAR ACIDOSIS, DISTAL, AD 179800;RENAL TUBULAR ACIDOSIS, DISTAL, AR 611590		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
SLC4A11	CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 4 613268		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC4A4	PROXIMAL RENAL TUBULAR ACIDOSIS WITH OCULAR ABNORMALITIES 604278		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC52A3	BROWN-VIALETTO-VAN LAERE SYNDROME 211530		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC5A5	THYROID HORMONOGENESIS DEFECT I 274400		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC5A7	Congenital Myasthenic Syndrome with Episodic Apnea		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC6A1	EPILEPSY WITH MYOCLONIC-ATONIC SEIZURES		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SLC6A3	PARKINSONISM-DYSTONIA, INFANTILE 319029		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC6A5	HYPEREKPLEXIA 149400		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SLC6A8	X-LINKED CREATINE DEFICIENCY SYNDROME 300352		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
SLC9A6	MENTAL RETARDATION SYNDROMIC X-LINKED CHRISTIANSON TYPE 300243		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
SLX4	FANCONI ANEMIA COMPLEMENTATION GROUP P 613951		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SMAD3	SMAD3-RELATED LOEYS-DIETZ SYNDROME 319643		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, OMIM 175050;MYHRE SYNDROME, OMIM 139210		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SMARCA2	COFFIN SIRIS 1 135900;NICOLAIDES-BARATSER SYNDROME 601358		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SMARCA4	COFFIN SIRIS 135900;RHABDOID TUMOR PREDISPOSITION SYNDROME 2 613325		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SMARCAL1	SCHIMKE IMMUNOOSSEOUS DYSPLASIA 242900		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SMARCB1	EHMT1-like SYNDROME;RHABDOID PREDISPOSITION SYNDROME 1 609322		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SMARCE1	COFFIN SIRIS 135900		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SMC1A	CORNELIA DE LANGE SYNDROME TYPE 2 300590;EPILEPTIC ENCEPHALOPATHY		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
SMC3	CORNELIA DE LANGE SYNDROME TYPE 3 610759		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SMOC1	ANOPHTHALMOCRANIOMELIC SYNDROME 206920		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SMOC2	DENTIN DYSPLASIA, TYPE I, WITH MICRODONTIA AND MISSHAPEN TEETH 125400		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SMPD1	NIEMANN-PICK DISEASE TYPE B 607616;NIEMANN-PICK DISEASE TYPE A 257200		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SMS	SNYDER-ROBINSON SYNDROME 309583		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
SNAP29	CEDNIK SYNDROME 609528		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SNAPC4	SNAPC4-related neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, OMIM 620515		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SOS1	NOONAN SYNDROME 4 610733		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SOX10	WAARDENBURG SYNDROME TYPE 2E 611584;KALLMANN SYNDROME WITH DEAFNESS;WAARDENBURG SYNDROME TYPE 4C 613266;YEMENITE DEAF-BLIND HYPOPIGMENTATION SYNDROME 601706;PERIPHERAL DEMYELINATING NEUROPATHY, CENTRAL DYSMYELINATING LEUKODYSTROPHY, WAARDENBURG SYNDROME, AND HIRSCHSPRUNG DISE		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SOX11	MENSOURETERAL REFLUX TYPE 3 613674		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SOX2	AEG SYNDROME;MICROPHTHALMIA SYNDROMIC TYPE 3 206900		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency, OMIM 300123 Intellectual disability, X-linked, with panhypopituitarism, MONDO:0010252:Panhypopituitarism, X-linked, OMIM:312000:Panhypopituitarism, X-linked, MONDO:0010712		X-LINKED: hemizygous mutation in males, biallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
SOX5	12P12.5 INTRAGENIC DELETIONS ASSOCIATED WITH INTELLECTUAL DISABILITY		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SOX6	SOX6-related neurodevelopmental syndrome		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SOX9	PIERRE ROBIN SEQUENCE;CAMPOMELIC DYSPLASIA 114290		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SPAST	SPAST-related developmental disorder (monoallelic)		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SPECC1L	FACIAL CLEFTING, OBLIQUE, 1 609251		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SPG11	SPASTIC PARAPLEGIA-11 604360		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SPR	DOPA-RESPONSIVE DYSTONIA DUE TO SEPIAPTERIN REDUCTASE DEFICIENCY 612716		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SPRED1	LEGIUS SYNDROME 611431		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SPTAN1	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, TYPE 5 613477		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SPTBN2	SCA14 infantile ataxia with oculomotor and pyramidal signs		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
SPTLC2	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE IC, OMIM 613640		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SRCAP	FLOATING-HARBOR SYNDROME 136140		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SRD5A3	CONGENITAL DISORDERS OF GLYCOSYLATION 612379		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SRY	46XY SEX REVERSAL 1 400044		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
ST14	ICHTHYOSIS AUTOSOMAL RECESSIVE WITH H/PTOTRICHOSIS 610765		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
ST3GAL3	Developmental and epileptic encephalopathy, 15, OMIM:615006;developmental and epileptic encephalopathy, 15, MONDO:0014003:Intellectual developmental disorder, autosomal recessive 12, OMIM:611090:Intellectual disability, autosomal recessive 12, MONDO:0012612		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ST3GAL5	AMISH INFANTILE EPILEPSY SYNDROME 319543		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
STAR	CHOLESTEROL DESMOLASE-DEFICIENT CONGENITAL ADRENAL HYPERPLASIA 201710		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
STATSB	GROWTH HORMONE INSENSITIVITY WITH IMMUNODEFICIENCY 245590		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
STIL	MICROCEPHALY PRIMARY TYPE 7 612703		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
STRA6	MICROPHTHALMIA SYNDROMIC TYPE 9 601186		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, OMIM:611087		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
STS	ICHTHYOSIS, X-LINKED 308100		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
STX1A	STX1A-associated neurodevelopmental disorder with epilepsy;STX1A-associated neurodevelopmental disorder without epilepsy		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
STXBP1	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, TYPE 4 612164;ANGELMAN/PITT-HOPKINS SYNDROME-LIKE DISORDER		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SUCLG1	FATAL INFANTILE LACTIC ACIDOSIS 308078		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SUFU	Joubert Syndrome with Cranio-facial and Skeletal Defects;SUFU-related Joubert and congenital ocular motor apraxia		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
SUMF1	SULFATIDOSIS, JUVENILE; AUSTIN TYPE 272200		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SUOX	Sulfite oxidase deficiency, OMIM:272300		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SURF1	LEIGH SYNDROME 256000;COMPLEX IV DEFICIENCY 220110		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SYN1	EPILEPSY, X-LINKED, WITH VARIABLE LEARNING DISABILITIES AND BEHAVIOR DISORDERS 290027		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
SYNE1	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 8, OMIM 610743		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
SYNGAP1	MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 5 612621;EPILEPTIC ENCEPHALOPATHY		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SYP	MENTAL RETARDATION X-LINKED SYP-RELATED 300802		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
SYT2	SYT2-related congenital onset presynaptic myasthenic syndrome		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
TAB2	CONGENITAL HEART DISEASE, NONSYNDROMIC 2 612863		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
TAC3	HYPOGONADOTROPIC HYPOGONADISM 146110		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
TACO1	LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX IV DEFICIENCY 256000		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
TACR3	HYPOGONADOTROPIC HYPOGONADISM 146110		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
TAF1	Dysmorphic Features, Intellectual Disability, and Neurological Manifestations		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
TAT	TYROSINEMIA TYPE 2 276600		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
TAZ	BARTH SYNDROME 302060		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
TBC1D23	Non-degenerative Pontocerebellar Hypoplasia		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
TBC1D24	DOORS SYNDROME 220500;MYOCLONIC EPILEPSY, INFANTILE, FAMILIAL, 605021;NON SYNDROMAL HEARING LOSS 614617		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
TBCE	Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy;HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME, OMIM:241410		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
TBL1XR1	Pierpont syndrome;Intellectual disability with autism spectrum disorder		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
TBX1	22Q11.2 DELETION SYNDROME 188400		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
TBX15	Craniofacial Dysmorphism, Hypoplasia of Scapula and Pelvis, and Short Stature;Cousin Syndrome		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
TBX20	ATRIAL SEPTAL DEFECT TYPE 4 611363		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
TBX22	CLEFT PALATE, X-LINKED 304401		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
TBX3	ULNAR-MAMMARY SYNDROME 181450		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
TBX4	SMALL PATELLA SYNDROME 147891		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
TBX5	HOLT-ORAM SYNDROME 142900		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
TBXAS1	GHOSAL HEMATODIAPHYSEAL SYNDROME 231095		BIALLELIC; autosomal or pseudoautosomal	DDG2P			5.1
TCF4	PITT-HOPKINS SYNDROME 610954		MONOALLELIC; autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1

TCF7L2	TCF7L2-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TCN2	Transcobalamin II deficiency		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TCOF1	TREACHER COLLINS SYNDROME TYPE 1 154500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TCTN1	JOUBERT SYNDROME AND RELATED DISORDERS 194263		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TCTN2	JOUBERT SYNDROME AND RELATED DISORDERS 194263		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TDRD7	CATARACT CONGENITAL AUTOSOMAL RECESSIVE TYPE 4 613887		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TEK	VENOUS MALFORMATIONS, MULTIPLE CUTANEOUS AND MUCOSAL 600195		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TERC	Dyskeratosis congenita, autosomal dominant 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TERT	Dyskeratosis congenita, autosomal recessive 4		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TFAP2A	BRANCHIOOCULOFACIAL SYNDROME 113620		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TFAP2B	CHAR SYNDROME 169100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TGFB1	Camurati-Engelmann disease, OMIM:131300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TGFB2	LOEYS-DIETZ SYNDROME, TYPE 4, OMIM:614816		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TGFB3	LOEYS-DIETZ SYNDROME 610168		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TGFBR1	LOEYS-DIETZ SYNDROME TYPE 2A 608967;LOEYS-DIETZ SYNDROME TYPE 1A 609192;AORTIC ANEURYSM FAMILIAL THORACIC TYPE 5 609192		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TGFBR2	LOEYS-DIETZ SYNDROME;TGFBR2-RELATED LOEYS-DIETZ SYNDROME 249163		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TGIF1	HOLOPROSENCEPHALY 609637		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TH	DOPA-RESPONSIVE DYSTONIA 605407		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
THAP1	DYSTONIA 6, TORSION 602629		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
THRA	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 8 614450		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TIMM8A	MOHR-TRANEBJAERG SYNDROME 304700;JENSEN SYNDROME 311150		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
TINF2	DEGENERATIVE RETINOPATHY WITH BONE MARROW FAILURE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TK2	MITOCHONDRIAL DNA DEPLETION SYNDROME, MYOPATHIC FORM 258276		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TMCO1	CRANIOFACIAL DYSMORPHISM, SKELETAL ANOMALIES, AND MENTAL RETARDATION SYNDROME 213980		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TMEM165	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IIK 614727		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TMEM216	JOUBERT SYNDROME 2 608091		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TMEM237	JOUBERT SYNDROME 14 614424		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TMEM67	NEPHRONOPHTHISIS,TYPE 11 613550;MECKEL SYNDROME TYPE 3 607361;COACH SYNDROM 216360;JOUBERT SYNDROME TYPE 6 610688		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TMEM70	MITOCHONDRIAL COMPLEX V (ATP SYNTHASE) DEFICIENCY, NUCLEAR TYPE 2 614052		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TMPRSS6	IRON-REFRACTORY IRON DEFICIENCY ANEMIA 206200		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TNFRSF13B	IMMUNODEFICIENCY, COMMON VARIABLE, 2 240500		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TNNT1	TNNT1-associated congenital myopathy (biallelic)		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TOR1A	TOR1A-associated arthrogryposis multiplex congenita (AR)		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TP53BK	GALLOWAY-MOWAT SYNDROME 4, 617730		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TP63	ECTODERMAL DYSPLASIA RAPP-HODGKIN TYPE 129400;NON-SYNDROMIC OROFACIAL CLEFT TYPE 8 129400;SPLIT-HANDFOOT MALFORMATION TYPE 4 605288;ANKYLOBLEPHARON-ECTODERMAL DEFECTS-CLEFT LIP/PALATE 106260;ECTRODACTYLY-ECTODERMAL DYSPLASIA-CLEFT LIP/PALATE SYNDROME TYPE 3 604292;LIMB-MAMMARY SYNDROME	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TP73	TP73-related ciliary dyskinesia and bronchiectasis, OMIM:619466		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TPM2	ARTHROGRYPOSIS, DISTAL, TYPE 1 108120		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TPM3	Nemaline/Cap myopathy		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TPP1	NEURONAL CEROID LIPOFUSCINOSIS TYPE 2 204500		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TPP2	Autoimmunity,Developmental delay,immunodeficiency,TPP2-related immune deficiency, autoimmune disease and intellectual disability		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TRAPPC2	SPONDYLOEPIPHYSEAL DYSPLASIA TARDA 313400		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
TRAPPC9	MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 13 613192		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TREX1	AICARDI-GOUTIERES SYNDROME 1, DOMINANT AND RECESSIVE 225750		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TRIM32	BARDET-BIEDL SYNDROME TYPE 11 209900;LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2H 254110		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TRIM37	MULIBREY NANISM 213300		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TRIO	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TRIP11	ACHONDROGENESIS TYPE 1A 200600		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TRIP12	TRIP12-related intellectual disability with/without autism spectrum disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TRPM1	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1C 613216		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TRPM3	TRPM3-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TRPS1	TRICHO-RHINO-PHALANGEAL SYNDROME TYPE 1 190350		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TRPV3	OLMSTED SYNDROME 614594		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TRPV4	METATROPIC DYSPLASIA 156530;SPONDYLOMETAPHYSEAL DYSPLASIA, KOZLOWSKI TYPE 184252		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TRRAP	Autism and Syndromic Intellectual Disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TSC1	TUBEROUS SCLEROSIS TYPE 1 191100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TSC2	TUBEROUS SCLEROSIS TYPE 2 613254;LYMPHANGIOLEIOMYOMATOSIS 606690		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TSEN2	PONTOCEREBELLAR HYPOPLASIA TYPE 2 AND TYPE 4 316970		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TSEN34	PONTOCEREBELLAR HYPOPLASIA TYPE 2 AND TYPE 4 316970		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TSEN54	PONTOCEREBELLAR HYPOPLASIA TYPE 2 AND TYPE 4 316970		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TSHB	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS 4 275100		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TSHR	SEVERE HYPERTHYROIDISM, FAMILIAL GESTATIONAL, 603273;HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 1 275200	BOTH monoallelic and biallelic, autosomal or pseudoautosomal		DDG2P		5.1
TSPAN7	MENTAL RETARDATION X-LINKED TYPE 58 300210		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
TTC19	MITOCHONDRIAL COMPLEX III DEFICIENCY 124000		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TTC37	TRICHOHEPATOENTERIC SYNDROME 222470		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TTC8	BARDET-BIEDL SYNDROME TYPE 8 209900;RETINITIS PIGMENTOSA TYPE 51 613464		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TTI2	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TTN	CAUSE OF EARLY-ONSET MYOPATHY WITH FATAL CARDIOMYOPATHY 611705		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TUBA1A	LISSENCEPHALY TYPE 3 611603;INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TUBB2B	POLYMICROGYRIA ASYMMETRIC 610031		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TUBB3	CONGENITAL FIBROSIS OF THE EXTRAOCULAR MUSCLES 600638;CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFORMATIONS 1 614039		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TUBGCP4	AUTOSOMAL-RECESSIVE MICROCEPHALY WITH CHORIORETINOPATHY		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TUBGCP6	MICROCEPHALY AND CHORIORETINOPATHY WITH OR WITHOUT MENTAL RETARDATION 251270		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TUFM	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 4 610678		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TUSC3	MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 7 611093		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TWIST1	CRANIOSYNOSTOSIS, TYPE 1 123100;SAETHRE-CHOTZEN SYNDROME 101400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
TWIST2	SETLEIS SYNDROME, OMIM:227260;ABLEPHARON MACROSTOMIA SYNDROME; OMIM:200110	BOTH monoallelic and biallelic, autosomal or pseudoautosomal		DDG2P		5.1
TYR	OCULOCUTANEOUS ALBINISM TYPE 1 203100		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
TYRP1	OCULOCUTANEOUS ALBINISM TYPE 3 203290		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
UBE2A	UBE2A-RELATED X-LINKED SYNDROMIC MENTAL RETARDATION 313562		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
UBE3A	ANGELMAN SYNDROME 105830		MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)	DDG2P		5.1
UBR1	JOHANSON-BLIZZARD SYNDROME 243800		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
UBR7	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
UGT1A1	CRIGLER-NAJJAR SYNDROME, TYPE I 218800		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
UMPS	OROTIC ACIDURIA TYPE 1 258900		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
UNC80	Persistent Hypotonia, Encephalopathy, Growth Retardation, and Severe Intellectual Disability		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
UPF3B	MENTAL RETARDATION SYNDROMIC X-LINKED TYPE 14 300676		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
UQCC2	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX III DEFICIENCY, UQCC2-RELATED 257827		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
UQCRQ	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX III DEFICIENCY, UQCRQ RELATED 319211		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
UROC1	UROCANASE DEFICIENCY 276880		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
UROS	CONGENITAL ERYTHROPOIETIC PORPHYRIA 263700		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
USB1	Poikiloderma with neutropenia		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
USP7	USP7-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
USP9X	MENTAL RETARDATION, X-LINKED 99 300919		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P		5.1
UVSSA	UV-SENSITIVE SYNDROME 609413		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
VDR	RICKETS, VITAMIN D-DEPENDENT, TYPE 2A 277440		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
VIPAS39	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS 2 613404		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
VLDLR	CEREBELLAR ATAXIA MENTAL RETARDATION AND DYSEQUILIBRIUM SYNDROME TYPE 1 224050		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
VPS13B	COHEN SYNDROME 216550		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
VPS33B	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS 1 208085		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
VRK1	PONTOCEREBELLAR HYPOPLASIA TYPE 1 607596		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
VSX2	MICROPHTHALMIA ISOLATED TYPE 2 610093;MICROPHTHALMIA ISOLATED WITH COLOBOMA TYPE 3 610092;MICROPHTHALMIA WITH CATARACTS AND IRIS ABNORMALITIES 610092		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
WDFY3	Primary Microcephaly or macrocephaly with developmental delay		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
WDPCP	BARDET-BIEDL SYNDROME TYPE 15 209900		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
WDR11	KALLMANN SYNDROME		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
WDR19	CRANIOECTODERMAL DYSPLASIA 4 614378;ASPHYXIATING THORACIC DYSTROPHY 5 614376		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
WDR35	CRANIOECTODERMAL DYSPLASIA 2 613610;SHORT RIB-POLYDACTYLY SYNDROME, TYPE V 614091		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
WDR45	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P		5.1
WDR4S8	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
WDR62	MICROCEPHALY, CORTICAL MALFORMATIONS AND MENTAL RETARDATION 604317		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
WFS1	Wolfram-like syndrome, autosomal dominant, OMIM:614296;WOLFRAM SYNDROME 1, OMIM:222300	BOTH monoallelic and biallelic, autosomal or pseudoautosomal		DDG2P		5.1
WNT10B	Split-hand/foot malformation 6, OMIM:225300	BIALLELIC, autosomal or pseudoautosomal		DDG2P		5.1
WNT3	TETRA-AMELIA SYNDROME 276060	BIALLELIC, autosomal or pseudoautosomal		DDG2P		5.1
WNT4	MULLERIAN APLASIA AND HYPERANDROGENISM 158330;SERKAL SYNDROME 611812	BOTH monoallelic and biallelic, autosomal or pseudoautosomal		DDG2P		5.1
WNT5A	WNT5A-RELATED ROBINOW SYNDROME, AUTOSOMAL DOMINANT 180700		BIALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
WNT7A	FUHRMANN SYNDROME 228930;LIMB/PELVIS-HYPOPLASIA/APLASIA SYNDROME 276820		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
WRAP53	DYSKERATOSIS CONGENITA, AUTOSOMAL RECESSIVE 3 613988		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
WT1	DENYS-DRASH SYNDROME 194080;FRASIER SYNDROME FRASIER SYNDROME 136680		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
WWOX	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12 614322;EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 28 616211		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
XPA	XERODERMA PIGMENTOSUM, GROUP A 278700		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
XPC	XERODERMA PIGMENTOSUM, GROUP C 278720		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
XRCC4	PRIMORDIAL DWARFISM 615272		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
XYLT1	DESBUQUOIS DYSPLASIA 2, OMIM:615777;Baratela Scott Syndrome, OMIM:615777		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
XYLT2	SPONDYLOCOSTAL DYSOSTOSIS 4 605822		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
YAP1	COLOBOMA, OCULAR, WITH OR WITHOUT HEARING IMPAIRMENT, CLEFT LIP/PALATE, AND/OR MENTAL RETARDATION 120433		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
YARS2	MYOPATHY, LACTIC ACIDOSIS, AND SIDEROBLASTIC ANEMIA 2, OMIM:613561		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
YWHAE	YWHAE-related developmental delay, seizures, hypotonia and brain abnormalities		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
YY1	INTELLECTUAL DISABILITY, OMIM:616579		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ZBTB18	ZBTB18 syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type, OMIM:300799		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
ZEB2	MOWAT-WILSON SYNDROME 235730		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ZFHX3	ZFHX3-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ZFHX4	ZFHX4-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ZFP57	DIABETES MELLITUS, 6Q24-RELATED TRANSIENT NEONATAL 601410		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ZFYVE26	SPASTIC PARAPLEGIA 15, AUTOSOMAL RECESSIVE TYPE 15 270700		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ZIC1	CRANIOSYNOSTOSIS 6 616602		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ZIC2	HOLOPROSENCEPHALY 609637		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ZIC3	VACTERL ASSOCIATION, X-LINKED, WITH OR WITHOUT HYDROCEPHALUS 314983;HETEROTAXY SYNDROME 207574		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
ZMPSTE24	MANDIBULOACRAL DYSPLASIA WITH TYPE B LIPODYSTROPHY 608612;LETHAL RESTRICTIVE DERMOPATHY, ZMPSTE24-RELATED 319186		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ZMYND11	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ZNF711	MENTAL RETARDATION X-LINKED ZNF711-RELATED 300803		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
ZNF750	SEBORRHEA-LIKE DERMATITIS WITH PSORIASIFORM ELEMENTS 610227		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ABCB6	MICROPHTHALMIA, ISOLATED, WITH COLOBOMA 7 614497		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ACBD5	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ACVR2B	HETEROTAXY SYNDROME 207574		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ADARB1	ADARB1-associated Microcephaly, Intellectual Disability, and Seizures		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ADK	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ADRA2B	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
AFG3L2	SPINOCEREBELLAR ATAXIA 28 610246;ATAXIA, SPASTIC, 5, AUTOSOMAL RECESSIVE 614487		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ALAD	ACUTE HEPATIC PORPHYRIA 612740		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ANK2	ANK2-related neurodevelopmental disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
ARHGEF6	MENTAL RETARDATION X-LINKED TYPE 46 300436		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
ARL14EP	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ASCC1	Prenatal Spinal Muscular Atrophy and Congenital Bone Fractures		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ASCC3	Intellectual developmental disorder, autosomal recessive 81, OMIM:620700		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ASCL1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ATOH7	RETINAL NON-ATTACHMENT CONGENITAL NON-SYNDROMIC 221900		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
ATP6AP2	MENTAL RETARDATION X-LINKED WITH EPILEPSY 300423		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
ATP8A2	CEREBELLAR ATAXIA, MENTAL RETARDATION, AND DYSEQUILIBRIUM SYNDROME 4 615268		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
AXIN1	CAUDAL DUPLICATION ANOMALY, OMIM:607864		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
B3GAT3	MULTIPLE JOINT DISLOCATIONS, SHORT STATURE, AND CRANIOFACIAL DYSMORPHISM WITH OR WITHOUT CONGENITAL HEART DEFECTS 245600		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
BCORL1	Shukla-Vernon Syndrome		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
CACNA1H	EPILEPSY, CHILDHOOD ABSENCE, SUSCEPTIBILITY TO, 6 611942		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
CACNA2D1	CACNA2D1-related neurodevelopmental disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
CACNB4	JUVENILE MYOCLONIC EPILEPSY, OMIM:611136		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
CAPN10	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
CAV1	CONGENITAL GENERALIZED LIPODYSTROPHY TYPE 3 612526		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
CCNA2	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
CC96	C-SYNDROME 212750		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
CDC42BPB	CDC42BPB-related Neurodevelopmental Disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
CDH15	MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 5 612580		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
CHRM1	CHRM1-associated intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
CHRNA2	CHRNA2-RELATED NOCTURNAL FRONTAL LOBE EPILEPSY, AUTOSOMAL DOMINANT, OMIM:291607		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P		5.1
CLCNKA	Bartter syndrome, type 4b, digenic, OMIM:613090;Bartter disease type 4B, MONDO:0009099		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
CLIC2	Mental retardation, X-linked, syndromic 32 300856		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P		5.1
CNKSR1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1
COA5	MITOCHONDRIAL COMPLEX IV DEFICIENCY 220110		BIALLELIC, autosomal or pseudoautosomal	DDG2P		5.1

COMP	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 1, OMIM:132400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
COQ5	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
COX14	MITOCHONDRIAL COMPLEX IV DEFICIENCY 220110		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
CRADD	INTELLECTUAL DEVELOPMENTAL DISORDER, autosomal recessive 34, with variant lissencephaly, OMIM:614499		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
CRBN	MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 2A 607417		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
CRKL	Bladder exstrophy plus		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CRYAB	MYOPATHY, MYOFIBRILLAR, FATAL INFANTILE HYPERTONIC, ALPHA-B CRYSTALLIN-RELATED 613869;CATARACT POSTERIOR POLAR TYPE 2 613763		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
CRYBA4	MICROPHTHALMIA ISOLATED WITH CATARACT TYPE 4, OMIM:610426		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CRYGD	Cataract 2, multiple types, OMIM:115700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CSDE1	CSDE1-associated intellectual disability and autism		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
CYP24A1	Hereditary 1,25-dihydroxyvitamin D-resistant rickets		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
D2HGDH	D-2-HYDROXYGLUTARIC ACIDURIA 1 600721		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DCAF17	WOODHOUSE-SAKATI SYNDROME 241080		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DDX58	SINGLETON-MERTEN SYNDROME 182250		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
DGAT1	CONGENITAL DIARRHEAL DISORDERS		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DHX16	Intellectual Disability, Central Nervous System anomalies and Seizures		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
DHX37	Intellectual Disability and Central Nervous System anomalies		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DIP2B	MENTAL RETARDATION, FRA12A TYPE 136630		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
DISP1	HOLOPROSENCEPHALY, OMIM:609637		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
DLG5	DLG5-associated developmental disorder (biallelic);DLG5-associated developmental disorder (monoallelic)		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
DLL1	INTELLECTUAL DISABILITY 616579		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
DLX5	SPLIT HAND AND FOOT MALFORMATION 220600		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
DMPK	DYSTROPHIA MYOTONICA TYPE 1, OMIM:160900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
EEF1B2	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ELP2	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EMX2	FAMILIAL SCHIZENCEPHALY, EMX2-RELATED 269160		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ENTPD1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EOMES	POLYMICROGYRIA AND CORPUS CALLOSUM AGENESIS		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
EPB41L1	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ERLIN2	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FASN	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FBLN1	SYNPOLYDACTYLY, 3/3-PRIME/4, ASSOCIATED WITH METACARPAL AND METATARSAL SYNOSTOSES 608180		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FBXW4	SPLIT-HAND/FOOT MALFORMATION TYPE 3 246560		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
FDFT1	Defect in Cholesterol Biosynthesis		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FLG	ICHTHYOSIS VULGARIS 146700		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FRMD7	NYSTAGMUS 1, CONGENITAL, X-LINKED 310700		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
FRY	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
FTO	GROWTH RETARDATION DEVELOPMENTAL DELAY COARSE FACIES AND EARLY DEATH 612938		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GABRG1	GABRG1-associated epileptic encephalopathy		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GAD1	CEREBRAL PALSY SPASTIC QUADRIPLEGIC TYPE 1 603513		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GDF3	MICROPHTHALMIA ISOLATED TYPE 7 613704;KLIPPEL-FEIL SYNDROME TYPE 3 613702;MICROPHTHALMIA ISOLATED WITH COLOBOMA TYPE 6 613703		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
GDI1	MENTAL RETARDATION X-LINKED TYPE 48 300849;MENTAL RETARDATION X-LINKED TYPE 41 300849		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
GFER	MITOCHONDRIAL PROGRESSIVE MYOPATHY WITH CONGENITAL CATARACT HEARING LOSS AND DEVELOPMENTAL DELAY (MPMCHD 613076		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GNB3	GNB3 Autosomal-Recessive Congenital Stationary Night Blindness.		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GNE	GNE-associated congenital myopathy;GNE-associated sialuria, OMIM:269921		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
GON4L	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GPHN	GPHN-related molybdenum cofactor deficiency, OMIM:615501		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
GTF2IRD1	GTF2IRD1-related neurodevelopmental disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HARS	USHER SYNDROME 614504		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HIST3H3	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
HOXA11	Radioulnar Synostosis with Amegakaryocytic Thrombocytopenia		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
HTRA2	Early-onset mitochondrial syndrome associated with 3-methylglutaconic aciduria		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
IGBP1	AGENESIS OF THE CORPUS CALLOSUM WITH MENTAL RETARDATION-OCULAR COLOBOMA-MICROGNATHIA 300472		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
INPP4A	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ITGA6	EPIDERMOLYSIS BULLOSA WITH PYLORIC ATRESIA 226730		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
JMJD1C	JMJD1C-related neurodevelopmental disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KANK1	CEREBRAL PALSY SPASTIC QUADRIPLEGIC TYPE 2 612900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KCNA1	KCNA1-related epileptic encephalopathy, biallelic;KCNA1-related epileptic encephalopathy, monoallelic		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
KCNK9	BIRK-BAREL SYNDROME, OMIM:612292		MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)	DDG2P			5.1
KDM5A	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
KIRREL3	MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 4 612581		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
KITLG	WAARDENBURG SYNDROME TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
LDB3	MYOPATHY MYOFIBRILLAR TYPE 4, OMIM:609452		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
LEFTY2	HETEROTAXY SYNDROME 207574		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
LRP6	Tooth Agenesis		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
LRPAP1	MYOPIA 23, AUTOSOMAL RECESSIVE 615431;EXTREME MYOPIA		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MAGI2	EARLY ONSET EPILEPTIC ENCEPHALOPATHY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MAGT1	MENTAL RETARDATION X-LINKED TYPE 95 300716		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
MAPK10	EPILEPTIC ENCEPHALOPATHY LENNOX-GASTAUT TYPE 606369		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MED23	MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 18 614249		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MIR184	EDICT SYNDROME 614303;KERATOCONUS WITH CATARACT		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MMP14	WINCHESTER SYNDROME		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MPDZ	HYDROCEPHALUS, NONSYNDROMIC, AUTOSOMAL RECESSIVE 2 615219		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MYH11	Megacystis-microcolon-intestinal hypoperistalsis syndrome		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
MYH6	CARDIOMYOPATHY DILATED TYPE 1EE 613252;CARDIOMYOPATHY FAMILIAL HYPERTROPHIC TYPE 14 613251;ATRIAL SEPTAL DEFECT TYPE 3 614089		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MYOC	PRIMARY CONGENITAL GLAUCOMA TYPE 3A 231300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
MYT1	OAVS/Goldenhar syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NCAPD2	Microcephaly with short stature		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NCKAP1	NCKAP1-related Neurodevelopmental Disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NDUFA9	LEIGH SYNDROME 256000		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NOP10	DYSKERATOSIS CONGENITA, AUTOSOMAL RECESSIVE 1 224230;NOP10-RELATED DYSKERATOSIS CONGENITA 318811		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
NOTCH3	INFANTILE MYOFIBROMATOSIS 615293		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NR1I3	EHMT1-LIKE INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
NRXN3	AUTISM 209850		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PARP1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PECR	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PIGM	Glycosylphosphatidylinositol deficiency		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PLCG2	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, OMIM:614878;Familial cold autoinflammatory syndrome 3, OMIM:614468		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
PLOD3	LYSYL HYDROXYLASE 3 DEFICIENCY 612394		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
POMP	KERATOSIS LINEARIS WITH ICHTHYOSIS CONGENITA AND SCLEROSING KERATODERMA 601952		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
PTPRF	ATHELIA		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
QKI	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RABL6	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RAD50	NIJMEGEN BREAKAGE SYNDROME-LIKE DISORDER 613078		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RAD51	MIRROR MOVEMENTS 2 614508		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RALGDS	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RANBP2	ACUTE NECROTIZING ENCEPHALOPATHY 1, SUSCEPTIBILITY TO 285648		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RASA1	PARKES WEBER SYNDROME 608355;CAPILLARY MALFORMATION-ARTERIOVENOUS MALFORMATION 608354		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RBFOX1	RBFOX1-related neurodevelopmental disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RBM28	ALOPECIA NEUROLOGIC DEFECTS AND ENDOCRINOPATHY SYNDROME 612079		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RGS7	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RMI1	Bloom Syndrome like Disorder		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RNF113A	X-LINKED TRICHOTHIODYSTROPHY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
RNF135	MACROCEPHALY, MACROSOMIA, FACIAL DYSMORPHISM SYNDROME 614192		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RNF168	RIDDLE SYNDROME 611943		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
RRM1	RRM1-related mitochondrial DNA depletion/deletions syndrome		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	DDG2P			5.1
RYR2	RYR2-related Catecholaminergic polymorphic ventricular tachycardia and intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
RYR3	EPILEPTIC ENCEPHALOPATHY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SCRIB	8Q24.3 DELETION-LIKE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SEC23A	CRANIOLENTICULOSUTURAL DYSPLASIA 607812		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
SH3BP2	Cherubism, OMIM:118400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SIX6	MICROPHTHALMIA, ISOLATED, WITH CATARACT 2 212550		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SLC24A1	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1D 613830		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
SLC9A9	SUSCEPTIBILITY TO AUTISM TYPE 16 613410		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SMAD2	CONGENITAL HEART DISEASE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SMAD6	Non-syndromic craniosynostosis		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SNIP1	SYMPTOMATIC EPILEPSY AND SKULL DYSPLASIA 614501		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
SNX3	MICROPHTHALMIA SYNDROMIC TYPE 8 601349		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SOBP	MENTAL RETARDATION-ANTERIOR MAXILLARY PROTRUSION-STRABISMUS 613671		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
SRGAP3	SLIT-ROBO RHO GTPASE-ACTIVATING PROTEIN 3 606525		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SRPX2	BILATERAL PERISYLVIAN POLYMICROGYRIA 300388;ROLANDIC EPILEPSY WITH SPEECH DYSPRAXIA AND MENTAL RETARDATION X-LINKED 300643		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
STIM1	TUBULAR-AGGREGATE MYOPATHY 160565		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SUMO1	CLEFT LIP +/- CLEFT PALATE 608874		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
SUPT16H	SUPT16H-related neurodevelopmental disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
TAF2	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
TFRC	Combined immunodeficiency		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
TLL1	ATRIAL SEPTAL DEFECT TYPE 6 613087		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
TMEM114	CONGENITAL AND JUVENILE CATARACT 611579		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
TMEM135	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
TRAPPC10	TRAPPC10-associated intellectual disability		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
TRMT1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
TRPC5	TRPC5-related neurodevelopmental disorder		X-LINKED: hemizygous mutation in males, biallelic mutations in females	DDG2P			5.1
TSHZ1	AURAL ATRESIA, CONGENITAL 607842		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
TUBA8	POLYMICROGYRIA WITH OPTIC NERVE HYPOPLASIA 613180		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
UBE3C	UBE3C-related neurodevelopmental disorder with absent speech and movement and behavioural abnormalities		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
VANGL1	NEURAL TUBE DEFECTS 182940		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
VCP	VCP-related developmental disorder (monoallelic)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
VIP	ASPERGER		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
WDR4	GALLOWAY-MOWAT SYNDROME 6, 618347		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
WDR81	CEREBELLAR ATAXIA, MENTAL RETARDATION, AND DYSEQUILIBRIUM SYNDROME 2 610185		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
XPNPEP3	Nephronophthisis-like nephropathy 1 OMIM:613159;nephronophthisis-like nephropathy 1 MONDO:0013163		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ZBTB16	SKELETAL DEFECTS GENITAL HYPOPLASIA AND MENTAL RETARDATION 612447		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ZBTB40	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ZCCHC8	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
ZDHHC15	MENTAL RETARDATION X-LINKED TYPE 91 300577		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	DDG2P			5.1
ZEB1	CORNEAL DYSTROPHY FUCHS ENDOTHELIAL TYPE 6 613270;POSTERIOR POLYMORPHOUS CORNEAL DYSTROPHY TYPE 3 609141		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ZFPM2	TETRALOGY OF FALLOT 187500;DIAPHRAGMATIC HERNIA 3 610187;46,XY GONADAL DYSGENESIS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	DDG2P			5.1
ZNF526	Dentici-Novelli neurodevelopmental syndrome, OMIM:619877		BIALLELIC, autosomal or pseudoautosomal	DDG2P			5.1
SLC4A10	SLC4A10-related neurodevelopmental disorder with hypotonia and characteristic brain abnormalities	620746	biallelic_autosomal				
DIP2C	DIP2C-related developmental disorder with speech delay	No disease mim	monoallelic_autosomal				
GABRD	GABRD-related neurodevelopmental disorder with epilepsy	No disease mim	monoallelic_autosomal				
TCP1	TCP1-related neurodevelopmental disorder with polymicrogyria	No disease mim	monoallelic_autosomal				
CCT5	CCT5-related neurodevelopmental disorder with brain abnormalities	No disease mim	monoallelic_autosomal				
ABCC9	Hypertrichotic osteochondrodysplasia_23985 (Cantu syndrome);Hypertrichotic osteochondrodysplasia239850		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ACAN	Osteochondritis dissecans, short stature, and early-onset osteoarthritis 165800;Spondyloepimetaphyseal dysplasia, aggrecan type 61283;Spondyloepiphyseal dysplasia, Kimberley type 608361		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ACP5	Spondyloenchondrodysplasia with immune dysregulation 607944		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ACVR1	Fibrodysplasia ossificans progressiva 135100		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ADAMTS10	Weill-Marchesani syndrome 1, recessive, 277600		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ADAMTS17	Weill-Marchesani syndrome type 4		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ADAMTSL2	Geleophysic dysplasia 1 231050		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
AFF3	KINSSHIP syndrome, OMIM:619297		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
AGA	Aspartylglucosaminuria 208400 (Patients may be tall for their age, but lack of a growth spurt in puberty typically causes adults to be short)		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
AGPS	Foundation Trust)Rhizomelic chondrodysplasia punctata, type 3 600121;Rhizomelic chondrodysplasia punctata, type 3 600121		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ALG12	Congenital disorder of glycosylation, type Ig 607143		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ALG3	Congenital disorder of glycosylation, type Id 601110		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ALG9	Gillessen-Kaesbach-Nishimura syndrome 263210;Congenital disorder of glycosylation, type II 608776;Gillessen-Kaesbach-Nishimura syndrome263210		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ALPL	hypophosphatasia; skeletal dysplasias;skeletal dysplasias;Osteogenesis Imperfecta and Decreased Bone Density		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ALX1	Frontonasal dysplasia type 3 613456;Frontonasal dysplasia 3 613456		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ALX3	Frontonasal dysplasia 1 136760 (frontorhiny);Frontonasal dysplasia 1 136760		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ALX4	Frontonasal dysplasia 2 613451		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
AMER1	Osteopathia striata with cranial sclerosis300373;Osteopathia striata with cranial sclerosis 300373		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ANAPC1	Rothmund Thomson syndrome type 1, OMIM:618625, MONDO:0016368		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ANKH	Chondrocalcinosis 2 118600;Craniometaphyseal dysplasia 123000		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ANKRD11	KBG syndrome 148050		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ANO5	Gnathodiaphyseal dysplasia OMIM:166260;gnathodiaphyseal dysplasia MONDO:0008151;Osteogenesis Imperfecta and Decreased Bone Density;skeletal dysplasias;skeletal dysplasias;Disproportionate Short Stature		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ANTXR2	Hyaline fibromatosis syndrome 228600		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5

ARHGAP31	Adams-Oliver syndrome 1 100300		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ARL6	Polydactyly;Bardet-Biedl syndrome 3 600151		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy) 253200;Mucopolysaccharidosis type VI (Maroteaux-Lamy) 253200		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ARSE	CDPXL;Chondrodysplasia punctata, X-linked recessive, 302950;CHONDRODYSPLASIA PUNCTATA 1, X-LINKED;X-linked recessive chondrodysplasia punctata		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ASXL1	Bohring-Opitz syndrome 605039		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA 219200;Cutis laxa, autosomal recessive, type IIA219200		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ATP7A	Spinal muscular atrophy, distal, 300489;Menkes disease 309400;Occipital horn syndrome 304150		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
AXIN1	neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome, MONDO:0018681		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
B3GALT6	Ehlers-Danlos syndrome, progeroid type, 2 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures 271640		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, 245600;Larsen alike phenotype (skd incl)		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
B4GALT7	Ehlers-Danlos syndrome with short stature and limb anomalies 130070		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BBS1	Polydactyly;Bardet-Biedl syndrome 1 OMIM:209900;Bardet-Biedl syndrome 1 MONDO:0008854		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BBS10	Polydactyly;Bardet Biedl syndrome 10, 615987		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BBS12	Polydactyly;Bardet Biedl syndrome 12, 615989		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BBS2	Polydactyly;Bardet-Biedl syndrome 2, 615981		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BBS4	Polydactyly;Bardet-Biedl syndrome 4, 615982		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BBS5	Polydactyly;Bardet Biedl syndrome 5, 615983		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BBS7	Polydactyly;Bardet-Biedl syndrome 7, 615984		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BBS9	Polydactyly;Bardet Biedl syndrome 9, 615986		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BHLHA9	Syndactyly, mesoaxial synostotic, with phalangeal reduction 609432		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BMP1	Osteogenesis imperfecta, type XIII, 614856		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BMP2	short stature, facial dysmorphism and skeletal anomalies with or without cardiac aomalies 617877.;Brachydactyly, type A2 112600;{HFE hemochromatosis, modifier of} 235200		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BMPER	Diaphanospondylodysostosis 608022		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BMPR1B	Acromesomelic dysplasia, Demirhan type, OMIM:609441;Brachydactyly, type A1, D, OMIM:616849;Brachydactyly, type A2, OMIM:112600		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis 259730		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CANT1	Desbuquois dysplasia 1 251450;multiple epiphyseal dysplasia type 7, 617719.		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CASR	Hypocalcemia, autosomal dominant 601198;Hypocalciuric hypercalcemia, type I 145980;Hyperparathyroidism, neonatal 239200;Hypocalcemia, autosomal dominant, with Bartter syndrome 601198		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CBFB	Cleidocranial dysplasia 2, OMIM:620099;cleidocranial dysplasia 2, MONDO:0859307		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CC2D2A	Meckel syndrome 6 612284		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CCDC8	3-M syndrome 3, 614205		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy 225280		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CDKN1C	IMAGE syndrome 614732		MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CDT1	Meier-Gorlin syndrome 4, OMIM:613804		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CEP290	Meckel syndrome 4 611134;Senior-Loken syndrome 6 610189;Joubert syndrome 5 610188;Bardet-Biedl syndrome 14 615991;Leber congenital amaurosis 10		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1 601776		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations (recessive Larsen syndrome) 143095		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CHSY1	Temtamy preaxial brachydactyly syndrome 605282		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CLCN5	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis 308990;Dent disease 300009;Nephrolithiasis, type I 310468;Hypophosphatemic rickets 300554		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CLCN7	Osteopetrosis, autosomal recessive 4, OMIM:611490;Osteopetrosis, autosomal dominant 2, OMIM:166600		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COG1	Congenital disorder of glycosylation, type IIg 611209		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COG4	Saul-Wilson syndrome, OMIM:618150;microcephalic osteodysplastic dysplasia, Saul-Wilson type, MONDO:0019407		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COL10A1	Metaphyseal chondrodysplasia, Schmid type 156500		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COL11A1	Stickler syndrome, type II, OMIM:604841;Marshall syndrome, OMIM:154780;Fibrochondrogenesis 1, OMIM:228520		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COL11A2	Stickler syndrome, type III 184840;Otospondylomegaepiphyseal dysplasia 215150;Fibrochondrogenesis 2 614524?;Fibrochondrogenesis 2 614524;Weissenbacher-Zweymuller syndrome 277610		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COL1A1	Osteogenesis imperfecta, type I 166200;Caffey disease 114000;Osteogenesis imperfecta, type III 259420;Osteogenesis imperfecta, type II 166210;Ehlers-Danlos syndrome, type VIIA 130060;Ehlers-Danlos syndrome, classic 130000;Osteogenesis imperfecta, type IV 166220		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COL1A2	Ehlers-Danlos syndrome, cardiac valvular form, OMIM:225320;Ehlers-Danlos syndrome, type VIIB, OMIM:130060;Osteogenesis imperfecta, type II, OMIM:166210;Osteogenesis imperfecta, type III, OMIM:259420;Osteogenesis imperfecta, type IV, OMIM:166220		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COL2A1	Epiphyseal dysplasia, multiple, with myopia and deafness 132450;Spondyloepiphyseal dysplasia, Stanescu type 616583;Stickler sydrome, type I, nonsyndromic ocular 609508;Achondrogenesis, type II or hypochondrogenesis 200610;Kniest dysplasia 156550;Legg-Calve-Perthes disease 150600;Otospondylomegaepiphyseal dysplasia 215150;Stickler syndrome, type I 108300;SM		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COL9A1	Stickler syndrome, type IV, OMIM:614134;Epiphyseal dysplasia, multiple, 6, OMIM:614135		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COL9A2	Stickler syndrome, type V, OMIM:614284;Epiphyseal dysplasia, multiple, 2, OMIM:600204		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy, OMIM:600969		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COLEC11	3MC syndrome 2 265050		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COMP	Epiphyseal dysplasia, multiple, 1 132400;Pseudoachondroplasia 177170		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CREBBP	Rubinstein-Taybi syndrome 180849;Rubinstein-Taybi syndrome180849		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CRTAP	Osteogenesis imperfecta, type VII 610682		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CSGALNACT1	Desbuquois dysplasia with mild joint laxity;non-proportionate short stature;Skeletal dysplasia, mild, with joint laxity and advanced bone age OMIM:618870;skeletal dysplasia, mild, with joint laxity and advanced bone age MONDO:0030029		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CTSA	Galactosialidosis 256540		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CTSC	Haim-Munk syndrome 245010,;Haim-Munk syndrome 245010		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CTSK	Pycnodysostosis 265800		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CUL7	3-M syndrome 1 273750		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CYP27B1	Vitamin D-dependent rickets, type I 264700		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CYP2R1	Rickets due to defect in vitamin D 25-hydroxylation, 600081		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type 271665, at least 3 cases reported;Spondylometaepiphyseal dysplasia, short limb-hand type 271665		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DHCR24	Desmosterolosis 602398		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DHCR7	Smith-Lemli-Opitz syndrome 270400		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DHODH	Miller syndrome (postaxial acrofacial dysostosis) 263750		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DIS3L2	Perlman syndrome 267000		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DLL3	Spondylocostal dysostosis 1, autosomal recessive 277300		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DLX3	Amelogenesis imperfecta, type IV 104510;Trichodontoosseous syndrome 190320		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DLX5	?Split-hand/foot malformation 1 with sensorineural hearing loss, OMIM:220600;Split-hand/foot malformation 1, OMIM:183600		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DMP1	Multiple synostoses syndrome 2, 610017; Chondrodysplasia, Grebe type, 200700; Brachydactyly, type C, 113100;Hypophosphatemic rickets, AR, 241520; Brachydactyly, type A1, C, 615072; Brachydactyly, type A2, 112600;Acromesomelic dysplasia, Hunter-Thompson type, 201250; {Osteoarthritis-5}, 612400;Hypophosphatemic rickets,autosomal recessive,type 1 (ARHR1); sk		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DNMT3A	Tatton-Brown-Rahman syndrome 615879		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DOCK6	Adams-Oliver syndrome 2 614219;Adams-Oliver syndrome 2614219		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DPAGT1	Congenital disorder of glycosylation, type Ij 608093;Myasthenic syndrome, congenital, 13, with tubular aggregates 614750;UDP-GlcNAc:Dol-P-GlcNac-P transferase deficiency (Disorders of protein N-glycosylation)		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DPM1	Congenital disorder of glycosylation, type Ie, OMIM:608799		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DYM	Dyggve-Melchior-Clausen disease 223800;Smith-McCort dysplasia 607326		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly, OMIM:613091		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
EBP	X-linked dominant chondrodysplasia punctata;CDPXLD;MEND syndrome;Chondrodysplasia punctata, X-linked dominant, 302960;MEND syndrome-300960 XLR.		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type 610536		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
EIF2AK3	Wolcott-Rallison syndrome226980;Wolcott-Rallison syndrome 226980		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ENPP1	Hypophosphatemic rickets, autosomal recessive, 2 613312;Arterial calcification, generalized, of infancy, 1 208000;Cole disease 615522		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ESCO2	SC phocomelia syndrome 269000;Roberts syndrome 268300		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
EVC	Ellis-van Creveld syndrome, 225500;ECV1;Ellis-van Creveld Syndrome;Ellis-van Creveld syndrome, 225500Weyers acrodental dysostosis, 193530		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
EVC2	Ellis-van Creveld syndrome 225500;Weyers acrofacial dysostosis 193530		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
EXT1	trichorhinophalangeal syndrome type 2 -150230;Exostoses, multiple, type 1133700;Exostoses, multiple, type 13370		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
EXT2	Exostoses, multiple, type 2, OMIM:133701		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities, OMIM:617425		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
EZH2	Weaver syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FAM20C	Raine syndrome 259775		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FAM58A	STAR syndrome300707;STAR syndrome 300707		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FBN1	Geleophysic dysplasia 2 614185;Stiff skin syndrome 184900;Marfan syndrome 154700;Acromicric dysplasia 102370;Weill-Marchesani syndrome 2, dominant 608328		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FBN2	Contractural arachnodactyly, congenital OMIM:121050;congenital contractural arachnodactyly MONDO:0007363		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FBXW11	Neurodevelopmental, jaw, eye, and digital syndrome, OMIM:618914;neurodevelopmental, jaw, eye, and digital syndrome, MONDO:0030057		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FERMT3	Leukocyte adhesion deficiency, type III 612840;(Moderate osteopetrosis) Kilic SS et al. The clinical spectrum of leukocyte adhesion deficiency (LAD) III due to defective CalDAG-GEF1. J Clin Immunol. 2009 Jan, 29(1):117-22.		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FGF10	LADD syndrome 149730		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FGF23	Hypophosphatemic rickets, autosomal dominant 193100;Osteomalacia, tumor-induced;Tumoral calcinosis, hyperphosphatemic, familial 211900		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FGF9	Multiple synostoses syndrome 3, OMIM:612961;multiple synostoses syndrome 3, MONDO:0013064		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FGFR1	Hypogonadotropic hypogonadism 2 with or without anosmia 147950;Encephalocraniocutaneous lipomatosis, somatic mosaism 613001;Pfeiffer syndrome 101600;Trigonocephaly 1 190440;Hartsfield syndrome 615465;Jackson-Weiss syndrome 123150;Osteoglophonic dysplasia 166250		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FGFR2	Beare-Stevenson cutis gyrata syndrome 123790;Craniosynostosis, nonspecific Crouzon syndrome 123500;Craniofacial-skeletal-dermatologic dysplasia 101600;Pfeiffer syndrome 101600;Gastric cancer, somatic 613659;Jackson-Weiss syndrome 123150;LADD syndrome 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis 207410;Apert synd		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FGFR3	Thanatophoric dysplasia, type I 187600;Muenke syndrome 602849;CATSHL syndrome 610474;SADDAN 616482;Thanatophoric dysplasia, type II 187601;Achondroplasia 100800;LADD syndrome 149730;Hypochondroplasia 146000;Crouzon syndrome with acanthosis nigricans 612247		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FIG4	Charcot-Marie-Tooth disease, type 4J, OMIM:611228;Charcot-Marie-Tooth disease type 4J MONDO:0012640;Yunis Varon syndrome OMIM:216340;Yunis-Varon syndrome MONDO:0008995		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FKBP10	Osteogenesis Imperfecta, Recessive;Brucks syndrome 1 - 259450;Osteogenesis imperfecta, type XI, 610968;Brucks syndrome;Osteogenesis Imperfecta and Decreased Bone Density; skeletal dysplasias		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FLNA	Frontometaphyseal dysplasia 1, OMIM:305620;Melnick-Needles syndrome, OMIM:309350;Otopalatodigital syndrome, type I, OMIM:311300;Otopalatodigital syndrome, type II, OMIM:304120;Terminal osseous dysplasia, OMIM:300244		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FLNB	Spondylocarpotarsal synostosis syndrome 272460;Atelosteogenesis, type III 108721;Boomerang dysplasia 112310;Atelosteogenesis, type I 108720;Larsen syndrome 150250		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FN1	Spondylometaphyseal dysplasia, corner fracture type 184255		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FUCA1	Fucosidosis 230000		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GALNS	Mucopolysaccharidosis IVA 253000		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial I 211900;Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome 211900		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GDF5	Brachydactyly, type C 113100;Acromesomelic dysplasia, Hunter-Thompson type 201250;Du Pan syndrome 228900;{Osteoarthritis-5} 612400;Chondrodysplasia, Grebe type 200700;Brachydactyly, type A2 112600;Brachydactyly, type A1, C 615072;Symphalangism, proximal, 1B 615298;Multiple synostoses syndrome 2 610017		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GDF6	Klippel-Feil syndrome 1, autosomal dominant 118100;Multiple synostoses syndrome type 4 - 617898.		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GHR	Laron dwarfism, OMIM:262500;Growth hormone insensitivity, partial, OMIM:604271;Increased responsiveness to growth hormone, OMIM:604271		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GJA1	Craniometaphyseal dysplasia, autosomal recessive, OMIM:218400;Oculodentodigital dysplasia, OMIM:164200;Oculodentodigital dysplasia, autosomal recessive, OMIM:257850;Syndactyly, type III, OMIM:186100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GLB1	GM1-gangliosidosis, type II 230600;GM1-gangliosidosis, type I 230500;GM1-gangliosidosis, type III 230650;Mucopolysaccharidosis type IVB (Morquio) 253010		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GLI3	Greig cephalopolysyndactyly syndrome, OMIM:175700;Pallister-Hall syndrome, OMIM:146510;Polydactyly, postaxial, types A1 and B, OMIM:174200;Polydactyly, preaxial, type IV, OMIM:174700		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GNAS	Pseudohypoparathyroidism Ia, OMIM:103580;pseudohypoparathyroidism type 1A, MONDO:0007078;Pseudohypoparathyroidism Ib, OMIM:603233;pseudohypoparathyroidism type 1B, MONDO:0011301;Pseudohypoparathyroidism Ic, OMIM:612462;pseudohypoparathyroidism type 1C, MONDO:0012911;McCune-Albright syndrome, somatic, mosaic, OMIM:174800;panostotic fibro		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GNPAT	Rhizomelic Chondrodysplasia Punctata;RCDP2;Rhizomelic chondrodysplasia punctata type 2;Chondrodysplasia punctata, rhizomelic, type 2, 222765		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GNPTAB	Mucolipidosis III alpha/beta 252600;Mucolipidosis II alpha/beta 252500		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GNPTG	Mucolipidosis III gamma 252605		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GNS	Mucopolysaccharidosis type IIID 252940		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GORAB	Geroderma osteodysplasticum 231070		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GPC6	Omodysplasia 1 258315		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GPX4	Spondylometaphyseal dysplasia, Sedaghatian type 250220		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GSC	Foundation Trust) Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities 602471;Foundation Trust)Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities 602471		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GUSB	Mucopolysaccharidosis VII 253220		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
HDAC8	Wilson-Turner syndrome 309585;Cornelia de Lange syndrome 5 300882		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
HES7	Spondylocostal dysostosis 4, autosomal recessive 613686		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C) 252930		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
HOXA13	Hand-foot-genital syndrome 140000;Hand-foot-uterus syndrome 140000;Guttmacher syndrome 176305		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
HOXD13	Syndactyly, type V 186300;Brachydactyly-syndactyly syndrome 610713;Brachydactyly, type E 113300;Synpolydactyly 1 186000;Brachydactyly, type D 113200		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
HPGD	Cranioosteoarthropathy 259100;Digital clubbing, isolated congenital 119900;Hypertrophic osteoarthropathy, primary, autosomal recessive 1 259100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
HSPG2	Schwartz-Jampel syndrome, type 1, OMIM:255800;Schwartz-Jampel syndrome, MONDO:0009717;Dyssegmental dysplasia, Silverman-Handmaker type, OMIM:224410;Silverman-Handmaker type dyssegmental dysplasia, MONDO:0009140		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ICK	Endocrine-cerebroosteodysplasia, OMIM:612651;Endocrine-cerebro-osteodysplasia syndrome, MONDO:0012980		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IDH1	Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria 614875;Maffucci syndrome 614569;Ollier disease/ Dyschondroplasia 166000		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IDS	Mucopolysaccharidosis II 309900		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IDUA	Mucopolysaccharidosis Ih 607014;Mucopolysaccharidosis Is 607016;Mucopolysaccharidosis Ih/s 607015		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IFIH1	Singleton-Merten syndrome 1, OMIM:182250		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IFITM5	Osteogenesis imperfecta, type V 610967		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IFT122	Cranioectodermal dysplasia 1 218330		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IFT140	Short-rib thoracic dysplasia 9 with of without polydactyly, 266920		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IFT43	Short-rib thoracic dysplasia 18 with polydactyly - 617866;?Cranioectodermal dysplasia 3 - 614099		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly 611263		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IHH	Acrocapitofemoral dysplasia, OMIM:607778;Brachydactyly, type A1, OMIM:112500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IKBKG	Ectodermal dysplasia and immunodeficiency 1, OMIM:300291		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IL11RA	Craniosynostosis and dental anomalies 614188		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IL1RN	Interleukin 1 receptor antagonist deficiency, OMIM:612852		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IMPAD1	Chondrodysplasia with joint dislocations, GPAPP type 614078		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
KAT6B	Genitopatellar syndrome 606170;SBBYSS syndrome 603736;GTPTS,Ohdo		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2 603546		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
KIF7	Joubert syndrome 12 200990;Acrocallosal syndrome 200990;Al-Gazali-Bakalinova syndrome 607131;Hydrolethalus syndrome 2 614120		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
KMT2D	Kabuki syndrome 1 - 147920		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LBR	Pelger-Huet anomaly with mild skeletal anomalies 618019;Greenberg skeletal dysplasia 215140;Pelger-Huet anomaly 169400		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LEMD3	Melorheostosis with osteopoikilosis 155950 IC;Osteopoikilosis 166700;Buschke-Ollendorff syndrome 166700		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, OMIM:601559		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LMBR1	Laurin-Sandrow syndrome 135750;Polydactyly, preaxial type II 174500;Hypoplastic or aplastic tibia with polydactyly 188740;Triphalangeal thumb, type I 174500;Triphalangeal thumb-polysyndactyly syndrome 174500;Syndactyly, type IV 186200;Acheiropody 200500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LMNA	Emery-Dreifuss muscular dystrophy 2, 181350;Heart-hand syndrome, Slovenian type 610140;Charcot-Marie-Tooth disease, type 2B1 605588;Emery-Dreifuss muscular dystrophy 3, 616516;Cardiomyopathy, dilated, 1A 115200;Hutchinson-Gilford progeria 176670;Mandibuloacral dysplasia 248370;Lipodystrophy, familial partial, 2 151660;Restrictive dermopathy, lethal 275210;Foun		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LMX1B	Nail-patella syndrome161200;Nail-patella syndrome 161200		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LPIN2	Majeed syndrome, OMIM:609628;Chronic recurrent multifocal osteomyelitis with congenital dyserythropoietic anemia		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LRP4	Sclerosteosis 2 614305;Cenani-Lenz syndactyly syndrome 212780		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LRP5	Exudative vitreoretinopathy 4 601813;[Bone mineral density variability 1] 601884;Osteopetrosis, autosomal dominant 1 607634;Osteosclerosis 144750;van Buchem disease, type 2 607636;Osteoporosis-pseudoglioma syndrome 259770;Hyperostosis, endosteal 144750;{Osteoporosis} 166710		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LTBP1	inherited cutis laxa MONDO:0100237;Cutis laxa, autosomal recessive, type IIE, OMIM:619451		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LTBP3	Geleophysic dysplasia 3, OMIM:617809;Dental anomalies and short stature, OMIM:610216;geleophysic dysplasia 3, MONDO:0054722,		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MAFB	Multicentric carpotarsal osteolysis syndrome 166300		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MAN2B1	Mannosidosis, alpha-, types I and II 248500		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MASP1	3MC syndrome 1 - 257920		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MATN3	MED;multiple epiphyseal dysplasia;Multiple Epiphyseal Dysplasia, Dominant;Disproportionate Short Stature;Epiphyseal dysplasia, multiple, 5, 607078; {Osteoarthritis susceptibility 2}, 140600;Spondyloepimetaphyseal dysplasia, 608728; Spondyloepimetaphyseal dysplasia, 608728		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MESP2	Spondylocostal dysostosis 2, autosomal recessive 608681		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MGP	Keutel syndrome, OMIM:245150;spondyloepiphyseal dysplasia, MONDO:0016761		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MKKS	Polydactyly;Bardet-Biedl syndrome 6, 605231;McKusick-Kaufman syndrome, 236700		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MKS1	Meckel syndrome 1 249000;Bardet-Biedl syndrome 13 615990		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MMP13	Metaphyseal anadysplasia 1 602111;Spondyloepimetaphyseal dysplasia, Missouri type 602111;Metaphyseal dysplasia, Spahr type - 250400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MMP2	Multicentric osteolysis, nodulosis, and arthropathy 259600		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MNX1	Currarino syndrome 176450		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MPDU1	Congenital disorder of glycosylation, type If 609180		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MSX2	Craniosynostosis, type 2 604757;Parietal foramina with cleidocranial dysplasia 168550;Parietal foramina 1 168500		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MYCN	Feingold syndrome (Microcephaly-oculo-digito-esophageal-duodenal) 164280		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MYH3	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, OMIM:178110;Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, OMIM:618469;contractures, pterygia, and spondylocarpotarsal fusion syndrome 1A, MONDO:0008338		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MYO18B	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, OMIM:616549		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5

NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B) 252920		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NBAS	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NEK1	Short rib thoracic dysplasia 6 with or without polydactyly - 263520;SRPS type 2 (Majewski);Short Rib Polydactyly Syndrome;Short rib-polydactyly syndrome, type IIA, 263520		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NEU1	Sialidosis, type II 256550;Sialidosis, type I 256550		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NF1	Neurofibromatosis, type 1 162200;Neurofibromatosis, type 1162200;Neurofibromatosis, familial spinal 162210;Neurofibromatosis-Noonan syndrome601321;Neurofibromatosis, familial spinal162210;Neurofibromatosis-Noonan syndrome 601321		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NFIX	Marshall-Smith syndrome 602535;Sotos syndrome 2 614753		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NIPBL	Cornelia de Lange syndrome 1 122470		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia 613330		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NLRP3	CINCA syndrome, OMIM:607115		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NOG	Symphalangism, proximal, 1A 185800;Brachydactyly, type B2 611377;Tarsal-carpal coalition syndrome 186570;Stapes ankylosis with broad thumb and toes 184460;Multiple synostoses syndrome 1 186500		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NOTCH1	Limb, scalp and skull defects;AOS;Combination of aplasia cutis congenita of the scalp vertex and terminal transverse limb defects (e.g., amputations, syndactyly, brachydactyly, or oligodactyly);Adams-Oliver syndrome 5, 616028		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NOTCH2	Hajdu-Cheney (Serpentine fibula polycystic kidney) syndrome 102500;Alagille syndrome 2 610205		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NPR2	Acromesomelic dysplasia, Maroteaux type 602875;Short stature with nonspecific skeletal abnormalities 616255;Epiphyseal chondrodysplasia, Miura type 615923		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NPR3	Tall stature;arachnodactyly;extra epiphyses;aortic dilatation		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NSD1	Sotos syndrome 1 117550		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NSDHL	Congenital hemidysplasia, ichthyosis, limb defects (CHILD) syndrome 308050;CK syndrome 300831		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
OBSL1	3-M syndrome 2 612921		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
OFD1	Joubert syndrome 10 300804;Simpson-Golabi-Behmel syndrome, type 2 300209 XLR;Orofaciodigital syndrome I 311200 XLD		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ORC1	Meier-Gorlin syndrome 1, OMIM:224690		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ORC4	Meier-Gorlin syndrome 2, OMIM:613800		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ORC6	Meier-Gorlin syndrome 3, OMIM:613803		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
OSTM1	Osteopetrosis, autosomal recessive 5 259720		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PAPSS2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes 612847		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PAX3	Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 1, 193500;Waardenburg syndrome, type 3, 148820		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, OMIM:210720		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PDE4D	Acrodysostosis 2, with or without hormone resistance 614613;Acrodysostosis 2, with or without hormone resistance614613		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PEX5	Peroxisome biogenesis disorder 2A (Zellweger) 214110;Rhizomelic chondrodysplasia punctata, type 5 616716		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PEX7	Rhizomelic chondrodysplasia punctata, type 1, 215100;Rhizomelic CDP type 1;Rhizomelic chondrodysplasia punctata, type 1, 215100		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PHEX	Hypophosphatemic rickets, X-linked dominant 307800		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PHGDH	Neu-Laxova syndrome 1 256520;Phosphoglycerate dehydrogenase deficiency 601815		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PIGV	Hyperphosphatasia with mental retardation syndrome 1 239300		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PIK3R1	SHORT syndrome 269880		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PITX1	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly 119800;Liebenberg syndrome 186550		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PLOD2	Bruck syndrome 2 609220		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
POLR1C	Treacher Collins syndrome 3 248390		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
POLR1D	Treacher Collins syndrome 2 613717		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
POP1	Anauxetic dysplasia 2, 617396		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase 613571		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PPIB	Osteogenesis imperfecta, type IX259440;Osteogenesis imperfecta, type IX 259440		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PRKAR1A	Acrodysostosis 1, with or without hormone resistance, OMIM:101800		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures 617157		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PSAT1	Neu-Laxova syndrome 2 616038		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PSPH	Phosphoserine phosphatase deficiency 614023		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PTH1R	Failure of tooth eruption, primary 125350;Eiken syndrome 600002;Metaphyseal chondrodysplasia, Murk Jansen type 156400;Chondrodysplasia, Blomstrand type 215045		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PTHLH	Brachydactyly, type E2 613382;Brachydactyly, type E2613382		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PTPN11	Metachondromatosis 156250;LEOPARD syndrome 1 151100;Noonan syndrome 1 163950;LEOPARD syndrome 1151100		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PYCR1	Cutis laxa, autosomal recessive, type IIB 612940;Cutis laxa, autosomal recessive, type IIIB 614438		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RAB23	Carpenter syndrome 201000		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RASGRP2	Bleeding disorder, platelet-type, 18 615888, also with osteopetrosis like bone abnormalities and neurodevelopmental defects;Bleeding disorder, platelet-type, 18 615888		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RBM8A	Thrombocytopenia-absent radius syndrome 274000		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RECQL4	Baller-Gerold syndrome, OMIM:218600;RAPADILINO syndrome, OMIM:266280;Rothmund-Thomson syndrome, type 2, OMIM:268400		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RFT1	Congenital disorder of glycosylation, type In 612015		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RMRP	Cartilage-hair hypoplasia 250250;Anauxetic dysplasia 607095;Metaphyseal dysplasia without hypotrichosis 250460		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RNU4ATAC	Lowry-Wood syndrome, OMIM:226960;Microcephalic osteodysplastic primordial dwarfism, type I, OMIM:210710;Roifman syndrome, OMIM:616651		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ROR2	Brachydactyly, type B1, OMIM:113000 (AD);Robinow syndrome, autosomal recessive, OMIM:268310 (AR)		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RPGRIP1L	COACH syndrome 216360;Meckel syndrome 5 611561;Joubert syndrome 7 611560		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RUNX2	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly 156510;Cleidocranial dysplasia, forme fruste, with brachydactyly 119600;Cleidocranial dysplasia, forme fruste, dental anomalies only 119600;Cleidocranial dysplasia 119600		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SALL1	Townes Brocks syndrome (Renal-Ear-Anal-Radial syndrome) 107480		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SALL4	Okihiro (Duane-radial ray) syndrome 607323;IVIC syndrome 147750		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SBDS	Shwachman-Diamond syndrome 260400;Shwachman-Diamond syndrome260400		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SCARF2	Van den Ende-Gupta syndrome 600920		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SERPINF1	Osteogenesis imperfecta, type VI, 613982;osteogenesis imperfecta;OI/osteoporosis;Osteogenesis Imperfecta, Recessive		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SERPINH1	Osteogenesis Imperfecta, Recessive;OI3;Osteogenesis Imperfecta and Decreased Bone Density;skeletal dysplasias;{Preterm premature rupture of the membranes, susceptibility to}, 610504;Osteogenesis imperfecta, type X, 613848		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SETD2	Luscan-Lumish syndrome616831;Luscan-Lumish syndrome 616831		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SF3B4	Acrofacial dysostosis 1, Nager type 154400		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SGSH	Mucopolysaccharidisis type IIIA (Sanfilippo A) 252900		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SH3BP2	Cherubism, OMIM:118400		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SH3PXD2B	Frank-ter Haar syndrome 249420		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SHOX	Langer mesomelic dysplasia, OMIM:249700 (PR);Leri-Weill dyschondrosteosis, OMIM:127300 (PD);Short stature, idiopathic familial, OMIM:300582;Dorsolateral bowed, short radii;Bowing and curving of radius;Radioulnar shortening		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SKI	Shprintzen-Goldberg syndrome 182212		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SLC17A5	Salla disease, OMIM:604369;Sialic acid storage disorder, infantile, OMIM:269920		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SLC26A2	ACG1B,DD,rMED;multiple epiphyseal dysplasia;Multiple Epiphyseal Dysplasia, Recessive;Epiphyseal dysplasia, multiple, 4		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome, OMIM:602782		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SLC34A1	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SLC34A3	Hypophosphatemic rickets with hypercalciuria 241530		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SLC35C1	Congenital disorder of glycosylation, type IIc 266265;GDP-fucose transporter deficiency (Disorders of multiple glycosylation and other glycosylation pathways)		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SLC35D1	Schneckenbecken dysplasia 269250;Schneckenbecken dysplasia269250		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SLC39A13	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like 612350		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive 2 614441;Hypertrophic osteoarthropathy, primary, autosomal recessive 2614441		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SMAD3	Loeys-Dietz syndrome 3 613795		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SMAD4	Myhre syndrome139210;Myhre syndrome 139210		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SMAD6	{Radioulnar synostosis, nonsyndromic}, OMIM:179300		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SMARCAL1	Schimke immunoosseous dysplasia, OMIM:242900		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SMC1A	Cornelia de Lange syndrome 2, OMIM:300590;Cornelia de Lange syndrome 2, MONDO:0010370		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SMC3	Cornelia de Lange syndrome 3 610759		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SMOC1	Microphthalmia with limb anomalies 206920;Ophthalmo-acromelic syndrome;Polydactyly		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SNX10	Osteopetrosis, autosomal recessive 8 615085		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SOST	Craniodiaphyseal dysplasia, autosomal dominant 122860;Van Buchem disease 239100;Sclerosteosis 1 269500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SOX9	Campomelic dysplasia with autosomal sex reversal 114290;Acampomelic campomelic dysplasia 114290;Campomelic dysplasia 114290		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SP7	Osteogenesis imperfecta, type XII 613849		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SUMF1	Multiple sulfatase deficiency 272200		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TALDO1	Transaldolase deficiency 606003		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TBCE	Kenny-Caffey syndrome, type 1 244460.;Hypoparathyroidism-retardation-dysmorphism syndrome 241410;Kenny-Caffey syndrome, type 1 244460		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TBX15	Cousin syndrome 260660		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TBX3	Ulnar-mammary syndrome 181450		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TBX4	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, OMIM:147891		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TBX5	Holt-Oram syndrome 142900		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TBX6	Spondylocostal dysostosis 5122600;Spondylocostal dysostosis 5 122600		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TBXAS1	Ghosal hematodiaphyseal syndrome, OMIM:231095		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TCIRG1	Osteopetrosis, autosomal recessive 1 259700		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TCOF1	Treacher Collins syndrome 1 154500		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TCTN2	Joubert syndrome 24 616654;Meckel syndrome 8 613885		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TERT	Dyskeratosis congenita, autosomal dominant 2, OMIM:613989;Dyskeratosis congenita, autosomal recessive 4, OMIM:613989		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TGFB1	Camurati-Engelmann disease, OMIM:131300		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TGFB2	Loeys-Dietz syndrome 4 614816		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TGFBR2	Loeys-Dietz syndrome 2 610168		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome 213980;Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome213980		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TMEM165	Congenital disorder of glycosylation, type IIk 614727		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TMEM216	Meckel syndrome 2 603194;Joubert syndrome 2 608091		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TNFRSF11A	Osteolysis, familial expansile 174810;Osteopetrosis, autosomal recessive 7 612301;Paget disease of bone 2, early-onset 602080		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TNFRSF11B	Paget disease of bone 5, juvenile-onset239000;Paget disease of bone 5, juvenile-onset 239000		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TNFSF11	Osteopetrosis, autosomal recessive 2 259710		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TP63	ADULT syndrome, OMIM:103285;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, OMIM:604292;Limb-mammary syndrome, OMIM:603543;Split-hand/foot malformation 4, OMIM:605289		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TRAPPC2	Spondyloepiphyseal dysplasia tarda 313400;Spondyloepiphyseal dysplasia tarda313400		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TREM2	Nasu-Hakola disease 221770		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TRIP11	Achondrogenesis, type IA200600;Achondrogenesis, type IA 200600		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TRPS1	Trichorhinophalangeal syndrome, type III 190351;Trichorhinophalangeal syndrome, type I 190350		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TRPV4	Brachyolmia type 3 113500;Hereditary motor and sensory neuropathy, type IIc 606071;Digital arthropathy-brachydactyly, familial 606835;SED, Maroteaux type 184095;Parastremmatic dwarfism 168400;Metatropic dysplasia 156530;Scapuloperoneal spinal muscular atrophy 181405;Spinal muscular atrophy, distal, congenital nonprogressive 600175;Spondylometaphyseal dysplasia		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TTC21B	Nephronophthisis 12, 613820;Asphyxiating Thoracic Dystrophy;SRTD4		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TTC8	Polydactyly;Bardet-Biedl syndrome 8, 615985		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TWIST1	Robinow-Sorauf syndrome 180750;Saethre-Chotzen syndrome 101400;Craniosynostosis, type 1 123100;Saethre-Chotzen syndrome with eyelid anomalies 101400		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TYROBP	Nasu-Hakola disease 221770		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
VDR	Rickets, vitamin D-resistant, type IIA, 277440		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
WDPCP	?Congenital heart defects, hamartomas of tongue, and polysyndactyly 217085;?Bardet-Biedl syndrome 15, 615992		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
WDR19	Cranioectodermal dysplasia 4, 614378;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376;Asphyxiating thoracic dystrophy 5, 614376;SRTD5		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
WDR35	Short-rib thoracic dysplasia 7 with or without polydactyly 614091;Cranioectodermal dysplasia 2 613610		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood 208230;Spondyloepiphyseal dysplasia tarda with progressive arthropathy 208230		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
WNT10B	Split-hand/foot malformation 6, OMIM:225300		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
WNT5A	Robinow syndrome, autosomal dominant 1 180700		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
WNT7A	Ulna and fibula, absence of, with severe limb deficiency 276820;Fuhrmann syndrome 228930		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
XRCC4	Short stature, microcephaly, and endocrine dysfunction, OMIM:616541		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
XYLT1	Desbuquois dysplasia 2, OMIM:615777;Desbuquois dysplasia 2, MONDO:0014343		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
XYLT2	Spondyloocular syndrome 605822		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
YY1	Gabriele-de Vries syndrome617557;Gabriele-de Vries syndrome 617557		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy 608612;Restrictive dermopathy, lethal 275210		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
B9D1	Meckel syndrome 9, OMIM:614209;Meckel syndrome 9, MONDO:0013630;Joubert syndrome 27, OMIM:617120;Joubert syndrome 27, MONDO:0014927		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DCC	Gaze palsy, familial horizontal, with progressive scoliosis, 2, OMIM:617542		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DROSHA	Global developmental delay;Intellectual disability;Seizures;Cerebral white matter atrophy;Abnormality of the corpus callosum;Abnormality of movement;Stereotypic behavior;Abnormality of head or neck;Short foot		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FBLN1	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses 608180		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
HDAC4	Albright hereditary osteodystrophy type 3;Brachydactyly-intellectual disability;Albright hereditary osteodystrophy type 3, Albright hereditary osteodystrophy-like syndrome, Brachydactyly-intellectual disability, Del(2)(q37) 600430;Albright hereditary osteodystrophy-like syndrome;Del(2)(q37) 600430		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
KIF5B	kyphomelic dysplasia, MONDO:0008881		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LFNG	Spondylocostal dysostosis 3, autosomal recessive, OMIM:609813		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MANBA	Beta-mannosidosis, 248510		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MBTPS2	Osteogenesis imperfecta, type XIX, OMIM:301014;osteogenesis imperfecta, type 19, MONDO:0049223		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MIR17HG	FS2;Brachydactyly with short stature and microcephaly;Microcephaly-oculo-digito-esophageal-duodenal syndrome;Feingold syndrome 2, 614326		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MMP9	Metaphyseal anadysplasia 2 613073		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NMNAT1	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual disability, and Leber congenital amaurosis (SHILCA), MIM#619260		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NT5E	Calcification of joints and arteries, OMIM:211800;hereditary arterial and articular multiple calcification syndrome, MONDO:0008895		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PFN1	Paget s disease of bone;bone Paget disease MONDO:0005382		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PLEKHM1	Osteopetrosis, autosomal recessive 6 - 611497;Osteopetrosis, autosomal recessive 6 611497;Osteopetrosis, autosomal dominant 3 - 618107		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RAD21	Cornelia de Lange syndrome 4, OMIM:614701		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ACVR2B	Heterotaxy, visceral, 4, autosomal613751		Unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
AHI1				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
AIPL1				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
AKT1	Proteus syndrome, somatic 176920;Cowden syndrome 6 615109		Unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ARID1A	Coffin-Siris			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ARID1B	Coffin-Siris syndrome type 1 - 135900;Coffin-Siris			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ARL13B				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ATXN10	Spinocerebellar ataxia 10, OMIM:603516		Other	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
B9D2	Joubert syndrome 34, OMIM:614175;Meckel syndrome 10, OMIM:614175;Meckel syndrome, type 10, MONDO:0013609		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
BANF1	Nestor-Guillermo progeria syndrome614008		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
C2orf71				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
C5orf42				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CCDC28B	{Bardet-Biedl syndrome 1, modifier of}, 209900		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CCDC39				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CCDC40				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CD96	C-syndrome 217750 (opitz trigonocephaly)			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CDC6	Meier-Gorlin syndrome 5, OMIM:613805		BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CDH23				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CEP41				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CFTR				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CLRN1				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COL12A1	{Lung cancer, susceptibility to, association with}(Rudd (2006) Genome Res 16,693); Bethlem-like myopathy (Hicks (2014) Hum Mol Genet 23,2353); Joint hypermobility syndrome with myopathy (Zou (2014) Hum Mol Genet 23, 2339)			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
COL5A1				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CRB1				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
CRELD1				Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5

CRX			Skeletal dysplasias	Skeletal dysplasias	Skeletal disorders	7.5
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FLT4	Split-hand/foot malformation 1 183600 Split-hand/foot malformation 1 with sensorineural hearing loss 220600	Unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DNAAF1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DNAAF2			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DNAAF3			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DNAH11			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DNAH5			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DNAI1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DNAI2			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DNAL1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DPM3	Congenital disorder of glycosylation, type Io 612937	Unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
DSPP	Dentinogenesis imperfecta, Shields type II, 125490; Deafness, autosomal dominant 39, with dentinogenesis, 605594; Dentinogenesis imperfecta, Shields type III, 125500; Dentin dysplasia, type II, 125420 -3	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
EP300	Rubinstein-Taybi syndrome 180849 Rubinstein-Taybi syndrome		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ESR1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FBLIM1	Majeed syndrome (Chronic recurrent multifocal osteomyelitis with congenital dyserythropoietic anemia) 609628		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FBXW4	Split-hand/foot malformation 3 syndrome 246560	Unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FGF8	Numerous variants reported in Hypogonadotropic hypogonadism 6 with or without anosmia 612702, but this phenotype is not relevant to this panel.	Unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FMN1	Animal models with skeletal dysplastic phenotypes	Unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FOXC1		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
FOXH1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GDF1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GDF3	Klippel-Feil anomaly with laryngeal malformation - 613702	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GLIS2			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GREM1		Unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
GUCY2D			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
HOXA11	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1 605432	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
HYLS1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IDH2	D-2-hydroxyglutaric aciduria 2, OMIM:613657;d-2-hydroxyglutaric aciduria 2, MONDO:0013345	Other	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IMPDH1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
INVS			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
IQCB1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
KCNJ13			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LCA5			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LEFTY2			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LRAT			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LRP6			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
LTBP2	Weill-Marchesani		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MCM5	?Meier-Gorlin syndrome 8B17564	BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MIA3	short stature;skeletal dysplasia;amelogenesis;dentinogenesis imperfecta;short stature;brachydactyly;Platyspondyly;insulin-dependent diabetes mellitus;sensorineural hearing loss;mild intellectual disability	BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MMP14	Winchester syndrome 277950	BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MTAP	Diaphyseal medullary stenosis with malignant fibrous histiocytoma 112250	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
MYO7A			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NEK8			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NKX2-5			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NME8			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NODAL			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NPHP1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NPHP3	Meckel syndrome 7 267010	BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NPHP4			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
NPPC	Overgrowth syndrome with 2q37 translocations	Unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia 258870		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PCDH15			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PHF6	Coffin-Siris syndrome		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PIK3CA	CLOVES 612918		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PINI	No phenotype associated with this gene	Unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PKD2			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PKHD1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PLOD1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
PTPRQ	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RAB3GAP2	Martsolf syndrome 1, OMIM:212720	BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RD3			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RDH12			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RPE65			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RPGR			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RPGRIP1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RSPH4A			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
RSPH9			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SCN1A			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SCN1B			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SCN1G			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SDCCAG8	Bardet-Biedl syndrome 16, 615993	BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SIX1	Preaxial polydactyly type 1 (PPD1)	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SMARCA2	Coffin Siris syndrome		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SMARCA4	Coffin Siris syndrome		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SMARCB1	Coffin Siris syndrome		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SMARCE1	Coffin-Siris syndrome 5, OMIM:616938	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SPATA7			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SPECC1L	Facial clefting, oblique, 1 600251 Opitz GBBB syndrome, type I; 145410 Teebi hypertelorism like syndrome 145420	Other - please specify in evaluation comments	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
SULF1	Mesomelia-synostoses syndrome600383;Mesomelia-synostoses syndrome 600383	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TCTN1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
THPO	Thrombocythemia 1 187950 (rare presentation with congenital limb defects)	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TMEM138			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TMEM237			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TMEM67	COACH syndrome 216360;Meckel syndrome 3 607361;Joubert syndrome 6 610688;Bardet-Biedl syndrome 14, modifier of) 615991	BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TN3B			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TOPORS			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TP53	Central conventional chondrosrcoma	Other	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TRIM32	Bardet-Biedl syndrome 11, 615988 Polydactyly		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TSC1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TSC2			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
TULP1			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
UMOD			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
USH1C			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
USH1G			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
USH2A			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
USP9X	New syndrom with skd		Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
VHL			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
WNT3	Tetra-amelia syndrome 273395	BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
WRN	Werner syndrome 277700	BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
XPNPEP3			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ZBTB16	Skeletal defects, genital hypoplasia, and mental retardation 612447	BIALLELIC, autosomal or pseudoautosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
ZIC3			Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	7.5
EFNB1	Craniofrontonasal syndrome	XLD	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
LEPRE1	Osteogenesis Imperfecta Type VIII	AR	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
TGFBR1	Loeys-Dietz syndrome 1	AD	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
ATR	SECKEL SYNDROME TYPE 1	210600 biallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
TGFB3	LOEYS-DIETZ SYNDROME	615582 monoallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
PLCB4	AURICULOCONDYLAR SYNDROME	602483 monoallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
BRCA1	BRCA1-related Fanconi anaemia	227650 biallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
IGF2	BECKWITH-WIEDEMANN SYNDROME	130650 monoallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
TFAP2B	CHAR SYNDROME	169100 monoallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
SMS	SNYDER-ROBINSON SYNDROME	309583 monoallelic_x_hem	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
FANCA	FANCONI ANEMIA, COMPLEMENTATION GROUP A	227650 biallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
MID1	OPITZ GBBB SYNDROME, X-LINKED	300000 monoallelic_x_hem	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
SEC23A	CRANIOLENTICULOSUTURAL DYSPLASIA	607812 biallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
EDN1	AURICULOCONDYLAR SYNDROME	602483 biallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
PTEN	PROTEUS SYNDROME	176920 monoallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
HYAL1	MUCOPOLYSACCHARIDOSIS TYPE 9	601492 biallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
PORCN	FOCAL DERMAL HYPOPLASIA	305600 monoallelic_X_het	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
FGD1	AARSKOG-SCOTT SYNDROME	305400 monoallelic_X_hem	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
EDNRA	MANDIBULOFACIAL DYSOSTOSIS WITH ALOPECIA	616367 monoallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
TBC1D24	DOORS SYNDROME	220500 biallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
GPC3	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 1	312870 monoallelic_X_hem	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
CSF1R	BRAIN ABNORMALITIES, NEURODEGENERATION, AND DYSOSTEOSCLEROSIS	618476 biallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
GPC4	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 2	300037 monoallelic_X_hem	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
PRRX1	PRRX1-related craniosynostosis	No disease mim monoallelic_autosomal	Skeletal dysplasia	Skeletal dysplasias	Skeletal disorders	
AAAS	Achalasia-addisonianism-alacrimia syndrome, OMIM:231550;Triple-A syndrome, MONDO:0009279	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AARS2	Leukoencephalopathy, progressive, with ovarian failure, OMIM:615889;Combined oxidative phosphorylation deficiency 8, OMIM:614096;fetal hydrops;cardiomyopathy;polyhydramnios;pulmonary effusion	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ABCA12	Ichthyosis, congenital, autosomal recessive 242500	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ABCC6	ARTERIAL CALCIFICATION, GENERALIZED, OF INFANCY, 2	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ABCC9	CANTU SYNDROME HYPERTRICHOTIC OSTEOCHONDRODYSPLASIA	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ABHD5	CHANARIN-DORFMAN SYNDROME	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACADS	ACYL-COA DEHYDROGENASE FAMILY MEMBER TYPE 9 DEFICIENCY	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACADVL	VERY LONG CHAIN ACYL-COENZYME A DEHYDROGENASE DEFICIENCY	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACAN	SPONDYLOEPIPHYSEAL DYSPLASIA TYPE KIMBERLEY;SPONDYLOEPIMETAPHYSEAL DYSPLASIA AGGRECAN TYPE	BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACE	Renal tubular dysgenesis 267430	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACOX1	ADRENOLEUKODYSTROPHY PSEUDONEONATAL;Peroxisomal acyl-CoA oxidase deficiency, OMIM:264470	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACP5	SPONDYLOENCHONDRODYSPLASIA WITH IMMUNE DYSREGULATION	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACTA1	Nemaline myopathy 3, autosomal dominant or recessive, OMIM:161800	BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACTA2	AORTIC ANEURYSM, FAMILIAL THORACIC 6;MOYAMOYA DISEASE 5	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ACTB	BARATTSER-WINTER SYNDROME;ACTB Haploinsufficiency syndrome	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
ACTC1	Atrial septal defect 5 612794	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ACTG1	BARATTSER-WINTER SYNDROME	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ACVR2B	Heterotaxy, Visceroatrial Double outlet right ventricle Transposition of the great arteries Gut malrotation/polysplenia right-sided spleen/asplenia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2, OMIM:600376	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
ACY1	ARNOCYLASE-1 DEFICIENCY	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ADAMTS10	Weill-Marchesani syndrome 1, recessive 277600	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ADAMTS17	Weill-Marchesani 4 syndrome, recessive, 613195	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ADAMTSL2	Geleophysic dysplasia 1 231050	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ADAR	Aicardi-Goutieres syndrome 6, OMIM:615010;Dyschromatosis symmetrica hereditaria, OMIM:127400	BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
ADCY6	Lethal congenital contracture syndrome 8, OMIM:616287,MONDO:0014570	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ADNP	MENTAL RETARDATION, AUTOSOMAL DOMINANT, 28	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ADSL	ADENYLOSUCCINASE DEFICIENCY	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AFF3	KINESHIP syndrome, OMIM:619297	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
AGK	SENGERS SYNDROME	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AGL	GLYCOGEN STORAGE DISEASE TYPE III	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AGPS	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA TYPE 3	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AGTR1	Renal tubular dysgenesis, OMIM:267430	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AHCY	Fetal hydrops S-adenosylhomocysteine hydrolase deficiency;Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AHI1	JOUBERT SYNDROME	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AKT1	PROTEUS SYNDROME	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
AKT2	Hypoinsulinemic hypoglycemia with hemihypertrophy, OMIM:240900;Hypoinsulinemic hypoglycemia and body hemihypertrophy, MONDO:0009416	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, OMIM:615937	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ALDH18A1	CUTIS LAXA, AUTOSOMAL DOMINANT 3;SPASTIC PARAPLEGIA 9, AUTOSOMAL DOMINANT;MENTAL RETARDATION,JOINT HYPERMOBILITY,SKIN LAXITY WITH OR WITHOUT METABOLIC ABNORMALITIES	BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALDH1A2	Diaphragmatic hernia 4, with cardiovascular defects, OMIM:620025	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALDH3A2	SJOEGREN-LARSSON SYNDROME	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALDH7A1	PYRIDOXINE-DEPENDENT EPILEPSY	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALDOA	GLYCOGEN STORAGE DISEASE XII	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALG1	ALG1-CDG	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALG12	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1G	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALG2	ALG2-CDG	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALG3	ALG3-CDG	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALG6	ALG6-CDG	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALG8	ALG8-CDG	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALG9	Gilissen-Kaesbach-Nishimura syndrome, 263210;AR lethal skeletal dysplasia ALG9-CDG;Congenital disorder of glycosylation, type II, 608776;NIHF hydops fetalis	BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5

ALMS1	ALSTROM SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALOX12B	Ichthyosis, congenital, autosomal recessive 2, 242100		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALOXE3	Ichthyosis, congenital, autosomal recessive 3, 606545		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALPL	HYPOPHOSPHATASIA		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALX1	FRONTONASAL DYSPLASIA TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALX3	FRONTONASAL DYSPLASIA TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALX4	FRONTONASAL DYSPLASIA 2;PARIETAL FORAMINA 2		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AMER1	OSTEOPATHIA STRIATA WITH CRANIAL SCLEROSIS		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
AMT	GLYCINE ENCEPHALOPATHY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ANAPC1	Rothmund-Thomson Syndrome Type 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ANKH	CHONDROCALCINOSIS 2;CRANIOMETAPHYSEAL DYSPLASIA JACKSON TYPE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ANKRD11	KBG SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ANTXR1	GAPO syndrome, OMIM:230740		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ANTXR2	Hyaline fibromatosis syndrome 228600		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AP1S2	Pettigrew syndrome, OMIM:304340		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
AP4B1	Spastic paraplegia 47, autosomal recessive, OMIM:614066;Hereditary spastic paraplegia 47, MONDO:0013551		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AP4E1	Spastic paraplegia 51, autosomal recessive, OMIM:613744;Hereditary spastic paraplegia 51, MONDO:0013401		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AP4S1	Spastic paraplegia 52, autosomal recessive, OMIM:614067		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AR	ANDROGEN INSENSITIVITY SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ARFGEF2	Periventricular heterotopia with microcephaly, OMIM:608097		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ARHGAP31	ADAMS-OLIVER SYNDROME 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ARID1A	COFFIN-SIRIS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ARID1B	MENTAL RETARDATION, AUTOSOMAL DOMINANT 12;COFFIN SIRIS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ARL13B	Joubert syndrome 8, 612291		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ARL6	BARDET-BIEDL SYNDROME TYPE 3;RETINITIS PIGMENTOSA TYPE 55		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ARSA	ARYLSULFATASE A DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ARSB	MUCOPOLYSACCHARIDOSIS TYPE 6		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ARSE	CHONDRODYSPLASIA PUNCTATA 1, X-LINKED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ARX	PARTINGTON SYNDROME;MENTAL RETARDATION X-LINKED ARX-RELATED;LISSENCEPHALY X-LINKED TYPE 2;AGENESIS OF THE CORPUS CALLOSUM WITH ABNORMAL GENITALIA;EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 1		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ASAH1	SPINAL MUSCULAR ATROPHY ASSOCIATED WITH PROGRESSIVE MYOCLONIC EPILEPSY;FARBER LIPOGRANULOMATOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ASCC1	spinal muscular atrophy;arthrogryposis;fetal akinesia;hypotonia;contractures		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ASNS	Asparagine synthetase deficiency 615574		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ASPA	CANAVAN DISEASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ASPM	Microcephaly 5, primary, autosomal recessive, OMIM:608716		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ASS1	CITRULLINEMIA TYPE I		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ASXL1	BOHRING-OPITZ SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ATIC	AICA-RIBOSURIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ATN1	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, OMIM:618494		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
ATP1A2	hydrops fetalis;microcephaly;arthrogryposis;extensive cortical malformations		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ATP6V0A2	Wrinkly skin syndrome 219200;Cutis laxa, autosomal recessive, type IIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ATP7A	MENKES DISEASE;OCCIPITAL HORN SYNDROME;SPINAL MUSCULAR ATROPHY, DISTAL, X-LINKED 3		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ATR	Seckel syndrome 1, OMIM:210600;Seckel syndrome 1, MONDO:0008869		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ATRX	MENTAL RETARDATION SYNDROMIC X-LINKED WITH HYPOTONIC FACIES SYNDROME TYPE 1;ALPHA-THALASSEMIA MENTAL RETARDATION SYNDROME X-LINKED NON-DELETION TYPE		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
B3GALT6	SPONDYLOEPIMETAPHYSEAL DYSPLASIA WITH JOINT LAXITY TYPE 1;EHLERS-DANLOS SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects 245600		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
B4GALT7	EHLERS-DANLOS SYNDROME PROGEROID TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
B9D2	Joubert syndrome 34, OMIM:614175;Meckel syndrome 10, OMIM:614175;Meckel syndrome, type 10, MONDO:0013609		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BBS1	Bardet-Biedl syndrome 1 OMIM:209900;Bardet-Biedl syndrome 1 MONDO:0008854		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BBS10	BARDET-BIEDL SYNDROME TYPE 10		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BBS12	BARDET-BIEDL SYNDROME TYPE 12		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BBS2	BARDET-BIEDL SYNDROME TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BBS4	BARDET-BIEDL SYNDROME TYPE 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BBS5	BARDET-BIEDL SYNDROME TYPE 5		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BBS7	BARDET-BIEDL SYNDROME TYPE 7		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BBS9	BARDET-BIEDL SYNDROME TYPE 9		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BCAP31	DEAFNESS, DYSTONIA, AND CENTRAL HYPOMYELINATION WITH DISORGANIZATION OF THE GOLGI APPARATUS		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
BCL11A	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
BCOR	MICROPHTHALMIA SYNDROMIC TYPE 2		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
BCS1L	GRACILE SYNDROME;GRACILE syndrome, 603358		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BFSP2	CATARACT AUTOSOMAL DOMINANT BFSP2-RELATED		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
BHLHA9	?Camptosynpolydactyly, complex, OMIM:607539;Syndactyly, mesoaxial synostotic, with phalangeal reduction, OMIM:609432;SPLIT HAND AND FOOT MALFORMATION;MESOAXIAL SYNOSTOTIC SYNDACTYLY WITH PHALANGEAL REDUCTION, MALIK-PERCIN TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BIN1	CENTRONUCLEAR MYOPATHY 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BLM	Bloom syndrome, OMIM:210900		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BMP1	Osteogenesis imperfecta type XIII 614856		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BMP2	Short stature, palatal anomalies, congenital heart disease, and skeletal malformations;Brachydactyly, type A2 112600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
BMP4	MICROPHTHALMIA, SYNDROMIC 6;OROFACIAL CLEFT 11		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
BMPER	DIAPHANOSPONDYLODYSOSTOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BMPR1B	Acromesomelic dysplasia, Demirhan type, OMIM:609441		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BNC2	Lower urinary tract obstruction, congenital, 618612		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
BRAF	CARDIOFACIOCUTANEOUS SYNDROME;LEOPARD SYNDROME TYPE 3;NOONAN SYNDROME TYPE 7		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
BRAT1	LETHAL NEONATAL RIGIDITY AND SEIZURE SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BRCA1	Fanconi anaemia, complementation group S, OMIM:617883		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BRCA2	FANCONI ANEMIA COMPLEMENTATION GROUP D TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BRIP1	Fanconi anemia, complementation group J, OMIM:609054		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BSND	BARTTER SYNDROME TYPE 4A		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BTD	BIOTINIDASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BUB1B	MOSAIC VARIEGATED ANEUPLOIDY SYNDROME 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
C12orf57	Temtamy syndrome, OMIM:218340		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
C12orf65	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 7		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
C5orf42	JOUBERT SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
C8orf37	CONE-ROD DYSTROPHY 16		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CA2	OSTEOPETROSIS AUTOSOMAL RECESSIVE TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CA8	CEREBELLAR ATAXIA MENTAL RETARDATION AND DYSEQUILIBRIUM SYNDROME TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CACNA1C	Timothy syndrome, OMIM:601005;Timothy syndrome, MONDO:0010979;Long QT syndrome 8, OMIM:618447;long qt syndrome 8, MONDO:0032756;Brugada syndrome 3, OMIM:611875;Brugada syndrome 3, MONDO:0012742;CACNA1C-related disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CACNA1E	Developmental and Epileptic Encephalopathy with Contractures Macrocephaly and Dyskinesias;Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesia		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CACNA1G	Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CANT1	Epiphyseal dysplasia, multiple, 7, 617719;Desbuquois dysplasia 1, 251450		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CASK	MRX WITH/WITHOUT NYSTAGMUS;MENTAL RETARDATION X-LINKED CASK-RELATED;FG SYNDROME TYPE 4		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
CASR	Hypocalcemia, autosomal dominant, 601198;Hypocalciuric hypercalcemia, type I, 145980;Hyperparathyroidism, neonatal, 239200;Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
CBL	NOONAN SYNDROME-LIKE DISORDER WITH OR WITHOUT JUVENILE MEYLOMONOCYTIC LEUKEMIA		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CC2D2A	JOUBERT SYNDROME 9;COACH SYNDROME;MECKEL SYNDROME, TYPE 6		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, OMIM:235510		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CCDC103	PRIMARY CILIARY DYSKINESIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CCDC22	Ritscher-Schinzel syndrome 2, OMIM:300963		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
CCDC39	CILIARY DYSKINESIA, PRIMARY, 14		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CCDC40	CILIARY DYSKINESIA, PRIMARY, 15		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CCDC8	3-M syndrome 3, OMIM:614205;3M syndrome 3, MONDO:0013627		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CCDC88C	Hydrocephalus, congenital, 1, OMIM:236600;Hydrocephalus, nonsyndromic, autosomal recessive 1, MONDO:0009360		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CDAN1	Dyserythropoietic anemia, congenital, type Ia, OMIM:224120;Anemia, congenital dyserythropoietic, type 1a, MONDO:0009135		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CDC6	Meier-Gorlin syndrome 5, OMIM:613805		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CDH1	Blepharo-cheiro-dontic syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CDH3	EEM SYNDROME;HYPOTRICHOSIS, CONGENITAL, WITH JUVENILE MACULAR DYSTROPHY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CDK5RAP2	Microcephaly 3, primary, autosomal recessive, OMIM:604804;Microcephaly 3, primary, autosomal recessive, MONDO:0011488		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CDKL5	EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 2		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
CDKN1C	BECKWITH-WIEDEMANN SYNDROME;IMAGe Syndrome		MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)	Fetal anomalies			5.5
CDON	HOLOPROSENCEPHALY 11		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CDT1	Meier-Gorlin syndrome 4, OMIM:613804		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CENPJ	MICROCEPHALY PRIMARY TYPE 6;SECKEL SYNDROME TYPE 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CEP135	Microcephaly 8, primary, autosomal recessive, OMIM:614673;Microcephaly 8, primary, autosomal recessive, MONDO:0013849		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CEP152	MICROCEPHALY PRIMARY TYPE 4;SECKEL SYNDROME TYPE 5		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CEP290	JOUBERT SYNDROME TYPE 5;LEBER CONGENITAL AMAUROSIS TYPE 10;SENIOR-LOKEN SYNDROME TYPE 6;BARDET-BIEDL SYNDROME TYPE 14;MECKEL SYNDROME TYPE 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CEP41	JOUBERT SYNDROME 15		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CEP57	MOSAIC VARIEGATED ANEUPLOIDY SYNDROME 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CEP63	?Seckel syndrome 6, OMIM:614728;Seckel syndrome 6, MONDO:0013871		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CEP85L	Lissencephaly 10, posterior predominant, OMIM:618873		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
CFC1	CFC1-RELATED CONOTRUNCAL HEART MALFORMATIONS;Heterotaxy, visceral, 2, autosomal, 605376		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CFL2	Nemaline myopathy 7, autosomal recessive, OMIM:610687;Nemaline myopathy 7, MONDO:0012538		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CFTR	Cystic fibrosis 219700		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHAT	Myasthenic syndrome, congenital, 6, presynaptic 254210		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHD7	KALLMANN SYNDROME TYPE 5;IDIOPATHIC HYPOGONADOTROPIC HYPOGONADISM;CHARGE SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CHKB	Muscular dystrophy, congenital, megaconial type 602541		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHRNA1	MULTIPLE PTERYGIUM SYNDROME LETHAL TYPE;Multiple pterygium syndrome, lethal type, 253290		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHRNA3	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHRNB1	Myasthenic syndrome, congenital, 2A, slow-channel, 616313;?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHRND	Several associated, probably most relevant is lethal multiple pterygium syndrome 253290		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHRNE	Myasthenic syndrome, congenital, 4A, slow-channel, OMIM:605809;Myasthenic syndrome, congenital, 4B, fast-channel, OMIM:616324;Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, OMIM:608931		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHRNG	MULTIPLE PTERYGIUM SYNDROME ESCOBAR VARIANT		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHST14	EHLERS-DANLOS SYNDROME MUSCULOCONTRACTURAL TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHST3	SPONDYLOEPIPHYSEAL DYSPLASIA WITH CONGENITAL JOINT DISLOCATIONS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHSY1	TEMTAMY PREAXIAL BRACHYDACTYLY SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHUK	Cocoon syndrome, OMIM:613630;Popliteal pterygium syndrome, Bartsocas-Papas type 2, OMIM:619339		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CLCN7	CLCN7-RELATED OSTEOPETROSIS;Osteopetrosis, autosomal recessive 4, OMIM:611490;Osteopetrosis, autosomal dominant 2, OMIM:166600;Hypopigmentation, organomegaly, and delayed myelination and development, OMIM:618541		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
CNOT3	CNOT3 syndrome;Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CNTNAP2	CORTICAL DYSPLASIA-FOCAL EPILEPSY SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COG1	COG1-CDG		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COG4	COG4-CDG;Saul-Wilson syndrome, 618150		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COG5	Congenital disorder of glycosylation, type III, OMIM:613612;COG5-CDG, MONDO:0013325		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COG6	Congenital disorder of glycosylation, type III, 614576;Shaheen syndrome, 615328		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COG7	COG7-CDG		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COG8	COG8-CDG		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL10A1	SCHMID TYPE METAPHYSEAL CHONDRODYSPLASIA		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
COL11A1	FIBROCHONDROGENESIS;STICKLER SYNDROME, TYPE II		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL11A2	WEISSENBACHER-ZWEYMUELLER SYNDROME;DEAFNESS AUTOSOMAL RECESSIVE TYPE 53;AUTOSOMAL RECESSIVE OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA;DEAFNESS AUTOSOMAL DOMINANT TYPE 13;STICKLER SYNDROME TYPE 3		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL12A1	Bethlem myopathy 2, 616471;?Ullrich congenital muscular dystrophy 2, 616470		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL18A1	Knobloch syndrome, type 1, OMIM:267750		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL1A1	COL1A1/2-RELATED OSTEOGENESIS IMPERFECTA;EHLERS-DANLOS SYNDROME, CLASSIC TYPE, COL1A1-RELATED;OSTEOGENESIS IMPERFECTA TYPE III;EHLERS-DANLOS SYNDROME TYPE VIIA;OSTEOGENESIS IMPERFECTA TYPE IIA;CAFFEY DISEASE;OSTEOGENESIS IMPERFECTA TYPE I		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
COL1A2	Ehlers-Danlos syndrome;Osteogenesis imperfecta		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
COL2A1	SPONDYLOEPIMETAPHYSEAL DYSPLASIA STRUDWICK TYPE;SPONDYLOPERIPHERAL DYSPLASIA;STICKLER SYNDROME TYPE 1 NON-SYNDROMIC OCULAR;PLATYSPONDYLIC LETHAL SKELETAL DYSPLASIA TORRANCE TYPE;ACHONDROGENESIS TYPE 2;RHEGMATOGENOUS RETINAL DETACHMENT AUTOSOMAL DOMINANT;SPONDYLOEPIPHYSEAL DYS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
COL3A1	HP:0002126;HP:0001883;HP:0006496		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL4A1	PORENCEPHALY 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
COL4A2	PORENCEPHALY 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
COL6A1	Bethlem myopathy 1, OMIM:158810;Ullrich congenital muscular dystrophy 1, OMIM:254090		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL6A2	Ullrich congenital muscular dystrophy 1 254090;Bethlem myopathy 1 158810		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL6A3	Bethlem myopathy, OMIM:158810;Ullrich congenital muscular dystrophy, OMIM:254090		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL9A1	STICKLER SYNDROME TYPE 4;MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 6		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL9A2	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 2;STICKLER SYNDROME, TYPE V		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
COLEC11	3MC SYNDROME 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COLQ	Myasthenic syndrome, congenital, 5, 603034		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COQ4	COENZYME Q10 DEFICIENCY, PRIMARY, 7		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COQ9	COENZYME Q10 DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CPT2	CPT II deficiency, infantile, OMIM:600649;CPT II deficiency, lethal neonatal, OMIM:608836		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CRADD	Mental retardation, autosomal recessive 34, with variant lissencephaly, OMIM:614499		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CREBBP	CREBBP intellectual disability without typical RTS features;RUBINSTEIN-TAYBI SYNDROME TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CRLF1	Cold-induced sweating syndrome 1 272430		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CRTAP	Osteogenesis imperfecta, type VII 610682		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CRYAA	CATARACT, AUTOSOMAL RECESSIVE CONGENITAL 1;CATARACT, NUCLEAR		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
CRYBA1	CATARACT CONGENITAL ZONULAR WITH SUTURAL OPACITIES		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CRYBA4	CATARACT ZONULAR TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CRYBB1	CATARACT 17, MULTIPLE TYPES;CATARACT 17, MULTIPLE TYPES, MONOALLELIC;CATARACT, CONGENITAL NUCLEAR, AUTOSOMAL RECESSIVE 3		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
CRYBB2	CATARACT, COPPOCK-LIKE;CATARACT, CONGENITAL, CERULEAN TYPE, 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CRYBB3	Cataract 22, OMIM:609741		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
CRYGC	CATARACT AUTOSOMAL DOMINANT		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CRYGD	CATARACT AUTOSOMAL DOMINANT;CATARACT CONGENITAL CERULEAN TYPE 3		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CSF1R	Brain abnormalities, neurodegeneration, and dysosteosclerosis, OMIM:618476;BANDDOS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CTC1	CEREBRORETINAL MICROANGIOPATHY WITH CALCIFICATIONS AND CYSTS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5

CTNNB1	MENTAL RETARDATION, AUTOSOMAL DOMINANT 19		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CTNND1	Blepharo-cheiro-dontic syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CTSA	GALACTOSIALIDOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CTSD	NEURONAL CEROID LIPOFUSCINOSIS TYPE 10		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CTSK	PYCNODYSOSTOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CUL4B	MENTAL RETARDATION SYNDROMIC X-LINKED CABEZAS TYPE		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
CUL7	3-M SYNDROME 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CYBB	Chronic granulomatous disease, X-linked, OMIM:306400		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, OMIM:613743		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, OMIM:202010		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CYP17A1	17-alpha-hydroxylase/17,20-lyase deficiency;17,20-lyase deficiency, isolated		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CYP1B1	PRIMARY CONGENITAL GLAUCOMA TYPE 3A		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CYP21A2	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency;Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies,614416		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CYP4F22	Ichthyosis, congenital, autosomal recessive 5, 604777		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DAG1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C7		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, OMIM:611105		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DCC	Mirror movements 1 and/or agenesis of the corpus callosum, OMIM:157600;Gaze palsy, familial horizontal, with progressive scoliosis, 2, OMIM:617542		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
DCX	Lissencephaly, X-linked, OMIM:300067;Subcortical laminal heterotopia, X-linked, OMIM:300067		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
DDR2	SPONDYLOEPIMETAPHYSEAL DYSPLASIA SHORT LIMB-HAND TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DDX11	WARSAW BREAKAGE SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DHCR24	DESMOSTEROLOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DHCR7	SMITH-LEMLI-OPITZ SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DHFR	MEGALOBLASTIC ANEMIA DUE TO DIHYDROFOLATE REDUCTASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DHODH	POSTAXIAL ACROFACIAL DYSOSTOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DIAPH1	Seizures, cortical blindness, microcephaly syndrome, 616632		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DIS3L2	PERLMAN SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DISP1	Holoprosencephaly		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
DKC1	DYSKERATOSIS CONGENITA, X-LINKED;DKC1-RELATED DYSKERATOSIS CONGENITA		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
DLL1	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, OMIM:618709		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
DLL3	SPONDYLOCOSTAL DYSOSTOSIS TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DLX5	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600;Split-hand/foot malformation 1, 183600		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNAAF1	Primary ciliary dyskinesia 613193		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNAAF2	Ciliary dyskinesia, primary, 10, 612518		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNAAF3	PRIMARY CILIARY DYSKINEASIA;Ciliary dyskinesia, primary, 2, MIM:606763		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNAH11	Primary ciliary dyskinesia 611884		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNAH5	CILIARY DYSKINESIA, PRIMARY, 3;Primary ciliary dyskinesia 608644;heterotaxy		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNAH9	Motile Cilia Defects and Situs Inversus		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNAI1	Primary ciliary dyskinesia 244400		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus,612444		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNAL1	Ciliary dyskinesia, primary, 16, 614017		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNM1L	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNM2	Lethal congenital contracture syndrome 5, 615368		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Fetal anomalies			5.5
DNMT3A	OVERGROWTH SYNDROME WITH INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
DNMT3B	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DOCK6	ADAMS-OLIVER SYNDROME 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DOK7	?Fetal akinesia deformation sequence 3, 618389;Myasthenic syndrome, congenital, 10, 254300		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DOLK	CONGENITAL DISORDERS OF GLYCOSYLATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DPAGT1	MYASTHENIC SYNDROME, CONGENITAL, WITH TUBULAR AGGREGATES 2;DPAGT1-CDG		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DPM1	CONGENITAL DISORDERS OF GLYCOSYLATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DPM3	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DSP	Epidermolysis bullosa, lethal acantholytic, OMIM:609638 (AR);Skin fragility-woolly hair syndrome, OMIM:607655 (AR);Keratosis palmoplantaris striata II, OMIM:612908 (AD);Cardiomyopathy, dilated, with woolly hair and keratoderma, OMIM:605676 (AR);Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, OMIM:615821 (AD);Arrhythmogenic right ventricular		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
DYM	SMITH-MCCORT DYSPLASIA;DYGGVE-MELCHIOR-CLAUSEN SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DYNC1H1	SEVERE ID WITH NEURONAL MIGRATION DISORDER;SPINAL MUSCULAR ATROPHY, LOWER EXTREMITY-PREDOMINANT, AD		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
DYNC2H1	SHORT RIB-POLYDACTYLY SYNDROME TYPE 3;ASPHYXIATING THORACIC DYSTROPHY TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DYRK1A	MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 7		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
EARS2	Combined oxidative phosphorylation deficiency 12, OMIM:614924		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EBP	CHONDRODYSPLASIA PUNCTATA 2, X-LINKED		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
EDNRA	MANDIBULOFACIAL DYSOSTOSIS WITH ALOPECIA		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
EDNRB	ABCD SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EFEMP2	Cutis laxa, autosomal recessive, type IB, OMIM:614437		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EFNB1	Craniofrontonasal dysplasia, OMIM:304110		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
EFTUD2	MANDIBULOFACIAL DYSOSTOSIS WITH MICROCEPHALY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
EHMT1	9Q SUBTELOMERIC DELETION SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
EIF2AK3	WOLCOTT-RALLISON SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EIF2B2	Leukoencephalopathy with vanishing white matter, 603896		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EIF2B3	vanishing white matter disease 603896		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ELAC2	INFANTILE HYPERTROPHIC CARDIOMYOPATHY, LACTIC ACIDOSIS, AND ISOLATED COMPLEX I DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ELN	ELN-RELATED CUTIS LAXA;SUPRAVALVAR AORTIC STENOSIS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ELOVL4	ICHTHYOSIS, SPASTIC QUADRIPLEGIA, AND MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked 310300		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
EMX2	Schizencephaly, 269160		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ENPP1	Arterial calcification, generalized, of infancy, 1, OMIM:208000		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EP300	RUBINSTEIN-TAYBI SYNDROME TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ERBB3	Lethal congenital contractural syndrome 2, OMIM:607598		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ERCC1	CEREBROOCULOFACIOSKELETAL SYNDROME 4;FANCONI ANEMIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ERCC2	XERODERMA PIGMENTOSUM COMPLEMENTATION GROUP D;CEREBRO-OCULO-FACIO-SKELETAL SYNDROME TYPE 2;TRICHOTHIODYSTROPHY PHOTOSENSITIVE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ERCC3	XERODERMA PIGMENTOSUM COMPLEMENTATION GROUP B;TRICHOTHIODYSTROPHY PHOTOSENSITIVE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ERCC4	XFE PROGEROID SYNDROME;FANCONI ANEMIA, COMPLEMENTATION GROUP Q;PRIMORDIAL DWARFISM;XERODERMA PIGMENTOSUM, GROUP F;Xeroderma pigmentosum, group F, 278760		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ERCC5	Cerebrooculofacioskeletal syndrome 3, OMIM:616570;Cerebrooculofacioskeletal syndrome 3, MONDO:0014696		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ERCC6	UV-SENSITIVE SYNDROME;CEREBRO-OCULO-FACIO-SKELETAL SYNDROME TYPE 1;COCKAYNE SYNDROME TYPE B;DE SANCTIS-CACCHIONE SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ERCC8	COCKAYNE SYNDROME TYPE A		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ESCO2	SC PHOCOMELIA SYNDROME;ROBERTS SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ETFA	Glutaric acidemia IIA, OMIM:231680;multiple acyl-CoA dehydrogenase deficiency, MONDO:0009282		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ETFB	Glutaric acidemia IIB, OMIM:231680;multiple acyl-CoA dehydrogenase deficiency, MONDO:0009282		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ETFDH	GLUTARIC ACIDURIA TYPE 2C		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EVC	ELLIS-VAN CREVELD SYNDROME;ACROFACIAL DYSOSTOSIS WEYERS TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EVC2	ELLIS-VAN CREVELD SYNDROME;ACROFACIAL DYSOSTOSIS WEYERS TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EXOSC3	Pontocerebellar hypoplasia, type 1B, OMIM:614678		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EXT1	HEREDITARY MULTIPLE EXOSTOSES TYPE 1;TRICHO-RHINO-PHALANGEAL SYNDROME TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
EXT2	EXOSTOSES, MULTIPLE, TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities, OMIM:617425		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EYA1	BRANCHIOOTORENAL SYNDROME TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
EZH2	WEAVER SYNDROME 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FAH	TYROSINEMIA TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FAM126A	LEUKODYSTROPHY HYPOMYELINATING TYPE 5		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FAM20A	AMELOGENESIS IMPERFECTA AND GINGIVAL FIBROMATOSIS SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FAM20C	RAINE SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FAM58A	STAR SYNDROME		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
FANCA	FANCONI ANEMIA, COMPLEMENTATION GROUP A		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FANCB	FANCB-RELATED FANCONI ANEMIA		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
FANCC	FANCONI ANEMIA, COMPLEMENTATION GROUP C		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FANCD2	FANCONI ANEMIA, COMPLEMENTATION GROUP D2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FANCE	FANCONI ANEMIA, COMPLEMENTATION GROUP E		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FANCF	FANCONI ANEMIA, COMPLEMENTATION GROUP F		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FANCG	FANCONI ANEMIA, COMPLEMENTATION GROUP G		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FANCI	FANCI-RELATED FANCONI ANEMIA;FANCONI ANEMIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FANCL	Fanconi anemia, complementation group L, OMIM:614083;Fanconi anemia complementation group L, MONDO:0013566		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FBLN5	?Cutis laxa, autosomal dominant 2, OMIM:614434;Cutis laxa, autosomal recessive, type IA, OMIM:219100		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FBN1	SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME;MARFAN SYNDROME;MASS SYNDROME/OVERLAP CONNECTIVE TISSUE DISEASE		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
FBN2	Contractural arachnodactyly, congenital OMIM:121050;congenital contractural arachnodactyly MONDO:0007363		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
FGD1	AARSKOG-SCOTT SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
FGF10	LADD SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FGF3	DEAFNESS WITH LABYRINTHINE APLASIA, MICROTIA AND MICRODONTIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FGF8	Hypogonadotropic hypogonadism 6 with or without anosmia 612702		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FGFR1	IDIOPATHIC HYPOGONADOTROPIC HYPOGONADISM;OSTEOGLOPHONIC DYSPLASIA;PFEIFFER SYNDROME;KALLMANN SYNDROME TYPE 2;Hartsfield syndrome;Encephalocraniocutaneous lipomatosis		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FGFR2	BEARE-STEVENSON CUTIS GYRATA SYNDROME;LACRIMO-AURICULO-DENTO-DIGITAL SYNDROME;JACKSON-WEISS SYNDROME;ACROCEPHALOSYNDACTYLY TYPE V;FAMILIAL SCAPHOCEPHALY SYNDROME;ANTLEY-BIXLER SYNDROME;CROUZON SYNDROME;APERT SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FGFR3	LACRIMO-AURICULO-DENTO-DIGITAL SYNDROME;MUENKE SYNDROME;ACHONDROPLASIA;CROUZON SYNDROME WITH ACANTHOSIS NIGRICANS;CAMPTODACTYLY TALL STATURE AND HEARING LOSS SYNDROME;HYPOCHONDROPLASIA;THANATOPHORIC DYSPLASIA TYPE 2;THANATOPHORIC DYSPLASIA TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FH	Fumarase deficiency, OMIM:606812		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FIG4	Yunis-Varon syndrome, OMIM:216340;Yunis-Varon syndrome, MONDO:0008995;Charcot-Marie-Tooth disease, type 4J, OMIM:611228;Charcot-Marie-Tooth disease type 4J, MONDO:0012640;?Polymicrogyria, bilateral temporooccipital, OMIM:612691;Bilateral parasagittal parieto-occipital polymicrogyria, MONDO:0012986		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FKBP10	Bruck syndrome 1, OMIM:259450;Bruck syndrome 1, MONDO:0009806;Osteogenesis imperfecta, type XI, OMIM:610968;Osteogenesis imperfecta type 11, MONDO:0012592		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FKBP14	EHLERS-DANLOS SYNDROME WITH PROGRESSIVE KYPHOSCOLIOSIS, MYOPATHY, AND HEARING LOSS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FKRP	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH OR WITHOUT MENTAL RETARDATION TYPE B5;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C5;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH BRAIN AND EYE ANOMALIES TYPE A5		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FKTN	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITHOUT MENTAL RETARDATION TYPE B4;CARDIOMYOPATHY DILATED TYPE 1X;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C4;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH BRAIN AND EYE ANOMALIES TYPE A4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FLNA	TERMINAL OSSEOUS DYSPLASIA;OTOPALATODIGITAL SYNDROME TYPE 1;EPILEPTIC ENCEPHALOPATHY;MELNICK-NEEDLES SYNDROME;PERIVENTRICULAR NODULAR HETEROTOPIA TYPE 1;FG SYNDROME TYPE 2;X-LINKED CONGENITAL IDIOPATHIC INTESTINAL PSEUDOOBSTRUCTION;OTOPALATODIGITAL SYNDROME TYPE 2;FRONTOMETAPHYSEAL		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
FLNB	ATELOSTEOGENESIS TYPE 1;BOOMERANG DYSPLASIA;SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME;AUTOSOMAL DOMINANT LARSEN SYNDROME;ATELOSTEOGENESIS TYPE 3		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
FLNC	Arthrogryposis, MONDO:0008779		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
FLT4	MILROY DISEASE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FLVCR2	PROLIFERATIVE VASCULOPATHY AND HYDRAENCEPHALY-HYDROCEPHALY SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FOLR1	NEURODEGENERATION DUE TO CEREBRAL FOLATE TRANSPORT DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FOXC1	IRIDOGONIODYSGENESIS ANOMALY;AXENFELD-RIEGER SYNDROME TYPE 3;PETERS ANOMALY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FOXC2	LYMPHEDEMA-DISTICHIASIS SYNDROME;HEREDITARY LYMPHEDEMA II		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FOXE1	BAMFORTH-LAZARUS SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FOXE3	Anterior segment dysgenesis 2, multiple subtypes, OMIM:610256;Cataract 34, multiple types, OMIM:612968;{Aortic aneurysm, familial thoracic 11, susceptibility to}, OMIM:617349 CONGENITAL PRIMARY APHAKIA;ANTERIOR SEGMENT MESENCHYMAL DYSGENESIS		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
FOXF1	ALVEOLAR CAPILLARY DYSPLASIA WITH MISALIGNMENT OF PULMONARY VEINS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FOXG1	Rett Syndrome, congenital variant OMIM:613454;Rett syndrome, congenital variant MONDO:0013270		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FOXP3	IPEX SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
FOXRED1	MITOCHONDRIAL COMPLEX I DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FRA10AC1	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, OMIM:620113		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FRAS1	FRASER SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FREM1	MANITOBA OCULOTRICHOANAL SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FREM2	FRASER SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FTL	HEREDITARY HYPERFERRITINEMIA-CATARACT SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FUT8	Congenital disorder of glycosylation with defective fucosylation 1, OMIM:618005;Congenital disorder of glycosylation with defective fucosylation 1, MONDO:0020775		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FYCO1	CATARACT, AUTOSOMAL RECESSIVE CONGENITAL 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
G6PC3	Dursun syndrome;Neutropenia, severe congenital 4, autosomal recessive		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GAA	GLYCOGEN STORAGE DISEASE TYPE II		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GALC	Krabbe disease, OMIM:245200		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GALE	EPIMERASE-DEFICIENCY GALACTOSEMIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GALK1	GALACTOSEMIA II		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GALNS	MUCOPOLYSACCHARIDOSIS TYPE 4A		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GALNT2	Congenital disorder of glycosylation, type IIt OMIM:618885		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GATA1	Anaemia, X-linked, with/without neutropaenia and/or platelet abnormalities, OMIM:300835;Hemolytic anemia due to elevated adenosine deaminase, OMIM:301083		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
GATA2	EMBERGER SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, OMIM:146255;Hypoparathyroidism-deafness-renal disease syndrome, MONDO:0007797		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GATA4	ATRIAL SEPTAL DEFECT TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GATA6	ATRIAL SEPTAL DEFECT 9;PANCREATIC AGENESIS, DIAPHRAGMATIC HERNIA AND CONGENITAL HEART DEFECTS;ATRIOVENTRICULAR SEPTAL DEFECT 5		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GBA	GAUCHER DISEASE TYPE 3C;GAUCHER DISEASE TYPE 1;GAUCHER DISEASE PERINATAL LETHAL;GAUCHER DISEASE TYPE 3;GAUCHER DISEASE;GAUCHER DISEASE TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GBE1	Glycogen storage disease IV, OMIM:232500;Fetal akinesia deformation sequence		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GCDH	GLUTARICACIDEMIA TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GDF1	Congenital heart defects, multiple types, 6, OMIM:613854;Right atrial isomerism (Ivemark), OMIM:208530		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
GDF5	BRACHYDACTYLY TYPE C;SYMPHALANGISM PROXIMAL SYNDROME;DU PAN SYNDROME;BRACHYDACTYLY TYPE A1;ACROMESOMELIC CHONDRODYSPLASIA HUNTER-THOMPSON TYPE;ACROMESOMELIC CHONDRODYSPLASIA GREBE TYPE;BRACHYDACTYLY TYPE A2;MULTIPLE SYNOSTOSES SYNDROME TYPE 2		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
GDF6	MICROPHTHALMIA ISOLATED TYPE 4;KLIPPEL-FEIL SYNDROME TYPE 1;Syndromic CAKUT		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GFAP	ALEXANDER DISEASE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GFM1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GFPT1	Myasthenia, congenital, 12, with tubular aggregates, OMIM:610542;Congenital myasthenic syndrome 12, MONDO:0012518		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GFRA1	Renal agenesis		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GHR	Growth hormone insensitivity, partial, OMIM:604271;Laron dwarfism, OMIM:262500		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GJA1	AUTOSOMAL DOMINANT OCULODENTODIGITAL DYSPLASIA;HYPOPLASTIC LEFT HEART SYNDROME;AUTOSOMAL RECESSIVE OCULODENTODIGITAL DYSPLASIA;HALLERMANN-STREIFF SYNDROME		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
GJA3	CATARACT ZONULAR PULVERULENT CATARACT TYPE 3		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GJA8	CATARACT-MICROCORNEA SYNDROME;CATARACT ZONULAR PULVERULENT TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GJC2	LYMPHEDEMA, HEREDITARY, IC;LEUKODYSTROPHY, HYPOMYELINATING, 2;SPASTIC PARAPLEGIA, 44		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
GLA	Fabry disease, 301500		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
GLB1	GM1-GANGLIOSIDOSIS TYPE 1;GM1-GANGLIOSIDOSIS TYPE 2;GM1-GANGLIOSIDOSIS TYPE 3;MUCOPOLYSACCHARIDOSIS TYPE 4B		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GLDC	GLDC-RELATED GLYCINE ENCEPHALOPATHY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GLE1	ARTHROGRYPOSIS, LETHAL, WITH ANTERIOR HORN CELL DISEASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5

GLI1	Polydactyly, postaxial, type A8, OMIM:618123;Polydactyly, postaxial, type A8, MONDO:0029130;Polydactyly, preaxial I, OMIM:174400;Preaxial polydactyly of fingers, MONDO:0017425		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GLI2	GLI2-RELATED HOLOPROSENCEPHALY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GLI3	Greig cephalopolysyndactyly syndrome, OMIM:175700;Pallister-Hall syndrome, OMIM:146510;Polydactyly, postaxial, types A1 and B, OMIM:174200;Polydactyly, preaxial, type IV, OMIM:174700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GLIS3	DIABETES MELLITUS NEONATAL WITH CONGENITAL HYPOTHYROIDISM		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GLUL	CONGENITAL SYSTEMIC GLUTAMINE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GNAS	Pseudohypoparathyroidism Ia, OMIM:103580;pseudohypoparathyroidism type 1A, MONDO:0007078;Pseudohypoparathyroidism Ib, OMIM:603233;pseudohypoparathyroidism type 1B, MONDO:0011301;Pseudohypoparathyroidism Ic, OMIM:612462;pseudohypoparathyroidism type 1C, MONDO:0012911;McCune-Albright syndrome, somatic, mosaic, OMIM:174800;panostotic fibrou		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GNPAT	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GNPTAB	MUCOLIPIDOSIS TYPE II;MUCOLIPIDOSIS TYPE III COMPLEMENTATION GROUP A		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GNPTG	MUCOLIPIDOSIS TYPE III COMPLEMENTATION GROUP C		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GNS	MUCOPOLYSACCHARIDOSIS TYPE 3D		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GORAB	Geroderma osteodysplasticum		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GPC3	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 1		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
GPC6	Omodysplasia 1, OMIM:258315;Autosomal recessive omodysplasia, MONDO:0009779		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency 613470		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GPSM2	CHUDLEY-MCCULLOUGH SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GPX4	Spondylometaphyseal dysplasia, Sedaghatian type, OMIM:250220		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant OMIM:614254;intellectual disability, autosomal dominant 8 MONDO:0013655;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive OMIM:617820;neurodevelopmental disorder with or without hyperkinetic movements and seizu		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Fetal anomalies			5.5
GRIN2B	Intellectual developmental disorder, autosomal dominant 6, with or without seizures, OMIM:613970;Developmental and epileptic encephalopathy 27, OMIM:616139		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GRIP1	Fraser syndrome 219000		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GRM7	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, OMIM:618922		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GSC	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, OMIM:602471;Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome, MONDO:0011227		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GTF2H5	TRICHOTHIODYSTROPHY PHOTOSENSITIVE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GUCY2C	MECONIUM ILEUS;FAMILIAL DIARRHEA DIARRHEA 6		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
GUSB	MUCOPOLYSACCHARIDOSIS TYPE 7		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HADHA	LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HADHB	Trifunctional protein deficiency, OMIM:609015;Mitochondrial trifunctional protein deficiency, MONDO:0012172		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HBA1	Thalassemias, alpha-, OMIM:604131;Fatal hydrops fetalis;Hb Bart syndrome		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HBA2	Thalassemias, alpha-, OMIM:604131;Fatal hydrops fetalis;Hb Bart syndrome		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HCCS	MICROPHTHALMIA SYNDROMIC TYPE 7		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
HDAC8	WILSON-TURNER SYNDROME;CORNELIA DE LANGE-LIKE SYNDROME		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
HES7	Spondylocostal dysostosis 4, autosomal recessive 613686		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HESX1	Septooptic dysplasia, OMIM:182230;Septooptic dysplasia, MONDO:0008428		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
HIBCH	HIBCH DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HK1	Hemolytic anemia due to hexokinase deficiency, OMIM:235700;Neurodevelopmental disorder with visual defects and brain anomalies, OMIM:618547		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Fetal anomalies			5.5
HMGA2	Silver-Russell syndrome 5, OMIM:618908;Silver-Russell syndrome 5, MONDO:0020795		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
HMX1	Oculoauricular syndrome, OMIM:612109		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HNF1B	RENAL CYSTS AND DIABETES SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
HNF4A	ATYPICAL DOMINANT FANCONI SYNDROME WITH MODY;HNF4A-RELATED MATURITY-ONSET DIABETES OF THE YOUNG TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
HOXA1	BOSLEY-SALIH-ALORAINY SYNDROME;ATHABASKAN BRAINSTEM DYSGENESIS SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HOXA13	HAND-FOOT-GENITAL SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
HOXD13	SYNPOLYDACTYLY 1;BRACHYDACTYLY TYPE D;BRACHYDACTYLY-SYNDACTYLY SYNDROME;BRACHYDACTYLY TYPE E;VACTERL ASSOCIATION;SYNDACTYLY TYPE 5		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
HPSE2	UROFACIAL SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HR	ATRICHIA WITH PAPULAR LESIONS;ALOPECIA UNIVERSALIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HRAS	COSTELLO SYNDROME;CONGENITAL MYOPATHY WITH EXCESS OF MUSCLE SPINDLES		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
HSD17B3	Pseudohermaphroditism, male, with gynecomastia 264300		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HSD17B4	D-BIFUNCTIONAL PROTEIN DEFICIENCY;PERRAULT SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HSF4	Cataract 5, multiple types, OMIM:116800		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
HSPA9	Anemia, sideroblastic, 4, OMIM:182170;Even-plus syndrome, OMIM:616854		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HSPD1	LEUKODYSTROPHY HYPOMYELINATING TYPE 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HSPG2	Schwartz-Jampel syndrome, type 1, OMIM:255800;Schwartz-Jampel syndrome, MONDO:0009717;Dyssegmental dysplasia, Silverman-Handmaker type, OMIM:224410;Silverman-Handmaker type dyssegmental dysplasia, MONDO:0009140		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HUWE1	MENTAL RETARDATION SYNDROMIC X-LINKED TURNER TYPE		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
HYLS1	HYDROLETHALUS SYNDROME TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ICK	Endocrine-cerebroosteodysplasia, OMIM:612651;Endocrine-cerebro-osteodysplasia syndrome, MONDO:0012980		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IDH1	Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria 614875;Maffucci syndrome 614569;Ollier disease/ Dyschondroplasia 166000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
IDS	MUCOPOLYSACCHARIDOSIS TYPE 2		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
IDUA	MUCOPOLYSACCHARIDOSIS TYPE 1H;MUCOPOLYSACCHARIDOSIS TYPE 1H/S;MUCOPOLYSACCHARIDOSIS TYPE 1S		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome 614231		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IFIH1	Aicardi-Goutieres syndrome 7, OMIM:615846;Singleton-Merten syndrome 1, OMIM:182250		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
IFITM5	OSTEOGENESIS IMPERFECTA TYPE V		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
IFT122	Cranioectodermal dysplasia type 1 OMIM:218330;cranioectodermal dysplasia 1 MONDO:0021093		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IFT140	MAINZER-SALDINO SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IFT43	CRANIOECTODERMAL DYSPLASIA TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IFT80	ASPHYXIATING THORACIC DYSTROPHY 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IGF1	Insulin-like growth factor I deficiency, OMIM:608747		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IGF1R	Insulin-like growth factor I, resistance to, OMIM:270450		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
IGF2	CHROMOSOME 11P15.5-RELATED RUSSELL-SILVER SYNDROME;BECKWITH-WIEDEMANN SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)	Fetal anomalies			5.5
IGHMBP2	SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IHH	Acrocapitofemoral dysplasia, OMIM:607778;Brachydactyly, type A1, OMIM:112500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
IKBKG	SUSCEPTIBILITY TO X-LINKED FAMILIAL ATYPICAL MICOBACTERIOSIS TYPE 1;INCONTINENTIA PIGMENTI;ECTODERMAL DYSPLASIA ANHIDROTIC WITH IMMUNODEFICIENCY-OSTEOPETROSIS-LYMPHEDEMA;ECTODERMAL DYSPLASIA ANHIDROTIC WITH IMMUNODEFICIENCY X-LINKED;IMMUNODEFICIENCY NEMO-RELATED WITHOUT ANHIDROTIC ECTOD		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
IL11RA	Autosomal Recessive Craniosynostosis;Crouzon-like craniosynostosis;Craniosynostosis and dental anomalies, 614188		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IL1RAPL1	MENTAL RETARDATION X-LINKED TYPE 21		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
IMPAD1	CHONDRODYSPLASIA WITH JOINT DISLOCATIONS, GRAPP TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
INSR	DONOHUE SYNDROME 246200;Hyperinsulinemic hypoglycemia, familial, 5 609968;Diabetes mellitus, insulin-resistant, with acanthosis nigricans 610549;Rabson-Mendenhall syndrome 262190		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
INVS	Nephronophthisis 2 602088		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IQCB1	Senior-Loken syndrome 5 609254		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IRF6	POPLITEAL PTERYGIUM SYNDROME;VAN DER WOUDE SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
IRX5	Hamamy syndrome, OMIM:611174		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ISPD	WALKER WARBURG SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ITGA3	INTERSTITIAL LUNG DISEASE, NEPHROTIC SYNDROME, AND EPIDERMOLYSIS BULLOSA, CONGENITAL		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ITGA6	Epidermolysis Bullosa with Pyloric Atresia. 226730		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ITGB4	Epidermolysis Bullosa with Pyloric Atresia. 226730		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
JAG1	ALAGILLE SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
JAM3	Haemorrhagic destruction of the brain, subependymal calcification, and cataracts, OMIM:613730		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KANSL1	CHROMOSOME 17Q21.31 MICRODELETION SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KAT6B	BLEPHAROPHIMOSIS/INTELLECTUAL DISABILITY PHENOTYPE WHICH IS NOONAN-LIKE;GENITOPATELLAR SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KCNJ1	Bartter syndrome 241200		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KCNJ2	Andersen syndrome, OMIM:170390;Andersen-Tawil syndrome, MONDO:0008222		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type, OMIM:300534		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
KDM6A	KABUKI SYNDROME 2		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
KIF11	AUTOSOMAL-DOMINANT MICROCEPHALY ASSOCIATED WITH LYMPHEDEMA AND/OR CHORIORETINOPATHY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KIF1A	NEUROPATHY, HEREDITARY SENSORY, TYPE IIC, 614213;NESCAV SYNDROME, 614255		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
KIF21A	Arthrogryposis;fetal akinesia		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KIF22	SPONDYLOEPIMETAPHYSEAL DYSPLASIA WITH JOINT LAXITY, TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KIF7	AUTOSOMAL RECESSIVE MENTAL RETARDATION;ACROCALLOSAL SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KLF1	Dyserythropoietic anemia, congenital, type IV, OMIM:613673;Hydrops Fetalis		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
KLHL7	PERCHING syndrome, OMIM:617055;PERCHING syndrome, MONDO:0014890		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KMT2C	INTELLECTUAL DISABILITY;Kleefstra syndrome 2 617768		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KMT2D	KABUKI SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KRAS	NOONAN SYNDROME TYPE 3;CARDIOFACIOCUTANEOUS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KRIT1	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KYNU	Vertebral, cardiac, renal, and limb defects syndrome 2 617661		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
L1CAM	MENTAL RETARDATION-APHASIA-SHUFFLING GAIT-ADDUCTED THUMBS SYNDROME;PARTIAL AGENESIS OF THE CORPUS CALLOSUM;HYDROCEPHALUS DUE TO STENOSIS OF THE AQUEDUCT OF SYLVIUS;SPASTIC PARAPLEGIA X-LINKED TYPE 1		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
L2HGDH	L-2-HYDROXYGLUTARIC ACIDURIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LAMA1	CEREBELLAR DYSPLASIA WITH CYSTS WITH OR WITHOUT RETINAL DYSTROPHY;AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LAMA2	CONGENITAL MUSCULAR DYSTROPHY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LAMB1	Lissencephaly 5, OMIM:615191;Cobblestone lissencephaly without muscular or ocular involvement, MONDO:0014077		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LAMC3	OCCIPITAL CORTICAL MALFORMATIONS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LARS2	Hydrops, lactic acidosis, and sideroblastic anemia, OMIM:617021		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LBR	HYDROPS-ECTOPIC CALCIFICATION-MOTH-EATEN SKELETAL DYSPLASIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LFNG	Spondylocostal dysostosis 3, autosomal recessive, OMIM:609813		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LHX3	PITUITARY HORMONE DEFICIENCY COMBINED TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LHX4	LHX4-RELATED COMBINED PITUITARY HORMONE DEFICIENCY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, OMIM:601559		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LIG4	LIG4 syndrome, OMIM:606593		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LIPA	Fetal hydrops;Wolman disease, 278000;Lysosomal Acid Lipase Deficiency		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LMBR1	Triphalangeal thumb-polysyndactyly syndrome 174500;Laurin-Sandrow syndrome 135750;Hypoplastic or aplastic tibia with polydactyly 188740;Syndactyly, type IV 186200;Triphalangeal thumb, type I 174500;Polydactyly, preaxial type II 174500;Acheiropody 200500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
LMBRD1	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA TYPE CBLF		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LMNA	HUTCHINSON-GILFORD PROGERIA SYNDROME;MUSCULAR DYSTROPHY CONGENITAL LMNA-RELATED;HEART-HAND SYNDROME SLOVENIAN TYPE;MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY;EMERY-DREIFUSS MUSCULAR DYSTROPHY TYPE 2;CARDIOMYOPATHY DILATED WITH HYPERGONADOTROPIC HYPOGONADISM;LETHAL TIGH		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
LMNB1	Microcephaly 26, primary, autosomal dominant, OMIM:619179		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
LMNB2	Microcephaly 27, primary, autosomal dominant, OMIM:619180		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
LMX1B	NAIL-PATELLA SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
LRP2	DONNAI-BARROW SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LRP4	CENANI-LENZ SYNDACTYLY SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LRP5	VITREORETINOPATHY EXUDATIVE TYPE 4;OSTEOPOROSIS-PSEUDOGLIOMA SYNDROME;ENDOSTEAL HYPEROSTOSIS WORTH TYPE;OSTEOPETROSIS AUTOSOMAL DOMINANT TYPE 1;HIGH BONE MASS TRAIT		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LRRC6	PRIMARY CILIARY DISKINESIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LTBP1	Cutis laxa, autosomal recessive, type IIE, OMIM:619451		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LTBP3	PLATYSPONDYLY WITH AMELOGENESIS IMPERFECTA		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
LTBP4	Cutis laxa, autosomal recessive, type IC 613177		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LYST	CHEDIAK-HIGASHI SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LZTFL1	Bardet-Biedl syndrome 17 615994		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MAF	CATARACT, DEAFNESS, INTELLECTUAL DISABILITY, SEIZURES, AND A DOWN SYNDROME-LIKE FACIES;CATARACT CONGENITAL CERULEAN TYPE 4;CATARACT PULVERULENT JUVENILE-ONSET MAF-RELATED		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MAFB	Duane Syndrome, Aberrant Extraocular Muscle Innervation, and Inner-Ear Defects;MULTICENTRIC CARPOTARSAL OSTEOLYSIS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MAGEL2	Schaaf-Yang syndrome;ARTHROGRYPOSIS MULTIPLEX CONGENITA;Schaaf-Yang syndrome, 615547		MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)	Fetal anomalies			5.5
MAP2K1	CARDIOFACIOCUTANEOUS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MAP2K2	CARDIOFACIOCUTANEOUS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MAP3K1	46XY SEX REVERSAL 6		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MASP1	3MC SYNDROME 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MATN3	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 5		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MBTPS2	Keratosis follicularis spinulosa decalvans, X-linked 308800;IFAP syndrome with or without BRESHECK syndrome 308205		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
MCOLN1	MUCOLIPIDOSIS IV		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MCPH1	MICROCEPHALY PRIMARY TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MED12	LUJAN-FRYNS SYNDROME;OPITZ-KAVEGGIA SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
MED13L	Impaired intellectual development and distinctive facial features with or without cardiac defects, OMIM:616789		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MED25	Basel-Vanagait-Smirin-Yosef syndrome, OMIM:616449		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MEF2C	MENTAL RETARDATION-STEREOTYPIC MOVEMENTS-EPILEPSY AND/OR CEREBRAL MALFORMATIONS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MEGF10	MYOPATHY, EARLY-ONSET, AREFLEXIA, RESPIRATORY DISTRESS, AND DYSPHAGIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MEIS2	Cleft palate, cardiac defects, and mental retardation, OMIM:600987;Cardiac malformation, cleft lip/palate, microcephaly, and digital anomalies, MONDO:0010970		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MESP2	SPONDYLOCOSTAL DYSOSTOSIS TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MFRP	NANOPHTHALMOS 2;MICROPHTHALMIA ISOLATED TYPE 5		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MFSD2A	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain imaging abnormalities, 616486		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MGP	KEUTEL SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MID1	OPITZ G/BBB SYNDROME, X-LINKED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
MINPP1	Pontocerebellar hypoplasia, type 16, OMIM:619527		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MKKS	BARDET-BIEDL SYNDROME TYPE 6;MCKUSICK-KAUFMAN SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MKS1	BARDET-BIEDL SYNDROME TYPE 13;MECKEL SYNDROME TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MLC1	LEUKOENCEPHALOPATHY MEGALENCEPHALIC WITH SUBCORTICAL CYSTS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MLYCD	MALONYL-COA DECARBOXYLASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MMACHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, CBLC TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MMADHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA TYPE CBLD		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MMP13	METAPHYSEAL ANADYSPLASIA TYPE 1;SPONDYLOEPIMETAPHYSEAL DYSPLASIA MISSOURI TYPE		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
MNX1	CURRARINO SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MOCS1	MOLYBDENUM COFACTOR DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MOCS2	MOLYBDENUM COFACTOR DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MOGS	Congenital disorder of glycosylation, type IIb, OMIM:606056;MOGS-CDG, MONDO:0011629		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MPDU1	CONGENITAL DISORDERS OF GLYCOSYLATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MPDZ	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, OMIM:615219		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MPLKIP	TRICHOTHIODYSTROPHY NON-PHOTOSENSITIVE TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MRPS22	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 5		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis, OMIM:616834;Microcephaly-congenital cataract-psoriasiform dermatitis syndrome, MONDO:0014793		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MSX1	Orofacial cleft 5, OMIM:608874		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MSX2	ENLARGED PARIETAL FORAMINA/CRANIUM BIFIDUM;CRANIOSYNOSTOSIS, TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MTFMT	Combined oxidative phosphorylation deficiency 15, OMIM:614947;Mitochondrial complex I deficiency, nuclear type 27, OMIM:618248		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MTM1	Myopathy, centronuclear, X-linked, OMIM:310400		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
MTO1	INFANTILE HYPERTROPHIC CARDIOMYOPATHY AND LACTIC ACIDOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MUSK	Fetal akinesia deformation sequence;Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYBPC1	Lethal congenital contracture syndrome 4 614915;Arthrogryposis, distal, type 1B 614335		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5

MYCN	FEINGOLD SYNDROME TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MYH11	Megacystis Microcolon Intestinal Hypoperistalsis Syndrome (MMIH)		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYH2	Proximal myopathy and ophthalmoplegia, OMIM:605637;Myopathy, proximal, and ophthalmoplegia, MONDO:0011577		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYH3	DISTAL ARTHROGRYPOSIS TYPE 2B;DISTAL ARTHROGRYPOSIS TYPE 2A		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MYH6	ATRIAL SEPTAL DEFECT TYPE 3;CARDIOMYOPATHY DILATED TYPE 1EE;CARDIOMYOPATHY FAMILIAL HYPERTROPHIC TYPE 14		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYH7	Laing distal myopathy, OMIM:160500;Laing early-onset distal myopathy, MONDO:0008050;Cardiomyopathy, hypertrophic, 1, OMIM:192600;Hypertrophic cardiomyopathy 1, MONDO:0008647;Cardiomyopathy, dilated, 1S, OMIM:613426;Dilated cardiomyopathy 1S, MONDO:0013262;Left ventricular noncompaction 5, OMIM:613426		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MYH8	DISTAL ARTHROGRYPOSIS TYPE;CARNEY COMPLEX VARIANT		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MYH9	SEBASTIAN SYNDROME;DEAFNESS AUTOSOMAL DOMINANT TYPE 17;EPSTEIN SYNDROME;FECHTNER SYNDROME;MACROTHROMBOCYTOPENIA WITH PROGRESSIVE SENSORINEURAL DEAFNESS;MAY-HEGGLIN ANOMALY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MYO18B	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, OMIM:616549;Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome, MONDO:0014689		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYOCD	Megabladder, congenital, OMIM:618719;Megabladder, congenital, MONDO:0032879		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYPN	Nemaline myopathy 11, autosomal recessive, 617336		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYT1	Oculo-auriculo-vertebral spectrum (OAVS);OAVS/Goldenhar syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NAA10	X-linked anophthalmia syndrome/Lenz;OGDEN SYNDROME;NONPECIFIC SEVERE ID;X-linked anophthalmia syndrome		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
NAGA	SCHINDLER DISEASE;KANZAKI DISEASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NBAS	ACUTE LIVER FAILURE (ALF) IN INFANCY AND CHILDHOOD		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NBN	NIJMEGEN BREAKAGE SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDE1	LISSENCEPHALY 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDP	NORRIE DISEASE		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
NDUFA6	Mitochondrial complex I deficiency, nuclear type 33, OMIM:618253		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16, 618238		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFB3	Mitochondrial complex I deficiency, nuclear type 25, OMIM:618246		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFS1	Mitochondrial complex I deficiency, nuclear type 5, OMIM:618226		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NEB	AUTOSOMAL RECESSIVE TYPICAL NEMALINE MYOPATHY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NEDD4L	Periventricular nodular heterotopia 7, OMIM:617201;Periventricular nodular heterotopia 7, MONDO:0014966		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NEK1	SHORT RIB-POLYDACTYLY SYNDORME, TYPE II;SHORT RIB-POLYDACTYLY SYNDROME, TYPE II;Short-rib thoracic dysplasia 6 with or without polydactyly, 263520		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NEK8	?Nephronophthisis 9, OMIM:613824;Nephronophthisis 9, MONDO:0013444;Renal-hepatic-pancreatic dysplasia 2, OMIM:615415;Renal-hepatic-pancreatic dysplasia 2, MONDO:0014174		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NEU1	SIALIDOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NF1	NEUROFIBROMATOSIS-NOONAN SYNDROME;FAMILIAL SPINAL NEUROFIBROMATOSIS;NEUROFIBROMATOSIS TYPE 1;WATSON SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NFIA	Brain malformations with or without urinary tract defects, OMIM:613735		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
NFIX	MARSHALL-SMITH SYNDROME;SOTOS-LIKE SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, OMIM:611291		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NHS	CATARACT CONGENITAL X-LINKED;NANCE-HORAN SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
NIPAL4	Ichthyosis, congenital, autosomal recessive 6, OMIM:612281;Autosomal recessive congenital ichthyosis 6, MONDO:0012847		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NIPBL	CORNELIA DE LANGE SYNDROME TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NKX2-5	ATRIAL SEPTAL DEFECT WITH ATRIOVENTRICULAR CONDUCTION DEFECTS;TETRALOGY OF FALLOT;CONGENITAL HYPOTHYROIDISM NON-GOITROUS TYPE 5		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NKX3-2	SPONDYLO-MEGAEPIPHYSEAL-METAPHYSEAL DYSPLASIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NODAL	HETEROTAXY SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NOG	BRACHYDACTYLY TYPE B2;SYMPHALANGISM PROXIMAL SYNDROME;MULTIPLE SYNOSTOSES SYNDROME TYPE 1;TARSAL-CARPAL COALITION SYNDROME;STAPES ANKYLOSIS WITH BROAD THUMB AND TOES		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NOTCH1	ADAMS OLIVER SYNDROME;LEFT VENTRICULAR OUTFLOW TRACT OBSTRUCTION		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NOTCH2	HAJDU-CHENEY SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NPC1	NIEMANN-PICK DISEASE, TYPE C1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NPC2	NIEMANN-PICK DISEASE, TYPE C2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NPHP1	SENIOR-LOKEN SYNDROME TYPE 1;NEPHRONOPHTHISIS TYPE 1;JOUBERT SYNDROME TYPE 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NPHP3	MECKEL SYNDROME TYPE 7;RENAL-HEPATIC-PANCREATIC DYSPLASIA;NEPHRONOPHTHISIS TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NPHP4	NEPHRONOPHTHISIS TYPE 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NPHS1	NEPHROTIC SYNDROME TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NPR2	ACROMESOMELIC DYSPLASIA MAROTEAUX TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NR0B1	Adrenal hypoplasia, congenital 300200;46XY sex reversal 2, dosage-sensitive 300018		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
NR5A1	46XY SEX REVERSAL 3;SPERMATOGENIC FAILURE 8		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NRAS	NOONAN SYNDROME TYPE 6		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NSD1	WEAVER SYNDROME;BECKWITH-WIEDEMANN SYNDROME;SOTOS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NSDHL	CONGENITAL HEMIDYSPLASIA WITH ICHTHYOSIFORM ERYTHRODERMA AND LIMB DEFECTS;CK SYNDROME		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
NUBPL	MITOCHONDRIAL COMPLEX I DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
OBSL1	3-M SYNDROME 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
OCLN	Band-like calcification with simplified gyration and polymicrogyria 251290		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
OCRL	Dent disease 2, OMIM:300555;Lowe syndrome, OMIM:309000		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
OFD1	JOUBERT SYNDROME TYPE 10;SIMPSON-GOLABI-BEHMEL SYNDROME TYPE 2;ORAL-FACIAL-DIGITAL SYNDROME TYPE 1		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ORC1	Meier-Gorlin syndrome 1, OMIM:224690		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ORC4	Meier-Gorlin syndrome 2, OMIM:613800		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ORC6	Meier-Gorlin syndrome 3, OMIM:613803		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
OSTM1	Osteopetrosis 259720		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
OTX2	MICROPHTHALMIA SYNDROMIC TYPE 5		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PAFAH1B1	Lissencephaly 1, OMIM:607432;Subcortical laminar heterotopia, OMIM:607432		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PAK3	MENTAL RETARDATION X-LINKED TYPE 30;AGENESIS OF THE CORPUS CALLOSUM		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
PALB2	FANCONI ANEMIA, COMPLEMENTATION GROUP N		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PAPSS2	SPONDYLOEPIMETAPHYSEAL DYSPLASIA PAKISTANI TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PAX1	Otofaciocervical syndrome 2, OMIM:615560		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PAX2	RENAL-COLOBOMA SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PAX3	WAARDENBURG SYNDROME, TYPE 1;CRANIOFACIAL-DEAFNESS-HAND SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PAX6	COLOBOMA OF OPTIC NERVE;ANIRIDIA;BILATERAL OPTIC NERVE HYPOPLASIA;ANIRIDIA CEREBELLAR ATAXIA AND MENTAL DEFICIENCY;KERATITIS HEREDITARY;PETERS ANOMALY;FOVEAL HYPOPLASIA		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PAX7	Myopathy, congenital, progressive, with scoliosis, OMIM:618578;Myopathy, congenital, progressive, with scoliosis, MONDO:0032821		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PAX8	CONGENITAL HYPOTHYROIDISM NON-GOITROUS TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PC	Pyruvate carboxylase deficiency, OMIM:266150		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, OMIM:210720		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PDCD10	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 3		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PDE4D	ACRODYSOSTOSIS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency;X-LINKED LEIGH SYNDROME;PYRUVATE DEHYDROGENASE E1-ALPHA DEFICIENCY IN FEMALES;INTELLECTUAL DISABILTIY		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
PDHB	Pyruvate dehydrogenase E1-beta deficiency, OMIM:614111		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PDHX	Lacticacidemia due to PDX1 deficiency, OMIM:245349		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEPD	PROLIDASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX1	ADRENOLEUKODYSTROPHY NEONATAL;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 1;INFANTILE REFSUM DISEASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX10	ADRENOLEUKODYSTROPHY NEONATAL;ZELLWEGER SYNDROME;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 7		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX11B	Peroxisome biogenesis disorder 14B		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX12	ZELLWEGER SYNDROME;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX13	ADRENOLEUKODYSTROPHY NEONATAL;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 13		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX14	PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP K;ZELLWEGER SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX16	PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 9;ZELLWEGER SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX19	ZELLWEGER SYNDROME;PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 14		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX2	PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 5;ZELLWEGER SYNDROME;INFANTILE REFSUM DISEASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX26	PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 8;ADRENOLEUKODYSTROPHY NEONATAL;ZELLWEGER SYNDROME;INFANTILE REFSUM DISEASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX3	PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 12;ZELLWEGER SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX5	ZELLWEGER SYNDROME;ADRENOLEUKODYSTROPHY NEONATAL;INFANTILE REFSUM DISEASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX6	Heimler syndrome 2, OMIM:616617;Peroxisome biogenesis disorder 4A (Zellweger), OMIM:614862;Peroxisome biogenesis disorder 4B, OMIM:614863		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
PEX7	PEROXISOME BIOGENESIS DISORDER COMPLEMENTATION GROUP 11;RHIZOMELIC CHONDRODYSPLASIA PUNCTATA TYPE 1;REFSUM DISEASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PFKM	Glycogen storage disease VII, OMIM:232800;Glycogen storage disease VII, MONDO:0009295		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PGM1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IT		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PHF6	Borjeson-Forssman-Lehmann syndrome, OMIM:301900		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
PHF8	MENTAL RETARDATION SYNDROMIC X-LINKED SIDERIUS TYPE		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
PHGDH	PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY;NEU-LAXOVA SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PHIP	Developmental delay, ID, obesity and dysmorphic features		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PHOX2B	NEUROBLASTOMA WITH HIRSCHSPRUNG DISEASE;CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL, WITH OR WITHOUT HIRSCHSPRUNG DISEASE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PIEZO2	ARTHROGRYPOSIS, DISTAL, TYPE 3;Ataxia, dysmetria, contractures & scoliosis with normal cognition but loss of discriminative touch perception		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
PIGA	MULTIPLE CONGENITAL ANOMALIES-HYPOTONIA-SEIZURES SYNDROME 2		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
PIGL	ZUNICH NEUROECTODERMAL SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, OMIM:614080;Multiple congenital anomalies-hypotonia-seizures syndrome 1, MONDO:0013563		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PIGO	HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PIGV	HYPERPHOSPHATASIA WITH MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PIK3CA	MEGALENCEPHALY-CAPILLARY MALFORMATION-POLYMICROGYRIA SYNDROME, SOMATIC 3;CLOVES: CONGENITAL LIPOMATOUS OVERGROWTH, VASCULAR MALFORMATIONS, AND EPIDERMAL NEVI;HEMIMEGALENCEPHALY PIK3CA		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PIK3R1	AGAMMAGLOBULINEMIA 7, AUTOSOMAL RECESSIVE;SHORT SYNDROME		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, OMIM:603387		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PITX1	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, OMIM:119800;Clubfoot, MONDO:0007342;Liebenberg syndrome, OMIM:186550;Brachydactyly-elbow wrist dysplasia syndrome, MONDO:0008520		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PITX2	RING DERMOID OF CORNEA;IRIDOGONIODYSGENESIS TYPE 2;AXENFELD-RIEGER SYNDROME TYPE 1;PETERS ANOMALY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PITX3	ANTERIOR SEGMENT MESENCHYMAL DYSGENESIS;CATARACT POSTERIOR POLAR TYPE 4;CATARACT AUTOSOMAL DOMINANT		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PKD1	Polycystic kidney disease, 173900;Autosomal recessive polycystic kidney disease (ARPKD);Autosomal dominant polycystic kidney disease (ADPKD)		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Fetal anomalies			5.5
PKD1L1	Laterality defects		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PKD2	Polycystic kidney disease 613095		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PKHD1	POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PKLR	Pyruvate kinase deficiency, OMIM:266200		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PLCB4	AURICULOCONDYLAR SYNDROME		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
PLEC	Muscular dystrophy, limb-girdle, autosomal recessive 17, OMIM:613723;Epidermolysis bullosa simplex 5A, Ogna type, OMIM:131950;Epidermolysis bullosa simplex 5C, with pyloric atresia, OMIM:612138;Epidermolysis bullosa simplex with muscular dystrophy, OMIM:226670		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
PLG	Plasminogen deficiency, type I, OMIM:217090;Dysplasminogenemia, OMIM:217090		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PLOD1	EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC FORM		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PLOD2	BRUCK SYNDROME TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PLXND1	Truncus arteriosus, HP:0001660		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PMM2	CONGENITAL DISORDERS OF GLYCOSYLATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PNKP	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 10;ATAXIA-OCULOMOTOR APRAXIA 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PNPLA1	Ichthyosis, congenital, autosomal recessive 10, OMIM:615024;Autosomal recessive congenital ichthyosis 10, MONDO:0014011		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
POGZ	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
POLG2	Mitochondrial DNA depletion syndrome syndrome 16 (hepatic type), OMIM:618528;Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), OMIM:619425;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, OMIM:610131		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Fetal anomalies			5.5
POLR1C	TREACHER COLLINS SYNDROME TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
POLR1D	TREACHER COLLINS SYNDROME TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
POLR3A	LEUKODYSTROPHY, HYPOMYELINATING, 7, WITH OR WITHOUT OLIGODONTIA AND/OR HYPOGONADOTROPIC HYPOGONADISM;Autosomal Recessive Wiedemann Rautenstrauch Syndrome, 264090		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, OMIM:614381		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
POMGNT1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH MENTAL RETARDATION TYPE B3;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH BRAIN AND EYE ANOMALIES TYPE A3 (MDDGA3;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
POMT1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C1;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH BRAIN AND EYE ANOMALIES TYPE A1;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH MENTAL RETARDATION TYPE B1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
POMT2	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH MENTAL RETARDATION TYPE B2;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY LIMB-GIRDLE TYPE C2;MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY CONGENITAL WITH BRAIN AND EYE ANOMALIES TYPE A2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
POP1	Anauxetic dysplasia 2, OMIM:617396;Anauxetic dysplasia 2, MONDO:0054561		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
POR	Disordered steroidogenesis due to cytochrome P450 oxidoreductase 613571;Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis 201750		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PORCN	FOCAL DERMAL HYPOPLASIA		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
POU1F1	Pituitary hormone deficiency, combined or isolated, 1, OMIM:613038		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PPIB	Osteogenesis imperfecta, type IX 259440		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PPP2R1A	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PQBP1	RENPENNING S(YNDROME 1		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
PRF1	Aplastic anaemia, OMIM:609135;Haemophagocytic lymphohistiocytosis, familial, 2, OMIM:603553		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome 208250		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PRKACA	Cardioacrofacial dysplasia 1, OMIM:619142		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
PRKAG2	Cardiomyopathy, hypertrophic 6, OMIM:600858;Hypertrophic cardiomyopathy 6, MONDO:0010946;Glycogen storage disease of heart, lethal congenital, OMIM:261740;Lethal congenital glycogen storage disease of heart, MONDO:0009867		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PRKAR1A	ACRODYSOSTOSIS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PRMT7	Pseudohypoparathyroidism-like disorder		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PRRX1	Agnathia-otocephaly complex, OMIM:202650		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
PRSS56	MICROPHTHALMIA ISOLATED TYPE 6		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PSAP	ATYPICAL KRABBE DISEASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PSAT1	Neu-Laxova syndrome 2, OMIM:616038;Neu-Laxova syndrome 2, MONDO:0014466		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PSPH	PHOSPHOSERINE PHOSPHATASE DEFICIENCY;NEU-LAXOVA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PTCH1	Holoprosencephaly 7, OMIM:610828		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PTF1A	DIABETES MELLITUS, PERMANENT NEONATAL, WITH CEREBELLAR AGENESIS;PANCREATIC AGENESIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PTH1R	CHONDRODYSPLASIA BLOMSTRAND TYPE;EIKEN SKELETAL DYSPLASIA;JANSEN METAPHYSEAL CHONDRODYSPLASIA;PRIMARY FAILURE OF TOOTH ERUPTION		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
PTHLH	BRACHYDACTYLY, TYPE E2;CLUBBING WITH SKELETAL DYSPLASIA INC ACROOSTEOLYSIS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PTPN11	LEOPARD SYNDROME TYPE 1;NOONAN SYNDROME 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PTPN14	Choanal atresia and lymphedema, OMIM:613611;Lymphedema-posterior choanal atresia syndrome, MONDO:0013324		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PTS	6-PYRUVOYLTETRAHYDROPTERIN SYNTHASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PXDN	Anterior segment dysgenesis 7, with sclerocornea, OMIM:269400		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PYCR1	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE IIB		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PYGM	McArdle disease, OMIM:232600;Glycogen storage disease V, MONDO:0009293		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RAB18	WARBURG MICRO SYNDROME TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RAB23	ACROCEPHALOPOLYSYNDACTYLY TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RAB3GAP1	WARBURG MICRO SYNDROME TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RAB3GAP2	MARTSOLF SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RAC1	Developmental Disorders with Diverse Phenotypes		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RAD21	COHESINOPATHY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RAD50	Nijmegen breakage syndrome-like disorder, OMIM:613078;MONDO:0013118		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RAD51	Fanconi anaemia, complementation group R, OMIM:617244		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
RAF1	NOONAN SYNDROME 5		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RAI1	SMITH-MAGENIS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RAPSN	FETAL AKINESIA DEFORMATION SEQUENCE;CONGENITAL MYASTHENIC SYNDROME WITH ACETYLCHOLINE RECEPTOR DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5

RARS2	PONTOCEREBELLAR HYPOPLASIA TYPE 6		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RASA1	PARKES WEBER SYNDROME;CAPILLARY MALFORMATION-ARTERIOVENOUS MALFORMATION		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RAX	Microphthalmia, isolated 3, OMIM:611038;isolated microphthalmia 3, MONDO:0012604		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RBBP8	Seckel syndrome 2, OMIM:606744;Seckel syndrome 2, MONDO:0011715		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RBM10	TARP syndrome, OMIM:311900;Tarp syndrome, MONDO:0010711		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
RBM8A	THROMBOCYTOPENIA-ABSENT RADIUS SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RBP4	Microphthalmia, isolated, with coloboma 10, OMIM:616428		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
RECQL4	Baller-Gerold syndrome, OMIM:218600;RAPADILINO syndrome, OMIM:266280;Rothmund-Thomson syndrome, type 2, OMIM:268400		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RELN	LISSENCEPHALY 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
REN	Renal tubular dysgenesis 267430		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RET	RENAL AGENESIS;MULTIPLE ENDOCRINE NEOPLASIA IIB		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
RFT1	Congenital disorder of glycosylation, type In, OMIM:612015;RFT1-CDG, MONDO:0012783		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RFX6	MARTINEZ-FRIAS SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RIPK4	POPLITEAL PTERYGIUM SYNDROME, LETHAL TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RMRP	CARTILAGE-HAIR HYPOPLASIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RNASEH2A	AICARDI-GOUTIERES SYNDROME 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RNASEH2B	Aicardi-Goutieres syndrome 2, OMIM:610181		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RNASEH2C	AICARDI-GOUTIERES SYNDROME 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RNASET2	LEUKOENCEPHALOPATHY, CYSTIC, WITHOUT MEGALENCEPHALY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RNF113A	Trichothiodystrophy 5, nonphotosensitive, OMIM:300953		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
RNU4ATAC	Microcephalic osteodysplastic primordial dwarfism, type I, OMIM:210710		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ROBO1	Tetralogy of Fallot and septal defects;Neurooculorenal syndrome, OMIM:620305		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ROBO3	Gaze palsy, familial horizontal, with progressive scoliosis, 1, OMIM:607313;Gaze palsy, familial horizontal, with progressive scoliosis 1, MONDO:0020790		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ROGDI	KOHLSCHAYTTER-TANZ SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ROR2	Brachydactyly, type B1, OMIM:113000 (AD);Robinow syndrome, autosomal recessive, OMIM:268310 (AR)		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
RPGRIP1L	MECKEL SYNDROME TYPE 5;COACH SYNDROME;JOUBERT SYNDROME TYPE 7		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RPL10	Mental retardation, X-linked, syndromic, 35, OMIM:300998;Intellectual disability, X-linked, syndromic, 35, MONDO:0030908		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
RPL11	Diamond-Blackfan anemia 7 612562;Diamond-Blackfan anemia with cleft palate and abnormal thumbs		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RPL35A	Diamond-Blackfan anemia 5, OMIM:612528;Diamond-Blackfan anemia 5, MONDO:0012925		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RPL5	Diamond-Blackfan anemia 6 612561		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RPS10	Diamond-Blackfan anemia 9 613308		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RPS17	Diamond-Blackfan anemia 4 612527		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RPS19	RPS19-RELATED DIAMOND-BLACKFAN ANEMIA		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RPS24	Diamond-blackfan anemia 3, OMIM:610629;Diamond-Blackfan anemia 3, MONDO:0012529		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RPS26	Diamond-Blackfan anemia 10 613309		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RPS6KA3	Coffin-Lowry syndrome, OMIM:303600		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
RPS7	Diamond-Blackfan anemia 8, OMIM:612563;Diamond-Blackfan anemia 8, MONDO:0012939		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RRM2B	Mitochondrial depletion syndrome		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RSPH4A	Ciliary dyskinesia, primary 612649		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RSPH9	Ciliary dyskinesia, primary 612650		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RUNX2	CLEIDOCRANIAL DYSPLASIA		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
RYR1	MINICORE MYOPATHY WITH EXTERNAL OPHTHALMOPLEGIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SALL1	TOWNES-BROCKS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SALL4	DUANE-RADIAL RAY SYNDROME;ACRO-RENAL-OCULAR SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SAMD9	MIRAGE - myelodysplasia, infection, restriction of growth, adrenal hypoplasia, genital phenotypes, enteropathy		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SAMHD1	AICARDI-GOUTIERES SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SATB2	NONSPECIFIC SEVERE ID;SYNDROMAL PIERRE ROBIN SEQUENCE;CLEFT PALATE ISOLATED		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SBDS	SHWACHMAN-DIAMOND SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SCARF2	VAN DEN ENDE-GUPTA SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SCN1A	Dravet syndrome, OMIM:607208;Arthrogryposis multiplex congenita		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SCN2A	NONSPECIFIC SEVERE ID;INFANTILE EPILEPTIC ENCEPHALOPATHY;BENIGN FAMILIAL NEONATAL INFANTILE SEIZURES		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SCN3A	Epileptic encephalopathy, early infantile, 62, OMIM:617938;Epilepsy, familial focal, with variable foci 4, OMIM:617935;Intellectual disability;Malformations of cortical development		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
SCN4A	PARAMYOTONIA CONGENITA OF VON EULENBURG;HYPERKALEMIC PERIODIC PARALYSIS TYPE 1;HYPOKALEMIC PERIODIC PARALYSIS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SCN5A	{Sudden infant death syndrome, susceptibility to}, OMIM:272120;Long QT syndrome 3, OMIM:603830		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
SCO2	FATAL INFANTILE CARDIOENCEPHALOMYOPATHY DUE TO CYTOCHROME C OXIDASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SDCCAG8	SENIOR-LOKEN SYNDROME 7		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SEC23B	Dyserythropoietic anemia, congenital, type II, OMIM:224100		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SEMA3A	{Hypogonadotropic hypogonadism 16 with or without anosmia}, OMIM:614897;congenital heart disease;skeletal anomalies		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SEPSECS	Pontocerebellar hypoplasia type 2D		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SERPINF1	Osteogenesis imperfecta, type VI, OMIM:613982;Osteogenesis imperfecta type 6, MONDO:0013515		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SERPINH1	Osteogenesis imperfecta, type X, OMIM:613848;Osteogenesis imperfecta type 10, MONDO:0013459		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SETBP1	DEVELOPMENTAL AND EXPRESSIVE LANGUAGE DELAY;SCHINZEL-GIEDION MIDFACE RETRACTION SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SETD2	microcephaly;profound intellectual disability;congenital anomalies;dysmorphic facial features		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SF3B4	ACROFACIAL DYSOSTOSIS 1, NAGER TYPE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SGCG	Muscular dystrophy, limb-girdle, autosomal recessive 5, OMIM:253700;Autosomal recessive limb-girdle muscular dystrophy type 2C, MONDO:0009677		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SH3PXD2B	FRANK-TER HAAR SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SHANK3	Phelan-McDermid syndrome, OMIM:606232;Phelan-McDermid syndrome, MONDO:0011652		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SHH	HOLOPROSENCEPHALY TYPE 3;TRIPHALANGEAL THUMB-POLYSYNDACTYLY SYNDROME;MICROPHTHALMIA ISOLATED WITH COLOBOMA TYPE 5;SOLITARY MEDIAN MAXILLARY CENTRAL INCISOR		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SHOC2	NOONAN-LIKE SYNDROME WITH LOOSE ANAGEN HAIR		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SHOX	Langer mesomelic dysplasia, OMIM:249700 (PR);Leri-Weill dyschondrosteosis, OMIM:127300 (PD);Short stature, idiopathic familial, OMIM:300582		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Fetal anomalies			5.5
SIL1	Marinesco-Sjogren syndrome, 248800		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SIX3	HOLOPROSENCEPHALY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SIX5	BRANCHIOOTORENAL SYNDROME TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SIX6	Optic disc anomalies with retinal and/or macular dystrophy, OMIM:212550;Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome, MONDO:0008927		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SKI	SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SKIV2L	Trichohepatoenteric syndrome 2, OMIM:614602		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC12A1	Bartter syndrome, type 1, OMIM:601678;Bartter disease type 1, MONDO:0100344		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC12A6	Charcot-Marie-Tooth disease, axonal, type 2II, OMIM:620068;Agenesis of the corpus callosum with peripheral neuropathy, OMIM:218000		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC16A2	MCT8 (SLC16A2)-SPECIFIC THYROID HORMONE CELL TRANSPORTER DEFICIENCY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
SLC17A5	SALLA DISEASE;INFANTILE SIALIC ACID STORAGE DISORDER		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC25A19	Microcephaly, Amish type, OMIM:607196;Amish lethal microcephaly, MONDO:0011790		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC25A38	ANEMIA, SIDEROBLASTIC, PYRIDOXINE-REFRACTORY, AUTOSOMAL RECESSIVE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC26A2	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 4;ACHONDROGENESIS TYPE 1B;ATELOSTEOGENESIS TYPE 2;DIASTROPHIC DYSPLASIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC26A3	Chloride diarrhea, congenital, Finnish type 214700		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC27A4	ICHTHYOSIS PREMATURITY SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome, OMIM:602782;H syndrome, MONDO:0011273		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC2A10	ARTERIAL TORTUOSITY SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC33A1	AUTOSOMAL-RECESSIVE DISORDER WITH CONGENITAL CATARACTS, HEARING LOSS, AND LOW SERUM COPPER AND CERULOPLASMIN		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC35C1	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 2C		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC35D1	SCHNECKENBECKEN DYSPLASIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC5A7	Myasthenic syndrome, congenital, 20, presynaptic, OMIM:617143;Congenital myasthenic syndrome 20, MONDO:0014939		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLX4	FANCONI ANEMIA COMPLEMENTATION GROUP P		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SMAD2	Loeys-Dietz syndrome 6, OMIM:619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, OMIM:619657		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
SMAD3	SMAD3-RELATED LOEYS-DIETZ SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SMAD4	MYHRE SYNDROME;JUVENILE POLYPOSIS/HEREDITARY HEMORRHAGIC TELANGIECTASIA SYNDROME;JUVENILE POLYPOSIS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SMARCA2	NICOLAIDES-BARAITSER SYNDROME;COFFIN SIRIS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SMARCA4	RHABDOID TUMOR PREDISPOSITION SYNDROME 2;COFFIN SIRIS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SMARCB1	RHABDOID PREDISPOSITION SYNDROME 1;?COFFIN-SIRIS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SMARCE1	Coffin-Siris syndrome 5, OMIM:616938		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SMC1A	Cornelia de Lange syndrome 2, OMIM:300590;Cornelia de Lange syndrome 2, MONDO:0010370;Developmental and epileptic encephalopathy 85, with or without midline brain defects, OMIM:301044;Developmental and epileptic encephalopathy, 85, with or without midline brain defects, MONDO:0026771		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
SMC3	CORNELIA DE LANGE SYNDROME TYPE 3		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SMN1	Spinal muscular atrophy 253550;Spinal muscular atrophy 271150;Spinal muscular atrophy 253400;Spinal muscular atrophy 253300		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SMOC1	OPHTHALMOACROMELIC SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SMPD1	NIEMANN-PICK DISEASE TYPE B;NIEMANN-PICK DISEASE TYPE A		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SMS	Mental retardation, X-linked, Snyder-Robinson type, OMIM:309583;Syndromic X-linked intellectual disability Snyder type, MONDO:0010664		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
SNX10	Osteopetrosis, autosomal recessive 8, OMIM:615085;Autosomal recessive osteopetrosis 8, MONDO:0014040		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SOS1	NOONAN SYNDROME 4		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SOST	SOST-Related Sclerosing Bone Dysplasias 122860;Sclerosteosis 1, 269500;Craniodiaphyseal dysplasia, autosomal dominant, 122860		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
SOX10	PERIPHERAL DEMYELINATING NEUROPATHY, CENTRAL DYSMYELINATING LEUKODYSTROPHY, WAARDENBURG SYNDROME, AND HIRSCHSPRUNG DISEASE;KALLMANN SYNDROME WITH DEAFNESS;YEMENITE DEAF-BLIND HYPOPIGMENTATION SYNDROME;WAARDENBURG SYNDROME TYPE 4C;WAARDENBURG SYNDROME TYPE 2E		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SOX17	VESICOURETERAL REFLUX TYPE 3		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SOX18	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, OMIM:137940;Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, MONDO:0019073;Hypotrichosis-lymphedema-telangiectasia syndrome, OMIM:607823;Hypotrichosis-lymphedema-telangiectasia syndrome, MONDO:0011914		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
SOX2	AEG SYNDROME;MICROPHTHALMIA SYNDROMIC TYPE 3		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency, OMIM:300123;Intellectual disability, X-linked, with panhypopituitarism, MONDO:0010252;Panhypopituitarism, X-linked, OMIM:312000;Panhypopituitarism, X-linked, MONDO:0010712		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
SOX6	Tolchin-Le Caignec syndrome, OMIM:618971;Tolchin-Le Caignec syndrome, MONDO:0033544		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SOX9	PIERRE ROBIN SEQUENCE;CAMPOMELIC DYSPLASIA		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SP7	Osteogenesis imperfecta, type XII, OMIM:613849;Osteogenesis imperfecta type 12, MONDO:0013460		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SPECC1L	?Facial clefting, oblique, 1, OMIM:600251;Tessier number 4 facial cleft, MONDO:0010850;Hypertelorism, Teebi type, OMIM:145420;Hypertelorism, Teebi type, MONDO:0007780;Opitz GBBB syndrome, type II, OMIM:145410;Autosomal dominant Opitz G/BBB syndrome, MONDO:0007779		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SPG11	SPASTIC PARAPLEGIA-11		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SPINT2	Diarrhea 3, secretory sodium, congenital, syndromic, OMIM:270420;congenital secretory sodium diarrhea 3, MONDO:0010036		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SPRED1	LEGIUS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SPTB	Elliptocytosis-3, OMIM:617948;Anemia, neonatal hemolytic, fatal or near-fatal, OMIM:617948;Spherocytosis, type 2, OMIM:616649		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
SRCAP	FLOATING-HARBOR SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SRD5A2	Pseudovaginal perineoscrotal hypospadias 264600		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SRD5A3	CONGENITAL DISORDERS OF GLYCOSYLATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SRY	46XY SEX REVERSAL 1		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ST14	Ichthyosis, congenital, autosomal recessive 11, OMIM:602400;Autosomal recessive congenital ichthyosis 11, MONDO:0011218		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
STAR	CHOLESTEROL DESMOLASE-DEFICIENT CONGENITAL ADRENAL HYPERPLASIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
STIL	Microcephaly 7, primary, autosomal recessive, OMIM:612703;Microcephaly 7, primary, autosomal recessive, MONDO:0012989		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
STRA6	MICROPHTHALMIA SYNDROMIC TYPE 9		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, OMIM:611087;Polyhydramnios, megalencephaly, and symptomatic epilepsy, MONDO:0012611		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SUCLG1	FATAL INFANTILE LACTIC ACIDOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SUFU	Joubert syndrome 32, OMIM:617757;Joubert Syndrome with Cranio-facial and Skeletal Defects		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Fetal anomalies			5.5
SULT2B1	Ichthyosis, congenital, autosomal recessive 14, OMIM:617571;Ichthyosis, congenital, autosomal recessive 14, MONDO:0033091		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SUMF1	SULFATIDOSIS, JUVENILE, AUSTIN TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SYNE1	Arthrogryposis multiplex congenita 3, myogenic type, OMIM:618484;Arthrogryposis multiplex congenita 3, myogenic type, MONDO:0032778		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TAB2	Congenital heart defects, nonsyndromic, 2, OMIM:614980		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TAF1	Dysmorphic Features, Intellectual Disability, and Neurological Manifestations		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
TALDO1	Fetal hydrops;Transaldolase deficiency, 606003		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TAZ	BARTH SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
TBC1D23	Non-degenerative Pontocerebellar Hypoplasia		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TBC1D24	MYOCLONIC EPILEPSY, INFANTILE, FAMILIAL;DOORS SYNDROME;NON SYNDROMAL HEARING LOSS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TBCE	Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy;HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME;KENNY-CAFFEY SYNDROME TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TBL1XR1	Intellectual disability with autism spectrum disorder;Pierpont syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TBX1	22Q11.2 DELETION SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TBX15	Craniofacial Dysmorphism, Hypoplasia of Scapula and Pelvis, and Short Stature;Cousin Syndrome		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TBX20	ATRIAL SEPTAL DEFECT TYPE 4		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TBX3	ULNAR-MAMMARY SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TBX4	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, OMIM:147891		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TBX5	HOLT-ORAM SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TBX6	Spondylocostal dysostosis 5 122600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TCF4	PITT-HOPKINS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TCIRG1	Osteopetrosis, infantile malignant 259700		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TCOF1	TREACHER COLLINS SYNDROME TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TCTN1	Joubert syndrome 13 614173;JOUBERT SYNDROME AND RELATED DISORDERS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TCTN2	JOUBERT SYNDROME AND RELATED DISORDERS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TFAP2A	BRANCHIOOCULOFACIAL SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TFAP2B	CHAR SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TGFB2	LOEYS-DIETZ SYNDROME, TYPE 4		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TGFB3	LOEYS-DIETZ SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TGFBR1	LOEYS-DIETZ SYNDROME TYPE 2A;LOEYS-DIETZ SYNDROME TYPE 1A;AORTIC ANEURYSM FAMILIAL THORACIC TYPE 5		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TGFBR2	LOEYS-DIETZ SYNDROME;TGFBR2-RELATED LOEYS-DIETZ SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TGIF1	Holoprosencephaly 4 142946;HOLOPROSENCEPHALY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TGM1	Ichthyosis, congenital, autosomal recessive 242300		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
THRA	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 6		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TINF2	EXUDATIVE RETINOPATHY WITH BONE MARROW FAILURE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type), OMIM:609560		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TMCO1	CRANIOFACIAL DYSMORPHISM, SKELETAL ANOMALIES, AND MENTAL RETARDATION SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TMEM138	Joubert syndrome 16 614465		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TMEM165	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IIK		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TMEM216	Meckel syndrome 2, OMIM:603194;Meckel syndrome, type 2, MONDO:0011296		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TMEM237	JOUBERT SYNDROME 14		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TMEM67	JOUBERT SYNDROME TYPE 6;MECKEL SYNDROME TYPE 3;COACH SYNDROM;NEPHRONOPHTHISIS TYPE 11		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TMEM70	IUGR;Oligohydramnios;Anhydramnios;Cardiomyopathy		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TNNI2	Arthrogryposis multiplex congenita, distal, type 2B 601680		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TNNT1	Nemaline myopathy, Amish type 605355		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5

TNNT3	Arthrogryposis, distal, type 2B2, OMIM:618435;Arthrogryposis, distal, type 2B2, MONDO:0032750		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TOR1A	Arthrogryposis multiplex congenita 5, OMIM:618947;Arthrogryposis multiplex congenita 5, MONDO:0100218		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TP63	ADULT syndrome, OMIM:103285;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, OMIM:604292;Hay-Wells syndrome, OMIM:106260;Limb-mammary syndrome, OMIM:603543;Orofacial cleft 8, OMIM:618149;Rapp-Hodgkin syndrome, OMIM:129400;Split-hand/foot malformation 4, OMIM:605289		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TP73	Ciliary dyskinesia, primary, 47, and lissencephaly, OMIM:619466		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TPM2	ARTHROGRYPOSIS, DISTAL, TYPE 1;Arthrogryposis multiplex congenita, distal, type 1, 108120;Arthrogryposis, distal, type 2B, 601680		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TPM3	Congenital fiber-type disproportion myopathy 255310		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
TRAF3IP1	Senior-Loken syndrome 9		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TRAPPC9	MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 13		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TREX1	AICARDI-GOUTIERES SYNDROME 1, DOMINANT AND RECESSIVE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TRIM37	MULIBREY NANISM		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TRIP11	ACHONDROGENESIS TYPE 1A		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TRIP12	TRIP12-related intellectual disability with/without autism spectrum disorder		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TRPS1	TRICHO-RHINO-PHALANGEAL SYNDROME TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TRPV4	METATROPIC DYSPLASIA;SPONDYLOMETAPHYSEAL DYSPLASIA, KOZLOWSKI TYPE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TRRAP	Developmental delay with or without dysmorphic facies and autism, OMIM:618454;multiple congenital anomalies		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
TSC1	TUBEROUS SCLEROSIS TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TSC2	LYMPHANGIOLEIOMYOMATOSIS;TUBEROUS SCLEROSIS TYPE 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TSEN2	PONTOCEREBELLAR HYPOPLASIA TYPE 2 AND TYPE 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TSEN34	PONTOCEREBELLAR HYPOPLASIA TYPE 2 AND TYPE 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TSEN54	?Pontocerebellar hypoplasia type 5, OMIM:610204;Pontocerebellar hypoplasia type 2A, OMIM:277470;Pontocerebellar hypoplasia type 4, OMIM:225753		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TSFM	Combined oxidative phosphorylation deficiency 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TTC21B	Short-rib thoracic dysplasia 4 with or without polydactyly 613819		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TTC37	TRICHOHEPATOENTERIC SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TTC8	RETINITIS PIGMENTOSA TYPE 51;BARDET-BIEDL SYNDROME TYPE 8		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TTN	congenital titinopathy with arthrogryposis		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TUBA1A	INTELLECTUAL DISABILITY;LISSENCEPHALY TYPE 3		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TUBB2B	Cortical dysplasia, complex, with other brain malformations 7, OMIM:610031		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TUBB3	CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFORMATIONS 1;CONGENITAL FIBROSIS OF THE EXTRAOCULAR MUSCLES		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TUBGCP4	AUTOSOMAL-RECESSIVE MICROCEPHALY WITH CHORIORETINOPATHY.		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TUBGCP6	MICROCEPHALY AND CHORIORETINOPATHY WITH OR WITHOUT MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TWIST1	SAETHRE-CHOTZEN SYNDROME;CRANIOSYNOSTOSIS, TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TWIST2	Ablepharon-macrostomia syndrome, 200110;Barber-Say syndrome, 209885		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
UBA1	Spinal muscular atrophy, X-linked 2, infantile 301830		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
UBR1	JOHANSON-BLIZZARD SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
UBR7	Li-Campeau syndrome, OMIM:619189		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
UMPS	OROTIC ACIDURIA TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
UROS	CONGENITAL ERYTHROPOIETIC PORPHYRIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
USP9X	MENTAL RETARDATION, X-LINKED 99		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
VIPAS39	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
VLDLR	CEREBELLAR ATAXIA MENTAL RETARDATION AND DYSEQUILIBRIUM SYNDROME TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
VPS13B	COHEN SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
VPS33B	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
VRK1	PONTOCEREBELLAR HYPOPLASIA TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
VSX2	MICROPHTHALMIA WITH CATARACTS AND IRIS ABNORMALITIES;MICROPHTHALMIA ISOLATED WITH COLOBOMA TYPE 3;MICROPHTHALMIA ISOLATED TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WDPCP	BARDET-BIEDL SYNDROME TYPE 15		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WDR19	ASPHYXIATING THORACIC DYSTROPHY 5;CRANIOECTODERMAL DYSPLASIA 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WDR35	CRANIOECTODERMAL DYSPLASIA 2;SHORT RIB-POLYDACTYLY SYNDROME, TYPE V		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WDR4	Microcephaly, growth deficiency, seizures, and brain malformations, OMIM:618346		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WDR62	MICROCEPHALY CORTICAL MALFORMATIONS AND MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WDR81	Hydrocephalus, congenital, 3, with brain anomalies, OMIM:617967;Hydrocephalus, congenital, 3, with brain anomalies, MONDO:0054794		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WNT10B	Split-hand/foot malformation 6, OMIM:225300		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WNT5A	WNT5A-RELATED ROBINOW SYNDROME, AUTOSOMAL DOMINANT		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
WNT7A	FUHRMANN SYNDROME;LIMB/PELVIS-HYPOPLASIA/APLASIA SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WRAP53	DYSKERATOSIS CONGENITA, AUTOSOMAL RECESSIVE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WT1	FRASIER SYNDROME FRASIER SYNDROME FRASIER SYNDROME;DENYS-DRASH SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
WWOX	Developmental and epileptic encephalopathy 28, OMIM:616211		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
XRCC4	Short stature, microcephaly, and endocrine dysfunction, OMIM:616541		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
XYLT1	Desbuquois dysplasia 2, OMIM:615777;Desbuquois dysplasia 2, MONDO:0014343		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
XYLT2	SPONDYLOOCULAR SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
YY1	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ZBTB18	ZBTB18 syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ZEB2	MOWAT-WILSON SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ZFP57	DIABETES MELLITUS, 6Q24-RELATED TRANSIENT NEONATAL		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ZIC1	CRANIOSYNOSTOSIS 6		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ZIC2	HOLOPROSENCEPHALY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ZIC3	VACTERL ASSOCIATION, X-LINKED, WITH OR WITHOUT HYDROCEPHALUS;HETEROTAXY SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ZMPSTE24	MANDIBULOACRAL DYSPLASIA WITH TYPE B LIPODYSTROPHY;LETHAL RESTRICTIVE DERMOPATHY, ZMPSTE24-RELATED		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ZNF335	Microcephaly 10, primary, autosomal recessive, OMIM:615095		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ZNF526	Dentici-Novelli neurodevelopmental syndrome, OMIM:619877		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AARS	Developmental and epileptic encephalopathy 29, OMIM:616339;Developmental and epileptic encephalopathy, 29, MONDO:0014593		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AASS	Hyperlysinemia, OMIM:238700;Hyperlysinemia (disease), MONDO:0009388		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ABCD4	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, CBLJ TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACO2	INFANTILE CEREBELLAR-RETINAL DEGENERATION		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACVR1	Fibrodysplasia ossificans progressiva, OMIM:135100		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
AGT	Renal tubular dysgenesis, OMIM:267430		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AIFM1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 6;COWCHOCK SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ALG11	ALG11-CDG		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALG13	EPILEPTIC ENCEPHALOPATHY;CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IS;EPILEPTIC ENCEPHALOPATHIES.		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
ANKRD26	THROMBOCYTOPENIA 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
AP3B2	Epileptic Encephalopathy with Optic Atrophy		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AP4M1	Spastic paraplegia 50, autosomal recessive, OMIM:612936		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ATP1A3	Developmental and epileptic encephalopathy 99, OMIM:619606;Polymicrogyria		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
B9D1	Meckel syndrome 9, OMIM:614209;Meckel syndrome 9, MONDO:0013630;Joubert syndrome 27, OMIM:617120;Joubert syndrome 27, MONDO:0014927		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BANF1	NESTOR-GUILLERMO PROGERIA SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BLOC1S6	HERMANSKY-PUDLAK SYNDROME 9		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BOLA3	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CACNA1A	Developmental and epileptic encephalopathy 42, OMIM:617106		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities, OMIM:615474		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
CACNA1S	Congenital myopathy;arthrogryposis		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CAMTA1	CEREBELLAR ATAXIA, NONPROGRESSIVE, WITH MENTAL RETARDATION		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CD151	NEPHROPATHY WITH PRETIBIAL EPIDERMOLYSIS BULLOSA AND DEAFNESS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CD96	C SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CELSR1	Lymphatic malformation 9, OMIM:619319		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CHD3	Apraxia of speech		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CHD8	AUTISM		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CHRM3	Prune belly syndrome, OMIM:100100;Megacystis		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHRNB2	CHRNB2-RELATED NOCTURNAL FRONTAL LOBE EPILEPSY, AUTOSOMAL DOMINANT;NOCTURNAL FRONTAL LOBE EPILEPSY, AUTOSOMAL DOMINANT		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CITED2	Atrial septal defect 8, OMIM:614433;Ventricular septal defect 2, OMIM:614431;Congenital heart disease		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
CLCNKB	Bartter syndrome, type 3, OMIM:607364;Bartter disease type 3, MONDO:0011822;Bartter syndrome, type 4b, digenic, OMIM:613090;Bartter disease type 4B, MONDO:0000909		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CNKSR2	INTELLECTUAL DISABILITY WITH EPILEPSY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
CNTN1	Myopathy, congenital, Compton-North, OMIM:612540		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COX14	?Mitochondrial complex IV deficiency, nuclear type 10 , OMIM:619053		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CRELD1	HETEROTAXY SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CSTA	EXFOLIATIVE ICHTHYOSIS, AUTOSOMAL RECESSIVE, ICHTHYOSIS BULLOSA OF SIEMENS-LIKE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, OMIM:604168		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CYB5R3	METHEMOGLOBINEMIA DUE TO DEFICIENCY OF METHEMOGLOBIN REDUCTASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DCDC2	RENAL-HEPATIC CILIOPATHY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), OMIM:251880;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, OMIM:617070;Portal hypertension, noncirrhotic, 1, OMIM:617068		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DHDDS	Epilepsy and intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
DNAJC19	3-methylglutaconic aciduria, type V, OMIM:610198		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DNM1	EPILEPTIC ENCEPHALOPATHY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
DSG1	SEVERE DERMATITIS, MULTIPLE ALLERGIES AND METABOLIC WASTING		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EDN1	AURICULOCONDYLAR SYNDROME		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
EDN3	Central hypoventilation syndrome, congenital, OMIM:209880;Waardenburg syndrome, type 4B, OMIM:613265;{Hirschsprung disease, susceptibility to, 4}, OMIM:613712		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
EMG1	Bowen-Conradi syndrome;Bowen-Conradi syndrome, 211180		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ENG	Telangiectasia, hereditary hemorrhagic, type 1, OMIM:187300		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
ESAM	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, OMIM:620371		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FANCM	FANCM-RELATED FANCONI ANEMIA;FANCONI ANEMIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FBXW11	Neurodevelopmental, jaw, eye, and digital syndrome, OMIM:618914		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
FGF9	Multiple synostoses syndrome 3, OMIM:612961		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
FMN2	NONSYNDROMIC AUTOSOMAL-RECESSIVE INTELLECTUAL DISABILITY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FN1	Spondylometaphyseal Dysplasia with Corner Fractures		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FOXL2	BLEPHAROPHIMOSIS, PTOSIS, AND EPICANTHUS INVERSUS SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FOXP2	Speech-language disorder-1, OMIM:602081;Structural abnormalities of basal ganglia		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FTO	Growth retardation, developmental delay, facial dysmorphism		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FUCA1	FUCOSIDOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FZD6	Nail disorder, nonsyndromic congenital, 1, OMIM:161050		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GABRA1	EPILEPTIC ENCEPHALOPATHY;JUVENILE MYOCLONIC EPILEPSY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GABRG2	EPILEPSY, GENERALIZED, WITH FEBRILE SEIZURES PLUS, TYPE 3;GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 3		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GLIS2	NEPHRONOPHTHISIS 7		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GLMN	Glomulovenous malformations, OMIM:138000		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
GM2A	GM2-GANGLIOSIDOSIS TYPE AB		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GNA14	Congenital vascular tumours		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GNAQ	Congenital Hemangioma		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GNB5	Sinus Bradycardia and Cognitive Disability		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GRHL2	ECTODERMAL DYSPLASIA/SHORT STATURE SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GSPT2	XL INTELLECTUAL DISABILITY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
HGSNAT	MUCOPOLYSACCHARIDOSIS TYPE 3C		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HPD	TYROSINEMIA TYPE 3;HAWKINSINURIA		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
IGFBP7	RETINAL ARTERIAL MACROANEURYSM WITH SUPRAVALVULAR PULMONIC STENOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ITCH	AUTOIMMUNE DISEASE, SYNDROMIC MULTISYSTEM		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KCNC3	SPINOCEREBELLAR ATAXIA TYPE 13		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KCNJ6	KEPPEN-LUBINSKY SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KCNJ8	Cantu syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
KCNK9	Birk-Barel syndrome, OMIM:612292		MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)	Fetal anomalies			5.5
KRT74	HYPOTRICHOSIS SIMPLEX OF THE SCALP 2		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
LIAS	Neonatal-onset epilepsy, defective mitochondrial energy metabolism, and glycine elevation		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LIPN	ICHTHYOSIS, LAMELLAR, 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LOX	Aortopathy		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LRAT	LEBER CONGENITAL AMAUROSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MAMLD1	Hypospadias 2, OMIM:300758		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
MAN1B1	AUTOSOMAL RECESSIVE MENTAL RETARDATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MANBA	LYSOSOMAL BETA-MANNOSIDOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MAT1A	METHIONINE ADENOSYLTRANSFERASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, OMIM:613668		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MGAT2	Congenital disorder of glycosylation, type IIa, OMIM:212066		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MIR17HG	FEINGOLD SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
MITF	COMMAD syndrome, OMIM:617306		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MPZ	Hypomyelinating neuropathy, congenital, 2, OMIM:618184		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
MRPS16	Combined oxidative phosphorylation deficiency 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MTPAP	?Spastic ataxia 4, autosomal recessive, OMIM:613672		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MVK	Hyper-IgD syndrome, OMIM:260920;Mevalonic aciduria, OMIM:610377		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYBPC3	Cardiomyopathy, hypertrophic, 4, OMIM:115197		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYLK	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, OMIM:249210		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NAGLU	MUCOPOLYSACCHARIDOSIS TYPE 3B		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NCAPD2	Microcephaly 21, primary, autosomal recessive, OMIM:617983		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFA10	LEIGH SYNDROME DUP		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFAF2	LEIGH SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFC2	Mitochondrial complex I deficiency, nuclear type 36, OMIM:619170		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFV2	Mitochondrial complex I deficiency, nuclear type 7, OMIM:618229		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NEXN	Cardiomyopathy, dilated, 1CC, OMIM:613122;Cardiomyopathy, hypertrophic, 20, OMIM:613876		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Fetal anomalies			5.5
NHP2	DYSKERATOSIS CONGENITA, AUTOSOMAL RECESSIVE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NID1	Hydrocephalus with or without seizures;Dandy-Walker malformation and occipital cephalocele		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
NSUN2	AUTOSOMAL- RECESSIVE INTELLECTUAL DISABILITY MRT5		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NTRK2	Epilepsy and intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NUP62	INFANTILE STRIATONIGRAL DEGENERATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5

ORAI1	Myopathy, tubular aggregate, 2, OMIM:615883		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
PDE10A	Childhood-Onset Chorea with Bilateral Striatal Lesions		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PDE6H	RETINAL CONE DYSTROPHY 3 PDE6H;ACHROMATOPSIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PDSS1	COENZYME Q10 DEFICIENCY, PRIMARY, 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PHEX	Hypophosphatemic rickets, X-linked dominant, OMIM:307800		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
PKP2	Severe cardiomyopathy with left ventricular noncompaction		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PLCB1	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 12		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PLOD3	Lysyl hydroxylase 3 deficiency, OMIM:612394;IUGR;Contractures		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
POLD1	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, OMIM:615381		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
POLG	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), OMIM:607459;Mitochondrial DNA depletion syndrome 4A (Alpers type), OMIM:203700;Mitochondrial DNA depletion syndrome 4B (MNGIE type), OMIM:613662		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PREPL	HYPOTONIA-CYSTINURIA SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PTH	FAMILIAL ISOLATED HYPOPARATHYROIDISM		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RAD51C	Fanconi anemia, complementation group O, OMIM:613390		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RIN2	Macrocephaly, alopecia, cutis laxa, and scoliosis, OMIM:613075		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RORA	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SACS	SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SCNN1B	Pseudohypoaldosteronism, type I, OMIM:264350		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SECISBP2	THYROID HORMONE METABOLISM, ABNORMAL		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SGSH	MUCOPOLYSACCHARIDOSIS TYPE 3A		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SHANK2	SUSCEPTIBILITY TO AUTISM TYPE 17		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SHROOM3	NEURAL TUBE DEFECT		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SLC1A2	EPILEPTIC ENCEPHALOPATHY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SLC22A5	Carnitine deficiency, systemic primary, OMIM:212140;systemic primary carnitine deficiency disease, MONDO:0008919		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC25A22	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC25A4	Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SLC35A1	CONGENITAL DISORDERS OF GLYCOSYLATION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC4A1	Ovalocytosis, SA type, OMIM:166900		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
SMOC2	DENTIN DYSPLASIA, TYPE I, WITH MICRODONTIA AND MISSHAPEN TEETH		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, OMIM:609528;CEDNIK syndrome, MONDO:0012290		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SOX5	12P12.5 INTRAGENIC DELETIONS ASSOCIATED WITH INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SPTAN1	Developmental and epileptic encephalopathy 5, OMIM:613477;developmental and epileptic encephalopathy, 5, MONDO:0013277		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ST3GAL3	Developmental and epileptic encephalopathy 15, OMIM:615006;developmental and epileptic encephalopathy, 15, MONDO:0014003;Intellectual developmental disorder, autosomal recessive 12, OMIM:611090;intellectual disability, autosomal recessive 12, MONDO:0012612		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ST3GAL5	Infantile epilepsy		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
STAT3	Autoimmune disease, multisystem, infantile-onset, 1, OMIM:615952;Hyper-IgE recurrent infection syndrome, OMIM:147060		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
STAT5B	GROWTH HORMONE INSENSITIVITY WITH IMMUNODEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
STIM1	Myopathy, tubular aggregate, OMIM:160565;Immunodeficiency 10, OMIM:612783;Stormorken syndrome, OMIM:185070		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), OMIM:612073		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SYN1	EPILEPSY, X-LINKED, WITH VARIABLE LEARNING DISABILITIES AND BEHAVIOR DISORDERS		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
SYT2	Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, OMIM:619461;Myasthenic syndrome, congenital, 7, presynaptic, OMIM:616040		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
TAC3	HYPOGONADOTROPIC HYPOGONADISM		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TACO1	LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX IV DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TACR3	HYPOGONADOTROPIC HYPOGONADISM		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TBC1D1	CAKUT		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
TBX22	Abruzzo-Erickson syndrome, OMIM:302905;Cleft palate with ankyloglossia, OMIM:303400		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
TLL1	Atrial septal defect 6, OMIM:613087		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
TNFRSF11A	Osteopetrosis, autosomal recessive 7, OMIM:612301		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TNFRSF13B	IMMUNODEFICIENCY, COMMON VARIABLE, 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TRIM32	LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 2H;BARDET-BIEDL SYNDROME TYPE 11		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TRIO	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, OMIM:617061		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
TRPM7	Cardiac arrhythmia, stillbirth		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
TRPV3	OLMSTED SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TUBA8	POLYMICROGYRIA WITH OPTIC NERVE HYPOPLASIA;Cortical dysplasia, complex, with other brain malformations 8, 613180		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TUFM	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TXN2	?Combined oxidative phosphorylation deficiency 29 , OMIM:616811		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3, OMIM:608898		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
UQCRB	Mitochondrial complex III deficiency, nuclear type 3, OMIM:615158		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
UQCRQ	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX III DEFICIENCY, UQCRQ RELATED		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
VDR	RICKETS VITAMIN D-DEPENDENT TYPE 2A		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WNT3	TETRA-AMELIA SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WNT4	SERKAL SYNDROME;MULLERIAN APLASIA AND HYPERANDROGENISM		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, OMIM:120433		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, OMIM:614069		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ZFPM2	Diaphragmatic hernia 3, OMIM:610187;Tetralogy of Fallot, OMIM:187500		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
ZMYND11	INTELLECTUAL DISABILITY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ZNF750	SEBORRHEA-LIKE DERMATITIS WITH PSORIASIFORM ELEMENTS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ABCB11	ABCB11-RELATED INTRAHEPATIC CHOLESTASIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ABCB7	ANEMIA, SIDEROBLASTIC, WITH ATAXIA		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ABCC8	Hyperinsulinemic hypoglycemia, familial 256450		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
ABCD1	ADRENOLEUKODYSTROPHY, X-LINKED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ACADM	MEDIUM CHAIN ACYL-COENZYME A DEHYDROGENASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACADS	SHORT CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACAT1	ALPHA-METHYLACETOACETIC ACIDURIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ACSL4	Mental retardation, X-linked 63 , OMIM:300387		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ADA	Severe combined immunodeficiency due to ADA deficiency, OMIM:102700;Adenosine deaminase deficiency, partial, OMIM:102700		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AFF2	FRAGILE X-E MENTAL RETARDATION SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
AGA	ASPARTYLGLUCOSAMINURIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AGPAT2	Lipodystrophy 608594		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AGRN	Fetal akinesia deformation sequence (FADS)		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AGXT	HYPEROXALURIA, PRIMARY, TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AIMP1	Leukodystrophy, hypomyelinating, 3, OMIM:260600		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AIPL1	LEBER CONGENITAL AMAUROSIS 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AIRE	AUTOIMMUNE POLYENDOCRINOPATHY SYNDROME TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AK2	RETICULAR DYSGENESIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AKR1D1	BILE ACID SYNTHESIS DEFECT, CONGENITAL, 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALAD	ACUTE HEPATIC PORPHYRIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALB	Analbuminemia, OMIM:616000		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALDH4A1	HYPERPROLINEMIA TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALDH5A1	SUCCINATE SEMIALDEHYDE DEHYDROGENASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALDOB	HEREDITARY FRUCTOSE INTOLERANCE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ALS2	ALS2-RELATED DISORDERS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ANO5	MIYOSHI MUSCULAR DYSTROPHY TYPE 3;GNATHODIAPHYSEAL DYSPLASIA		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
AP3B1	Hermansky-Pudlak syndrome 2 608233		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
APTX	ATAXIA WITH OCULOMOTOR APRAXIA 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ARG1	ARGININEMIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ASL	ARGININOSUCCINATE LYASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ATM	ATAXIA-TELANGIECTASIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ATP13A2	PARKINSON DISEASE 9		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ATP6V1B1	DISTAL RENAL TUBULAR ACIDOSIS WITH DEAFNESS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ATP8B1	ATP8B1-RELATED INTRAHEPATIC CHOLESTASIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AUH	3-METHYLGLUTACONIC ACIDURIA TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
AUTS2	Intellectual developmental disorder, autosomal dominant 26, OMIM:615834		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
BCKDHA	MAPLE SYRUP URINE DISEASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BCKDHB	MAPLE SYRUP URINE DISEASE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
BRWD3	MENTAL RETARDATION X-LINKED TYPE 93		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
C2orf71	RETINITIS PIGMENTOSA 54		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CBS	CYSTATHIONINE BETA-SYNTHASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CC2D1A	Intellectual developmental disorder, autosomal recessive 3, OMIM:608443		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CHD2	EPILEPTIC ENCEPHALOPATHY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CHRDL1	MEGALOCORNEA, X-LINKED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
CHRNA4	NOCTURNAL FRONTAL LOBE EPILEPSY TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CIB2	USHER SYNDROME TYPE 1J;NONSYNDROMIC DEAFNESS DFNB48		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CISD2	WOLFRAM SYNDROME TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CLCN5	Dent disease 1, OMIM:300009;Hypophosphatemic rickets, OMIM:300554;Nephrolithiasis, type I, OMIM:310468;Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, OMIM:308990		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
CLDN19	HYPOMAGNESEMIA 5, RENAL, WITH OCULAR INVOLVEMENT		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CLMP	Congenital short bowel syndrome, OMIM:615237		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CLN3	NEURONAL CEROID LIPOFUSCINOSIS TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CLN5	NEURONAL CEROID LIPOFUSCINOSIS TYPE 5		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CLN6	CEROID LIPOFUSCINOSIS, NEURONAL, KUFS TYPE, ADULT ONSET;CEROID LIPOFUSCINOSIS, NEURONAL, 6		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CLN8	NEURONAL CEROID LIPOFUSCINOSIS TYPE 8 NORTHERN EPILEPSY VARIANT;NEURONAL CEROID LIPOFUSCINOSIS TYPE 8		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CNBP	Myotonic dystrophy 2, OMIM:602668		Other	Fetal anomalies			5.5
COL25A1	Arthrogryposis multiplex congenita, MONDO:0015168		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL4A3	ALPORT SYNDROME AUTOSOMAL RECESSIVE;ALPORT SYNDROME AUTOSOMAL DOMINANT		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL4A4	ALPORT SYNDROME AUTOSOMAL RECESSIVE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COL5A1	Ehlers-Danlos syndrome, classic type 130000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
COL5A2	Ehlers-Danlos syndrome, classic type 130000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
COL9A3	Stickler syndrome, type VI, OMIM:620022;Epiphyseal dysplasia, multiple, 3, with or without myopathy, OMIM:600969		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
COMP	ARE THE CAUSE OF PSEUDOACHONDROPLASIA;MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
COQ2	COENZYME Q10 DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COX10	Mitochondrial complex IV deficiency, nuclear type 3, OMIM:619046		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COX15	LEIGH SYNDROME;MITOCHONDRIAL COMPLEX IV DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
COX6B1	Mitochondrial complex IV deficiency, nuclear type 7, OMIM:619051		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CPS1	CARBAMOYL PHOSPHATE SYNTHETASE 1 DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CRB1	LEBER CONGENITAL AMAUROSIS 8;RETINITIS PIGMENTOSA-12, AUTOSOMAL RECESSIVE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CRX	CRX-RELATED LEBER CONGENITAL AMAUROSIS LEBER CONGENITAL AMAUROSIS 7		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), OMIM:254800		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CTNS	CYSTINOSIS NEPHROPATHIC TYPE;CYSTINOSIS LATE-ONSET JUVENILE OR ADOLESCENT NEPHROPATHIC TYPE;CYSTINOSIS ADULT NON-NEPHROPATHIC TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
CYP19A1	Aromatase deficiency, OMIM:613546;Aromatase excess syndrome, OMIM:139300		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
D2HGDH	D-2-hydroxyglutaric aciduria, OMIM:600721		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DBT	MAPLE SYRUP URINE DISEASEQ		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DDB2	XERODERMA PIGMENTOSUM, GROUP E, DDB-NEGATIVE SUBTYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DDC	Aromatic L-amino acid decarboxylase deficiency, OMIM:608643;Aromatic L-amino acid decarboxylase deficiency, MONDO:0012084		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DDOST	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IR		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DEAF1	Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, OMIM:617171;Vulto-van Silfout-de Vries syndrome, OMIM:615828		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DHH	46XY partial gonadal dysgenesis, with minifascicular neuropathy;46XY sex reversal 7		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DICER1	GLOW syndrome, somatic mosaic, OMIM:618272;Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors , OMIM:138800;Pleuropulmonary blastoma, OMIM:601200		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
DLAT	PYRUVATE DEHYDROGENASE E2 DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DLD	DIHYDROLIPOAMIDE DEHYDROGENASE (E3) DEFICIENCY;LEIGH SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DLG3	MENTAL RETARDATION X-LINKED TYPE 90		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
DMP1	HYPOPHOSPHATEMIC RICKETS, AR		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DMPK	Myotonic dystrophy 1, OMIM:160900		Other	Fetal anomalies			5.5
DOCK8	HYPERIMMUNOGLOBULIN E RECURRENT INFECTION SYNDROME AUTOSOMAL RECESSIVE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
DSPP	DENTINOGENESIS IMPERFECTA, SHIELDS TYPE II;DEAFNESS AUTOSOMAL DOMINANT TYPE 39 WITH DENTINOGENESIS IMPERFECTA 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
EDA	Ectodermal dysplasia 1, hypohidrotic, X-linked, OMIM:305100		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive;Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900		Unknown	Fetal anomalies			5.5
EGR2	NEUROPATHY, CONGENITAL HYPOMYELINATING, 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
EPHX1	Hypercholanemia, familial;?Fetal hydantoin syndrome;Diphenylhydantoin toxicity		Unknown	Fetal anomalies			5.5
ETHE1	ETHYLMALONIC ENCEPHALOPATHY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FAM161A	RETINITIS PIGMENTOSA 28		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FARS2	Neurometabolic disorder due to FARS2 deficiency			Fetal anomalies			5.5
FBP1	FRUCTOSE 1,6 BISPHOSPHATASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FGD4	Charcot-Marie-Tooth disease 609311		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FGF20	?Renal hypodysplasia/aplasia 2, 615721		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FHL1	EMERY-DREIFUSS MUSCULAR DYSTROPHY 6, X-LINKED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
FLVCR1	ATAXIA, POSTERIOR COLUMN, WITH RETINITIS PIGMENTOSA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FMR1	Fragile X syndrome, OMIM:300624;Fragile X tremor/ataxia syndrome, OMIM:300623		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
FOXN1	ALOPECIA AND T-CELL IMMUNODEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FOXP1	MENTAL RETARDATION WITH LANGUAGE IMPAIRMENT AND AUTISTIC FEATURES		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
FRMD7	NYSTAGMUS 1, CONGENITAL, X-LINKED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
FRMPD4	Intellectual Disability, X-linked 104, OMIM:300983		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
FTCD	GLUTAMATE FORMIMINOTRANSFERASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
FTSJ1	MENTAL RETARDATION X-LINKED TYPE 44		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
FUZ	Neural tube defects 182940		Unknown	Fetal anomalies			5.5
G6PD	Hemolytic anemia, G6PD deficient (favism), OMIM:300908		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
GABRB3	EPILEPTIC ENCEPHALOPATHIES;CHILDHOOD ABSENCE EPILEPSY TYPE 5		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GALT	GALACTOSEMIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GAMT	GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5

GATM	Cerebral creatine deficiency syndrome 3, OMIM:612718		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GCH1	GTP CYCLOHYDROLASE 1 DEFICIENCY;DYSTONIA TYPE 5		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
GDI1	MENTAL RETARDATION X-LINKED TYPE 48;MENTAL RETARDATION X-LINKED TYPE 41		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
GJB2	VOHWINKEL SYNDROME;ICHTHYOSIS HYSTRIX-LIKE WITH DEAFNESS SYNDROME;PALMOPLANTAR KERATODERMA WITH DEAFNESS;BART-PUMPHREY SYNDROME;DEAFNESS AUTOSOMAL RECESSIVE TYPE 1A		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GK	GLYCEROL KINASE DEFICIENCY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
GLUD1	HYPERINSULINISM-HYPERAMMONEMIA SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GRIA3	MENTAL RETARDATION X-LINKED TYPE 94		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
GRIK2	MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 6		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GRIN2A	EPILEPSY WITH NEURODEVELOPMENTAL DEFECTS;LANDAU-KLEFFNER SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
GRM1	Spinocerebellar ataxia, autosomal recessive 13, OMIM:614831		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
GRM6	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1B		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
H19	Wilms tumor 2 194071;Beckwith-Wiedemann syndrome 130650;Silver-Russell syndrome 180860		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
HACE1	HACE1 related disorder		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HADH	3-HYDROXYACYL-COENZYME A DEHYDROGENASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HAX1	NEUTROPENIA, SEVERE CONGENITAL 3, AUTOSOMAL RECESSIVE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HCN1	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 24		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
HDAC4	BRACHYDACTYLY-MENTAL RETARDATION SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
HEXA	GM2-GANGLIOSIDOSIS TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, OMIM:268800		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HLCS	HOLOCARBOXYLASE SYNTHETASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HMGCL	3-HYDROXY-3-METHYLGLUTARYL-COENZYME A LYASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HMGCS2	3-HYDROXY-3-METHYLGLUTARYL-COA SYNTHASE 2 DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HNRNPU	EPILEPTIC ENCEPHALOPATHY		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
HOXA2	Microtia with or without hearing impairment (AD), OMIM:612290		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
HPGD	CRANIOOSTEOARTHROPATHY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HPRT1	Hyperuricemia, HRPT-related, OMIM:300323;Lesch-Nyhan syndrome, OMIM:300322		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
HPS1	HERMANSKY-PUDLAK SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HSD17B10	2-METHYL-3-HYDROXYBUTYRYL-COA DEHYDROGENASE DEFICIENCY;MENTAL RETARDATION SYNDROMIC X-LINKED TYPE 10		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
HSD3B7	BILE ACID SYNTHESIS DEFECT, CONGENITAL, 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HYAL1	MUCOPOLYSACCHARIDOSIS TYPE 9		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
HYDIN	CILIARY DYSKINESIA, PRIMARY, 5		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
IQSEC2	MENTAL RETARDATION X-LINKED TYPE 1		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ITGA7	CONGENITAL MUSCULAR DYSTROPHY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ITPR1	Spinocerebellar ataxia 29, congenital nonprogressive, OMIM:117360;Spinocerebellar ataxia 15, OMIM:606658;Gillespie syndrome, OMIM:206700		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
IVD	ISOVALERIC ACIDEMIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
JAK3	SEVERE COMBINED IMMUNE DEFICIENCY, AUTOSOMAL RECESSIVE, T CELL-NEGATIVE, B CELL -POSITIVE, NK CELL-NEGATIVE, JAK3-RELATED		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KARS	CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE, B;DEAFNESS, AUTOSOMAL RECESSIVE 89		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KBTBD13	NEMALINE MYOPATHY 6		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KCNE1	Jervell and Lange-Nielsen syndrome 2, OMIM:612347		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KCNJ10	SEIZURES-SENSORINEURAL DEAFNESS-ATAXIA-MENTAL RETARDATION-ELECTROLYTE IMBALANCE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KCNJ11	DIABETES MELLITUS, KCNJ11-RELATED TRANSIENT NEONATAL;FAMILIAL HYPERINSULINISM		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
KCNQ1	Long QT syndrome 1, OMIM:192500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KCNQ2	BENIGN NEONATAL EPILEPSY TYPE 1;EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 7		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KCNQ3	KCNQ3 syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KCTD7	NEURONAL CEROID LIPOFUSCINOSIS;PROGRESSIVE MYOCLONIC EPILEPSY TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KISS1R	Hypogonadotropic hypogonadism 8 with or without anosmia 614837		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
KIT	HUMAN PIEBALDISM		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
KMT2E	INTELLECTUAL DISABILITY;O'Donnell-Luria-Rodan syndrome, 618512		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
LAMA3	Epidermolysis bullosa, junctional 226700		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LAMB3	Epidermolysis bullosa, junctional 226700;Epidermolysis bullosa, junctional 226650		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LAMC2	Epidermolysis bullosa, junctional 226700;Epidermolysis bullosa, junctional 226650		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LAMP2	DANON DISEASE		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
LDB3	CARDIOMYOPATHY DILATED TYPE 1C;MYOPATHY MYOFIBRILLAR TYPE 4;LEFT VENTRICULAR NON-COMPACTION TYPE 3		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
LEMD3	MELORHEOSTOSIS;BUSCHKE-OLLENDORFF SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
LRPPRC	LEIGH SYNDROME, FRENCH-CANADIAN TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
LTBP2	MICROSPHEROPHAKIA;PRIMARY CONGENITAL GLAUCOMA TYPE 3D		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MAN2B1	LYSOSOMAL ALPHA-MANNOSIDOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MAOA	BRUNNER SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
MC2R	GLUCOCORTICOID DEFICIENCY 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MCCC1	3-METHYLCROTONYL-COA CARBOXYLASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MCCC2	3-METHYLCROTONYL-COA CARBOXYLASE 2 DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MCEE	METHYLMALONYL-COA EPIMERASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MECP2	ENCEPHALOPATHY NEONATAL SEVERE DUE TO MECP2 MUTATIONS;MENTAL RETARDATION SYNDROMIC X-LINKED TYPE 13;MENTAL RETARDATION SYNDROMIC X-LINKED LUBS TYPE;CHROMOSOME XQ28 DUPLICATION SYNDROME;RETT SYNDROME (RTT)[		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
MFSD8	MFSD8-RELATED NEURONAL CEROID-LIPOFUSCINOSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MLH1	Mismatch repair cancer syndrome 276300		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MMAA	METHYLMALONIC ACIDURIA TYPE CBLA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MMAB	METHYLMALONIC ACIDURIA TYPE CBLB		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MPI	Congenital disorder of glycosylation, type Ib, OMIM:602579;MPI-CDG, MONDO:0011257		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MPV17	MITOCHONDRIAL DNA DEPLETION SYNDROME 6		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MSH2	Mismatch repair cancer syndrome 276300;Mismatch repair cancer syndrome		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MSH6	Mismatch repair cancer syndrome 276300		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MTHFR	METHYLENETETRAHYDROFOLATE REDUCTASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MTR	METHYLCOBALAMIN DEFICIENCY TYPE G		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MTRR	HOMOCYSTINURIA-MEGALOBLASTIC ANEMIA, CBL E TYPE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MUT	METHYLMALONIC ACIDURIA TYPE MUT		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYO5A	GRISCELLI SYNDROME TYPE 3;ELEJALDE SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYO5B	Diarrhea 2, with microvillus atrophy, OMIM:251850		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYO7A	DEAFNESS AUTOSOMAL RECESSIVE TYPE 2;USHER SYNDROME TYPE 1B		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
MYT1L	MYT1L syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NAGS	N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFA1	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
NDUFA12	Mitochondrial complex I deficiency, nuclear type 23, OMIM:618244		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFS4	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY;LEIGH SYNDROME DUP;LEIGH SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFS7	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFS8	MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX I DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NDUFV1	MITOCHONDRIAL COMPLEX I DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NFU1	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NKX2-1	BENIGN HEREDITARY CHOREA;CHOREOATHETOSIS, HYPOTHYROIDISM, AND NEONATAL RESPIRATORY DISTRESS		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NKX2-6	Persistent truncus arteriosus, OMIM:217095;Conotruncal heart malformations, OMIM:217095		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NLRP3	CINCA syndrome, OMIM:607115		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
NMNAT1	LEBER CONGENITAL AMAUROSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NPHS2	NEPHROTIC SYNDROME, TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NPL	Sialic aciduria		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NR2F1	BOSCH-BOONSTRA OPTIC ATROPHY SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NRXN2	AUTISM		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NSMF	Hypogonadotropic hypogonadism 9 with or without anosmia, OMIM:614838		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
NT5C3A	HEMOLYTIC ANEMIA DUE TO UMPH1 DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NTRK1	CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
NYX	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1A		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
OTC	ORNITHINE TRANSCARBAMYLASE DEFICIENCY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
OXCT1	SUCCINYL-COA-3-KETOACID-COA TRANSFERASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PAH	PHENYLKETONURIA;NON-PHENYLKETONURIA HYPERPHENYLALANINEMIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PAX9	TOOTH AGENESIS, SELECTIVE, 3		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PCBD1	HYPERPHENYLALANINEMIA, BH4-DEFICIENT, D		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PCCA	PROPIONIC ACIDEMIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PCCB	PROPIONIC ACIDEMIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PCDH19	EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 9		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
PDE6G	RETINITIS PIGMENTOSA 57		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PDSS2	COENZYME Q10 DEFICIENCY, PRIMARY, 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PGK1	PHOSPHOGLYCERATE KINASE 1 DEFICIENCY		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
PLA2G6	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 2B;INFANTILE NEUROAXONAL DYSTROPHY 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PLCE1	NEPHROTIC SYNDROME, TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PLP1	SPASTIC PARAPLEGIA X-LINKED TYPE 2;LEUKODYSTROPHY HYPOMYELINATING TYPE 1		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
PMP22	Neuropathy, recurrent, with pressure palsies 162500;Charcot-Marie-Tooth disease, type 1A 118220;Charcot-Marie-Tooth disease, type 1E 118300;Dejerine-Sottas disease 145900;Neuropathy, inflammatory demyelinating 139393;Roussy-Levy syndrome 180800		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
PMS2	MISMATCH REPAIR CANCER SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PPM1D	PPM1D syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PPT1	NEURONAL CEROID LIPOFUSCINOSIS TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PROK2	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
PROKR2	Hypogonadotropic hypogonadism 3 with or without anosmia 244200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PROP1	PROP1-RELATED COMBINED PITUITARY HORMONE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PRPS1	PHOSPHORIBOSYLPYROPHOSPHATE SYNTHETASE SUPERACTIVITY;ARTS SYNDROME;CHARCOT-MARIE-TOOTH DISEASE X-LINKED RECESSIVE TYPE 5;DEAFNESS X-LINKED TYPE 1		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
PRRT2	BENIGN FAMILIAL INFANTILE EPILEPSY AND INFANTILE CONVULSIONS WITH CHOREOATHETOSIS SYNDROME;AUTOSOMAL RECESSIVE MENTAL RETARDATION		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
PRSS12	MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PRX	Dejerine-Sottas disease, OMIM:145900		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PSMB8	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, OMIM:256040		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
PTCHD1	AUTISM/ID		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
PTEN	MACROCEPHALY/AUTISM SYNDROME;BANNAYAN-ZONANA SYNDROME;PROTEUS SYNDROME;VACTERL ASSOCIATION WITH HYDROCEPHALUS;COWDEN DISEASE;LHERMITTE-DUCLOS DISEASE		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
PYGL	GLYCOGEN STORAGE DISEASE TYPE VI		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
QDPR	BH4-DEFICIENT HYPERPHENYLALANINEMIA C		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RAB39B	Intellectual developmental disorder, X-linked 72, OMIM:300271;Waisman syndrome, OMIM:311510		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
RPE65	LEBER CONGENITAL AMAUROSIS		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RPGRIP1	LEBER CONGENITAL AMAUROSIS 6;CONE-ROD DYSTROPHY 13		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
RSPO4	ANONYCHIA CONGENITA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SCN1B	EPILEPSY, GENERALIZED, WITH FEBRILE SEIZURES PLUS, TYPE 1;BRUGADA SYNDROME 5		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SCN8A	COGNITIVE IMPAIRMENT WITH OR WITHOUT CEREBELLAR ATAXIA;EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 13		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SCNN1A	Pseudohypoaldosteronism, type I, OMIM:264350		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
SCNN1G	Pseudohypoaldosteronism, type IB3, autosomal recessive, OMIM:620126		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SCO1	Mitochondrial complex IV deficiency, nuclear type 4, OMIM:619048		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SDHA	LEIGH SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SDHAF1	MITOCHONDRIAL COMPLEX II DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SDHD	Mitochondrial complex II deficiency, nuclear type 3, OMIM:619167		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SGCA	Muscular dystrophy, limb-girdle, type 2D 608099		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome, 300434		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
SIM1	Severe obesity with neurobehavioral features		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
SIX1	BRANCHIOOTIC SYNDROME TYPE 3;DEAFNESS AUTOSOMAL DOMINANT TYPE 23		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SLC19A3	THIAMINE METABOLISM DYSFUNCTION SYNDROME 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC25A15	HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC2A1	GLUT1 DEFICIENCY SYNDROME TYPE 2;GLUT1 DEFICIENCY SYNDROME TYPE 1		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SLC2A2	FANCONI-BICKEL SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC37A4	Glycogen storage disease Ib 232220		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC39A13	SPONDYLOEPIMETAPHYSEAL DYSPLASIA WITH ABNORMAL DENTITION;EHLERS-DANLOS SYNDROME-LIKE SPONDYLOCHEIRODYSPLASIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC46A1	HEREDITARY FOLATE MALABSORPTION		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC4A11	CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC4A4	PROXIMAL RENAL TUBULAR ACIDOSIS WITH OCULAR ABNORMALITIES		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC52A3	BROWN-VIALETTO-VAN LAERE SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC5A5	Thyroid dyshormonogenesis 1, OMIM:274400		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC6A1	EPILEPSY WITH MYOCLONIC-ATONIC SEIZURES		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SLC6A3	PARKINSONISM-DYSTONIA, INFANTILE		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC6A5	Hyperekplexia 3, 614618		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SLC6A8	X-LINKED CREATINE DEFICIENCY SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
SLC9A6	MENTAL RETARDATION SYNDROMIC X-LINKED CHRISTIANSON TYPE		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
SMAD6	{Craniosynostosis 7, susceptibility to}, OMIM:617439;Aortic valve disease 2, OMIM:614823;{Radioulnar synostosis, nonsyndromic}, OMIM:179300		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal anomalies			5.5
SMARCAL1	Schimke immunoosseous dysplasia, OMIM:242900		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SP110	Hepatic venoocclusive disease with immunodeficiency 235550		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SPR	DOPA-RESPONSIVE DYSTONIA DUE TO SEPIAPTERIN REDUCTASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SPTA1	Elliptocytosis-2, OMIM:130600;Spherocytosis, type 3, OMIM:270970;Hydrops fetalis;Congenital anaemia		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
SPTBN2	SCA14;Infantile ataxia with oculomotor and pyramidal signs;Spinocerebellar ataxia, autosomal recessive 14, 615386		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SPTBN5	Multicystic kidney;Oligohydramnios;Sacral agenesis		Unknown	Fetal anomalies			5.5
SPTLC2	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE IC		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
STAT1	STAT1 DEFICIENCY COMPLETE;MENDELIAN SUSCEPTIBILITY TO MYCOBACTERIAL DISEASE;FAMILIAL CANDIDIASIS TYPE 7		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, OMIM:614868		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
STS	ICHTHYOSIS, X-LINKED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
STXBP1	EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 4;ANGELMAN/PITT HOPKINS SYNDROME-LIKE DISORDER		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SURF1	COMPLEX IV DEFICIENCY;LEIGH SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
SYNGAP1	EPILEPTIC ENCEPHALOPATHY;MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 5		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
SYP	MENTAL RETARDATION X-LINKED SYP-RELATED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5

TAT	TYROSINEMIA TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TBXAS1	GHOSAL HEMATODIAPHYSEAL SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TCN2	Transcobalamin II deficiency		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TEK	VENOUS MALFORMATIONS, MULTIPLE CUTANEOUS AND MUCOSAL		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TERT	Dyskeratosis congenita, autosomal recessive 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TG	Thyroid dyshormonogenesis 3, OMIM:274700		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TGFB1	Camurati-Engelmann disease, OMIM:131300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TH	DOPA-RESPONSIVE DYSTONIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
THAP1	DYSTONIA 6, TORSION		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
TIMM8A	MOHR-TRANEBJAERG SYNDROME;JENSEN SYNDROME		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
TMPRSS6	IRON-REFRACTORY IRON DEFICIENCY ANEMIA		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TNFRSF11B	Paget disease 239000		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TNXB	Ehlers-Danlos syndrome due to tenascin X deficiency 606408;Vesicoureteral reflux 8 615963		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal anomalies			5.5
TPO	Thyroid dyshormonogenesis 2A, OMIM:274500		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TPP1	NEURONAL CEROID LIPOFUSCINOSIS TYPE 2		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TRAPPC2	SPONDYLOEPIPHYSEAL DYSPLASIA TARDA		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
TRMU	Liver failure, transient infantile, OMIM:613070		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TRPM1	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1C		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TSHB	HYPOTHRYROIDISM, CONGENITAL, NONGOITROUS 4		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TSHR	Hyperthyroidism, nonautoimmune, OMIM:609152;Hypothyroidism, congenital, nongoitrous, 1, OMIM:275200		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TSPAN7	MENTAL RETARDATION X-LINKED TYPE 58		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
TTC19	MITOCHONDRIAL COMPLEX III DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TTI2	Mental retardation, autosomal recessive 39, OMIM:615541;Microcephaly		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TUSC3	Intellectual developmental disorder, autosomal recessive 7, OMIM:611093		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TYR	OCULOCUTANEOUS ALBINISM TYPE 1		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
TYRP1	OCULOCUTANEOUS ALBINISM TYPE 3		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
UBE2A	UBE2A-RELATED X-LINKED SYNDROMIC MENTAL RETARDATION		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
UBE3A	ANGELMAN SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)	Fetal anomalies			5.5
UGT1A1	CRIGLER-NAJJAR SYNDROME, TYPE I		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
UNC80	Persistent Hypotonia, Encephalopathy, Growth Retardation, and Severe Intellectual Disability		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
UPF3B	MENTAL RETARDATION SYNDROMIC X-LINKED TYPE 14		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
UROC1	UROCANASE DEFICIENCY		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
USB1	Poikiloderma with neutropenia		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
UVSSA	UV-SENSITIVE SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
WDR11	KALLMANN SYNDROME		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Fetal anomalies			5.5
WDR45	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal anomalies			5.5
XPA	XERODERMA PIGMENTOSUM, GROUP A		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
XPC	XERODERMA PIGMENTOSUM, GROUP C		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ZDHHC9	MENTAL RETARDATION SYNDROMIC X-LINKED ZDHHC9-RELATED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
ZFYVE26	SPASTIC PARAPLEGIA AUTOSOMAL RECESSIVE TYPE 15		BIALLELIC, autosomal or pseudoautosomal	Fetal anomalies			5.5
ZNF711	MENTAL RETARDATION X-LINKED ZNF711-RELATED		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal anomalies			5.5
BRAF	LEOPARD syndrome 3 613707;Cardiofaciocutaneous syndrome 115150;Noonan syndrome 7 613706		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia 613563		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
HRAS	Costello syndrome, 218040		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
KRAS	Noonan syndrome 3 609942;Cardiofaciocutaneous syndrome 2 615278		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279;LEOPARD syndrome;?Noonan syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
MAP2K2	Cardiofaciocutaneous syndrome 4 615280		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
NF1	Neurofibromatosis-Noonan syndrome 601321;Neurofibromatosis, type 1 162200		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
NRAS	Noonan syndrome 6 613224;Cardio-Facio-cutanenous syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
PTPN11	LEOPARD syndrome 1 151100;Noonan syndrome 1 163950		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
RAF1	LEOPARD syndrome 2 611554;Noonan syndrome 5 611553		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
SHOC2	Noonan syndrome-like with loose anagen hair 1, 607721		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
SOS1	Noonan syndrome 4 610733		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
SPRED1	Legius syndrome 611431		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	RASopathies	RASopathies	Dysmorphic and congenital abnormality syndromes	1.81
EFNB1	Craniofrontonasal dysplasia OMIM:304110		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Common craniosynostosis syndromes			1.15
FGFR1	Jackson-Weiss syndrome OMIM:123150;Osteoglophonic dysplasia OMIM:166250;Pfeiffer syndrome OMIM:101600;Trigonocephaly 1 OMIM:190440		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Common craniosynostosis syndromes			1.15
FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis OMIM:207410;Apert syndrome OMIM:101200;Beare-Stevenson cutis gyrata syndrome OMIM:123790;Pfeiffer syndrome OMIM:101600;Craniofacial-skeletal-dermatologic dysplasia OMIM:101600;Crouzon syndrome OMIM:123500;Jackson-Weiss syndrome OMIM:123150;Saethre-Chotzen syndrome O		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Common craniosynostosis syndromes			1.15
FGFR3	Muenke syndrome OMIM:602849;Crouzon syndrome with acanthosis nigricans OMIM:612247;Thanatophoric dysplasia, type I OMIM:187600;Thanatophoric dysplasia, type II OMIM:187601		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Common craniosynostosis syndromes			1.15
TWIST1	Craniosynostosis 1 OMIM:123100;Saethre-Chotzen syndrome with or without eyelid anomalies OMIM:101400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Common craniosynostosis syndromes			1.15
ACTB	Baraitser-Winter syndrome 1, OMIM:243310;Baraitser-Winter syndrome 1, MONDO:0009470		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ACTG1	Baraitser-Winter syndrome 2, OMIM:614583		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ADAMTSL4	Ectopia lentis 225200/225100;craniosynostosis with ectopia lentis, MONDO:0011347		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ALPL	hypophosphatasia		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ALX4	Parietal foramina;Parietal foramina 2, (AD), 609597;Frontonasal dysplasia 2, (AR), 613451		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ARID1B	Coffin-Siris syndrome 1, OMIM:135900		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ARSB	Mucopolysaccharidosis VI (MPS6) 253200		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ASXL1	Bohring-Opitz syndrome, 605039;Metopic synostosis frequently associated with Bohring-Opitz syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
B3GAT3	Craniosynostosis and bone fragility;Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects OMIM:245600;Larsen-like syndrome, B3GAT3 type MONDO:0009511		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
BRAF	cardiofaciocutaneous syndrome type 115150;Noonan syndrome type 7 613706		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
CHD7	CHARGE syndrome, OMIM:214800;CHARGE syndrome, MONDO:0008965		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
COLEC11	265050;3MC syndrome 2 265050		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
CTSK	Pycnodysostosis 265800;265800		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
EFNB1	Craniofrontonasal dysplasia, OMIM:304110		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FAM20C	Raine syndrome 259775		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FBN1	Marfan syndrome, OMIM:154700;Marfan lipodystrophy syndrome, OMIM:616914;craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FGF9	Multiple synostoses syndrome 3, OMIM:612961		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FGFR1	Craniosynostosis		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FGFR2	Crouzon syndrome, 123500; Jackson-Weiss syndrome, 123150; Beare-Stevenson cutis gyrata syndrome, 123790; Pfeiffer syndrome, 101600; Apert syndrome, 101200; Saethre-Chotzen; Craniosynostosis, nonspecific syndrome, 101400; Gastric cance;Craniosynostosis		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FGFR3	Muenke syndrome;Crouzon syndrome with acanthosis nigricans		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FLNA	Frontometaphyseal dysplasia 1, OMIM:305620;Melnick-Needles syndrome, OMIM:309350;Otopalatodigital syndrome, type I, OMIM:311300;Otopalatodigital syndrome, type II, OMIM:304120		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
GLI3	Greig cephalopolysyndactyly syndrome, OMIM:175700		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
GNAS	103580;pseudohypoparathyroidism type 1a 103580;pseudohypoparathyroidism type 1a		MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
GNPTAB	252500;Mucolipidosis II alpha/beta(I cell disease) 252500		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
HUWE1	X-linked intellectual disability with CSS		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
IDS	Mucopolysaccharidosis II (MPS2, Hunter syndrome) 309900;309900		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
IDUA	607016;Mucopolysaccharidosis Ih/s (Hurler syndrome) 607014;607014		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
IFT122	Cranioectodermal dysplasia type 1 OMIM:218330;cranioectodermal dysplasia 1 MONDO:0021093		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly, OMIM:266920		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
IHH	Syndactyly, type 1, with or without craniosynostosis, OMIM:185900;Chr2q35dup syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
IL11RA	Craniosynostosis and dental anomalies, 614188;Craniosynostosis And Dental Anomalies		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
JAG1	Alagille syndrome 1, OMIM:118450		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
KAT6B	KAT6B-related disorders;Genitopatellar syndrome, OMIM:606170		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
KMT2D	Kabuki syndrome 1, OMIM:147920		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
KRAS	cardiofaciocutaneous syndrome type 2 615278;Noonan syndrome type 3 609942;615278;609942		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
LTBP1	Cutis laxa, autosomal recessive, type IIE, OMIM:619451;craniosynostosis, MONDO:0015469		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
MAN2B1	Mannosidosis, alpha-, types I and II, OMIM:248500		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
MSX2	Craniosynostosis, type 2, 604757;Parietal foramina 1, 168500;Parietal foramina with cleidocranial dysplasia, 168550;Craniosynostosis;MSX2-related craniosynostosis		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
NFIA	Metopic synostosis, hydrocephalus, thin corpus callosum, mild developmental delay, autism, macrocephaly		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
NFIX	Malan syndrome, OMIM:614753;Marshall-Smith syndrome, OMIM:602535;craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
PHEX	X-linked hypophosphataemic rickets		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
POR	201750;Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis 201750		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
PRRX1	Agnathia-otocephaly complex, OMIM:202650;craniosynostosis, MONDO:0015469;craniosynostosis, various combinations of sutures		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
PTCH1	metopic craniosynostosis		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
PTPN11	Noonan syndrome type 1 163950;leopard syndrome 151100		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
RAB23	Carpenter syndrome 201000;201000		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
RECQL4	218600;Baller-Gerold syndrome 218600		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
RUNX2	Craniosynostosis;156510		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SIX1	sagittal synostosis, multi-suture synostosis		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SKI	Shprintzen-Goldberg syndrome 182212;182212		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SMAD6	metopic synostosis;sagittal synostosis;{Craniosynostosis 7, susceptibility to} 617439		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SOX6	Tolchin-Le Caignec syndrome, OMIM:618971;craniosynostosis		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SPECC1L	Opitz GBBB syndrome, type II 145410		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
STAT3	Hyper IgE recurrent infection syndrome 147060;147060		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
TFAP2B	Char syndrome 169100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
TGFBR1	Loeys-Dietz syndrome 1 609192;609192		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
TGFBR2	610168;Loeys-Dietz syndrome 2 610168		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
TMCO1	MR syndrome;Cerebrofaciothoracic dysplasia/ craniofacial dysmorphism;skeletal anomalies;Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
TWIST1	Saethre-Chotzen syndrome, 101400; Saethre-Chotzen syndrome with eyelid anomalies, 101400; Craniosynostosis, type 1, 123100; Robinow-Sorauf syndrome, 180750		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
WDR35	Cranioectodermal dysplasia type 2 (Sensenbrenner syndrome) 613610;613610		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ZEB2	Mowat-Wilson syndrome 235730;235730		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ZIC1	?Craniosynostosis 6, 616602;Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ANKH	Craniometaphyseal dysplasia, OMIM:123000;craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
AXIN2	Oligodontia-colorectal cancer syndrome, OMIM:608615;craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ESCO2	Roberts syndrome;SC phocomelia syndrome			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FGF10	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FREM1	Trigonocephaly 2, OMIM:614485		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
GPC3	Simpson-Golabi-Behmel syndrome, type 1, OMIM:312870		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
IRX5	Hamamy syndrome, OMIM:611174		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
MASP1	3MC syndrome 1, OMIM:257920;3MC syndrome 1, MONDO:0009770		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SHOC2	Noonan-like syndrome with loose anagen hair 607721			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SMAD3	Loeys-Dietz syndrome 3, OMIM:613795		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ABCC9	hypertrichotic osteochondrodysplasia, Cantu syndrome 239850		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ACVRL1	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ALX1	Frontonasal dysplasia type 3			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ALX3	Frontonasal dysplasia type 1 (frontorhiny)			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
ATR	Seckel syndrome 1 210600			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
AXIN1	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
BBS9	Bardet-Biedl syndrome 9 615986			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
BMP4	orofacial cleft;Microphthalmia, syndromic type 6 607932			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
BRWD3	craniosynostosis, MONDO:0015469		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
CACNA1E	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, OMIM:235510			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
CD96	C syndrome			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
CHD3	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
COL11A1	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
CRTAP	Cole Carpenter			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
EDNRB	Waardenburg syndrome;ABCD syndrome;Hirschprung disease			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
EHMT1	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities, OMIM:617425		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FGF3	congenital deafness with inner ear agenesis, microtia and microdontia			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FLNB	spondylo-carpel-tarsel dysplasia;Larsen syndrome (dominant);atelsteogenesis type 1/3			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FOXP1	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FOXP2	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FTO	craniosynostosis, MONDO:0015469		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
FUZ	craniosynostosis, MONDO:0015469		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
GLI2	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
GLIS3	craniosynostosis, MONDO:0015469		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
GPC4	craniosynostosis, MONDO:0015469		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
HDAC4	craniosynostosis, MONDO:0015469		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
IFRD1	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
IFT43	Short-rib thoracic dysplasia 18 with polydactyly 617866;Cranioectodermal dysplasia type 3 614099		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
IGF1R	Insulin-like growth factor I, resistance to, OMIM:270450			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
IMPAD1	Chondrodysplasia with joint dislocations, GPAPP type			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
KANSL1	Koolen-de Vries/KANSL haploinsufficiency syndrome.			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
KDM6A	Kabuki syndrome 2 300867			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
LMX1B	Nail-patella syndrome - LOF			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
LRP5				Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
MED13L	Impaired intellectual development and distinctive facial features with or without cardiac defects, OMIM:616789;craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
NOG	Tarsal-carpal coalition syndrome;Stapes ankylosis with broad thumb and toes (Teunissen-Cremers syndrome);Brachydactyly type B2;Multiple synostosis syndrome;Symphalangism, proximal			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
NTRK2	Obesity, hyperphagia, and developmental delay, OMIM:613886;craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
OSTM1	Osteopetrosis, autosomal recessive 5, OMIM:259720;craniosynostosis, MONDO:0015469		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
PAX3	Waardenburg syndrome;Craniofacial-deafness-hand syndrome			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
PITX2			MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
PTPRD				Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1

SCARF2	Van den Ende-Gupta syndrome		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SCN4A				Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SCN8A	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SH3PXD2B	Borrone dermato-cardio-skeletal syndrome;Frank-ter-har 249420;249420		BIALLELIC, autosomal or pseudoautosomal	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SIX2	six2-related frontonasal dysplasia, MONDO:0044628;craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SMAD2	Loeys-Dietz syndrome 6, OMIM:619656		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SMC1A	Cornelia de Lange syndrome 2, OMIM:300590;craniosynostosis, MONDO:0015469		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SOX10	Hirschsprung disease;Waardenburg syndrome;Peripheral demyelinating neuropathy;central dysmyelination			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SP7	craniosynostosis, MONDO:0015469		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
SRCAP			MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
TCOF1	Treacher-Collins syndrome			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
TWIST2	Ablepharon-macrostomia syndrome;Barber-Say syndrome;Focal facial dermal dysplasia 3, Setleis type		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
WDR19	Cranioectodermal dysplasia type 4 614378			Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis syndromes	Skeletal disorders	5.1
AHCY	non-immune hydrops;Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752;S-adenosylhomocysteine hydrolase deficiency;AHCY deficiency		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
ALG8	Congenital disorder of glycosylation, type Ih, MIM#608104		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
ASAH1	disseminated lipogranulomatosis (Farber disease); NIHF; nonimmune hydrops fetalis		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
BRAF	LEOPARD Syndrome;Noonan Syndrome;Cardiofaciocutaneous Syndrome;LEOPARD syndrome 3;Cardio-facio-cutaneous syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
CANT1	Nonimmune hydrops fetalis		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;NSLL;Noonan syndrome-like disorder associated with JMML;Fetal hydrops (in some patients)		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, OMIM:235510;generalised lymphatic dysplasia;fetal hydrops		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
CDAN1	Dyserythropoietic anemia, congenital, type Ia, OMIM:224120;Anemia, congenital dyserythropoietic, type 1a, MONDO:0009135		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
CTSA	Galactosialidosis, 256540		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
FLT4	Lymphedema, hereditary, IA, 153100;Hereditary lymphoedema type 1a;fetal hydrops		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
FOXC2	Lymphedema-distichiasis syndrome, 153400;fetal hydrops		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790;IPEX;X-linked syndrome of immunodysregulation, polyendocrinopathy, and enteropath;fetal hydrops;Absent skeletal Muscle		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
GALNS	Mucopolysaccharidosis IVA, 253000;Mucopolysaccharidosis, Type IV;Mucopolysaccharidosis Type IVA;MUCOPOLYSACCHARIDOSIS TYPE 4A; Morquio disease type A		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
GATA1	Anaemia, X-linked, with/without neutropaenia and/or platelet abnormalities, OMIM:300835;Hemolytic anemia due to elevated adenosine deaminase, OMIM:301083		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
GBA	Gaucher disease;Gaucher disease, type I, 230800;Gaucher disease, type II, 230900;Gaucher disease, type III, 231000;Gaucher disease, type IIIC, 231005;Gaucher disease, perinatal lethal, 608013		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
GBE1	Glycogen storage disease IV, OMIM:232500;Fetal hydrops (in perinatal or congenital neuromuscular forms)		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
GLA	Fabry disease, 301500; Fabry disease, cardiac variant, 301500;Fabry Disease		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
GLB1	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type II, 230600;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;Mucopolysaccharidosis, Type IV;Mucopolysaccharidosis Type IVB;MUCOPOLYSACCHARIDOSIS TYPE 4B		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
GNPTAB	Mucolipidosis, Type II;Mucolipidosis, Type III Alpha/Beta;Mucolipidosis II alpha/beta;Mucolipidosis III alpha/beta; I-cell disease		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
GUSB	Mucopolysaccharidosis VII, 253220;MPS VII (Sly syndrome); Hydrops fetalis		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
HBA1	Thalassemias, alpha-, OMIM:604131;Fatal hydrops fetalis;Hb Bart syndrome		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
HBA2	Thalassemias, alpha-, OMIM:604131;Fatal hydrops fetalis;Hb Bart syndrome		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
HRAS	Costello syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
KLF1	Dyserythropoietic anemia, congenital, type IV, OMIM:613673;Hydrops Fetalis		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
KMT2D	Kabuki syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
KRAS	Noonan syndrome 3;Noonan syndrome;Cardiofaciocutaneous syndrome 2;Cardiofaciocutaneous Syndrome;Cardio-Facio-Cutaneous syndrome;CFC syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
LARS2	Hydrops, lactic acidosis, and sideroblastic anemia, OMIM:617021		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
LBR	Greenberg skeletal dysplasia, 215140;hydrops-ectopic calcification-moth-eaten skeletal dysplasia		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
LIPA	Wolman disease, 278000; Lysosomal Acid Lipase Deficiency		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
MAP2K1	Cardiofaciocutaneous syndrome 3;Cardiofaciocutaneous Syndrome;Cardio-Facio-Cutaneous syndrome;CFC syndrome;LEOPARD syndrome;?Noonan syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
MAP2K2	Cardiofaciocutaneous syndrome 4;Cardio-Facio-Cutaneous syndrome type 4;Cardiofaciocutaneous Syndrome;Cardio-Facio-Cutaneous syndrome;CFC syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
NEU1	Sialidosis, type I, 256550;Sialidosis, type II, 256550;Sialidosis;fetal hydrops;Hydrops fetalis (type II, congenital)		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
NPC1	Niemann-Pick disease, type C1, 257220; Niemann-Pick disease, type C; NPC1		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
NRAS	Noonan syndrome;Noonan syndrome 6;Cardio-Facio-cutanenous syndrome;CFC Syndrome		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX1	Zellweger; peroxisome biogenesis disorder 1A, 214100		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX10	Zellweger, peroxisome biogenesis disorder 6A; Peroxisome biogenesis disorder 6A (Zellweger), 614870; Peroxisome biogenesis disorder 6B, 614871		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX11B	Peroxisome biogenesis disorder 14B, 614920		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX12	Peroxisome biogenesis disorder 3B, 266510; Peroxisome biogenesis disorder 3A (Zellweger), 614859		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX13	Zellweger; peroxisome biogenesis disorder 11A, 614883; Peroxisome biogenesis disorder 11B, 614885		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX14	Zellweger, peroxisome biogenesis disorder 13A; Peroxisome biogenesis disorder 13A (Zellweger), 614887		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX16	Peroxisome biogenesis disorder 8B, 614877; Peroxisome biogenesis disorder 8A, (Zellweger), 614876		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX19	Zellweger, peroxisome biogenesis disorder 12A; Peroxisome biogenesis disorder 12A (Zellweger), 614886		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX26	Peroxisome biogenesis disorder 7B, 614873; Peroxisome biogenesis disorder 7A (Zellweger), 614872		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX3	Zellweger, peroxisome biogenesis disorder 10A; Peroxisome biogenesis disorder 10A (Zellweger); 614882		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX5	Peroxisome biogenesis disorder 2B, 202370; Peroxisome biogenesis disorder 2A (Zellweger) , 214110		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX6	Peroxisome biogenesis disorder 4A (Zellweger), OMIM:614862;Peroxisome biogenesis disorder 4B, OMIM:614863		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PEX7	Peroxisome biogenesis disorder 9B, 614879		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PKLR	Pyruvate kinase deficiency, OMIM:266200		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PMM2	Congenital disorder of glycosylation, type Ia, 212065;nonimmune hydrops fetalis;CDG1A;CDG 1a		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PTPN11	LEOPARD syndrome;LEOPARD syndrome 1;Noonan syndrome;Noonan syndrome 1		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
RAF1	Noonan syndrome;Noonan syndrome 5;LEOPARD syndrome;LEOPARD syndrome 2		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
SHOC2	Noonan-like syndrome with loose anagen hair		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
SLC17A5	Sialic acid storage disorder, infantile, 269920; ISSD; Hydrops fetalis; Infantile sialic acid storage disease		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
SMPD1	Niemann-Pick disease, type A, 257200		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
SOS1	Noonan syndrome;Noonan syndrome 4		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
TALDO1	Transaldolase deficiency, 606003		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
ALG9	Congenital disorder of glycosylation, type II, MIM#608776		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
ATP1A2	hydrops fetalis;microcephaly;arthrogryposis;extensive cortical malformations		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
FZD6	Nail disorder, nonsyndromic congenital, 1, OMIM:161050		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
G6PD	Nonimmune hydrops fetalis		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
IDUA	Mucopolysaccharidosis Ih, 607014; Hurler syndrome, MPS I		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
SLC30A5	Perinatal lethal cardiomyopathy;cardiomyopathy, MONDO:0004994		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
ALG1	Congenital disorder of glycosylation, type Ik, 608540; Hydrops fetalis, nonimmune		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
DHCR7	Smith-Lemli-Opitz syndrome, 270400; SLOS		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
DMPK	Myotonic dystrophy 1, OMIM:160900		Other	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
EBP	Chondrodysplasia punctata, X-linked dominant, 302960; Conradi Huenermann: Chondrodysplasia punctata		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
FH	Fumarase deficiency, 606812; Citric-acid cycle defect		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
GLE1	Lethal congenital contracture syndrome 1, 253310;LCCS1;MULTIPLE CONTRACTURE SYNDROME, FINNISH TYPE;Arthrogryposis, lethal, with anterior horn cell disease, 611890		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
HADHA	LCHAD deficiency, 609016		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
HBZ	HEMOGLOBIN H HYDROPS FETALIS SYNDROME		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
PSAT1	Neu-Laxova syndrome 2, MIM#616038		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
SUMF1	Multiple sulfatase deficiency, 272200		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
UROS	Porphyria, congenital erythropoietic, 263700; Congenital erythropoietic porphyria; Gunther's disease		BIALLELIC, autosomal or pseudoautosomal	Fetal hydrops	Fetal disorders	Dysmorphic and congenital abnormality syndromes	1.88
AAAS	Achalasia-addisonianism-alacrimia syndrome, OMIM:231550;Triple-A syndrome, MONDO:0009279		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AARS	Charcot-Marie-Tooth disease, axonal, type 2N, OMIM:613287;Charcot-Marie-Tooth disease axonal type 2N, MONDO:0013212;Developmental and epileptic encephalopathy 29, OMIM:616339;Developmental and epileptic encephalopathy, 29, MONDO:0014593		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AARS2	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AASS	Hyperlysinemia, OMIM:238700;Hyperlysinemia (disease), MONDO:0009388		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABAT	GABA-transaminase deficiency, 613163		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCA1	HDL deficiency, familial, 1, 604091;Tangier disease, 205400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCA12	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500;Ichthyosis, congenital, autosomal recessive 4A, 601277		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3, 610921		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCA4	Retinal dystrophy, early-onset severe, 248200;Fundus flavimaculatus, 248200;Stargardt disease 1, 248200;Cone-rod dystrophy 3, 604116;Retinitis pigmentosa 19, 601718		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCB11	Cholestasis, progressive familial intrahepatic 2, 601847;Cholestasis, benign recurrent intrahepatic, 2, 605479		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCB4	Cholestasis, progressive familial intrahepatic 3, 602347;Gallbladder disease 1, 600803;Cholestasis, intrahepatic, of pregnancy, 3, 614972		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCB7	Anemia, sideroblastic, with ataxia, 301310		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ABCC2	Dubin-Johnson syndrome, 237500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCC6	Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, 264800;Pseudoxanthoma elasticum, forme fruste, 177850		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCC8	Diabetes mellitus, transient neonatal 2, 610374;Hypoglycemia of infancy, leucine-sensitive, 240800;Diabetes mellitus, permanent neonatal, 606176;Hyperinsulinemic hypoglycemia, familial, 1, 256450;Diabetes mellitus, noninsulin-dependent, 125853		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCC9	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia, 239850;Atrial fibrillation, familial, 12, 614050		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCD1	Adrenomyeloneuropathy, adult, 300100;Adrenoleukodystrophy, 300100		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCG5	Sitosterolemia, 210250		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABCG8	Sitosterolemia, 210250		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ABHD5	Chanarin-Dorfman syndrome, 275630		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACAD8	Isobutyryl-CoA dehydrogenase deficiency, 611283		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACAD9	Mitochondrial complex I deficiency, nuclear type 20, 611126		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACADSB	2-methylbutyrylglycinuria, 610006		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACADVL	VLCAD deficiency, 201475		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACAN	Spondyloepimetaphyseal dysplasia, aggrecan type, 612813;Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800;?Spondyloepiphyseal dysplasia, Kimberley type, 608361		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACAT1	Alpha-methylacetoacetic aciduria, 203750		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACE	Renal tubular dysgenesis, 267430		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACO2	Infantile cerebellar-retinal degeneration, 614559		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACP5	Spondyloenchondrodysplasia with immune dysregulation, 607944		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACSF3	Combined malonic and methylmalonic aciduria, 614265		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACSL4	Mental retardation, X-linked 63, 300387		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
ACTA1	Myopathy, actin, congenital, with excess of thin myofilaments, 161800;Nemaline myopathy 3, autosomal dominant or recessive, 161800;Myopathy, congenital, with fiber-type disproportion 1, 255310;Myopathy, actin, congenital, with cores, 161800		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACTA2	Multisystemic smooth muscle dysfunction syndrome, 613834;Moyamoya disease 5, 614042;Aortic aneurysm, familial thoracic 6, 611788		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ACTB	Baraitser-Winter syndrome 1, 243310		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ACTC1	Left ventricular noncompaction 4, 613424;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424;Cardiomyopathy, hypertrophic, 11, 612098		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ACTG1	Baraitser-Winter syndrome 2, 614583;Deafness, autosomal dominant 20/26, 604717		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ACTN4	Glomerulosclerosis, focal segmental, 1, 603278		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ACVR1	Fibrodysplasia ossificans progressiva, 135100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ACVR2B	Heterotaxy, visceral, 4, autosomal, 613751		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2, 600376		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ACY1	Aminoacylase 1 deficiency, 609924		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ADA	Adenosine deaminase deficiency, partial, 102700;Severe combined immunodeficiency due to ADA deficiency, 102700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ADAMTS10	Weill-Marchesani syndrome 1, recessive, 277600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ADAMTS13	Thrombotic thrombocytopenic purpura, hereditary, OMIM:274150		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ADAMTS17	Weill-Marchesani 4 syndrome, recessive, 613195		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ADAMTSL2	Geleophysic dysplasia 1, 231050		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ADAR	Aicardi-Goutieres syndrome 6, 615010;Dyschromatosis symmetrica hereditaria, 127400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ADCY5	Dyskinesia, familial, with facial myokymia, 606703		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ADNP	Helsmoortel-van der Aa syndrome, 615873		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ADSL	Adenylosuccinase deficiency, 103050		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AFF2	Mental retardation, X-linked, FRAXE type, 309548		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
AFG3L2	Spinocerebellar ataxia 28, 610246;Spastic ataxia 5, autosomal recessive, 614487		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AGA	Aspartylglucosaminuria, 208400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AGK	Sengers syndrome, 212350;Cataract 38, autosomal recessive, 614691		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AGL	Glycogen storage disease IIIb, 232400;Glycogen storage disease IIIa, 232400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AGPS	Rhizomelic chondrodysplasia punctata, type 3, 600121		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AGRN	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AGT	Renal tubular dysgenesis, 267430		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AGTR1	Renal tubular dysgenesis, 267430		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AGXT	Hyperoxaluria, primary, type 1, 259900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AHI1	Joubert syndrome 3, 608629		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AICDA	Immunodeficiency with hyper-IgM, type 2, 605258		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AIFM1	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Deafness, X-linked 5, 300614		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
AIMP1	Leukodystrophy, hypomyelinating, 3, 260600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AIPL1	Cone-rod dystrophy, 604393;Leber congenital amaurosis 4, 604393;Retinitis pigmentosa, juvenile, 604393		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AK2	Reticular dysgenesis, 267500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AKR1D1	Bile acid synthesis defect, congenital, 2, 235555		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AKT1	Cowden syndrome 6, 615109		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ALAD	Porphyria, acute hepatic, 612740		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALAS2	Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ALB	Analbuminemia, OMIM:616000;[Dysalbuminemic hyperthyroxinemia], OMIM:615999;?[Dysalbuminemic hypertriiodothyroninemia], OMIM:615999		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALDH18A1	Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal dominant 3, 616603;Spastic paraplegia 9B, autosomal recessive, 616586		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALDH3A2	Sjogren-Larsson syndrome, 270200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALDH4A1	Hyperprolinemia, type II, 239510		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency, 614105		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALDOA	Glycogen storage disease XII, 611881		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALDOB	Fructose intolerance, hereditary, 229600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALG1	Congenital disorder of glycosylation, type Ik, 608540		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208

ALG11	Congenital disorder of glycosylation, type Ip, 613661		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALG12	Congenital disorder of glycosylation, type Ig, 607143		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALG13	Epileptic encephalopathy, early infantile, 36, 300884		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
ALG3	Congenital disorder of glycosylation, type Id, 601110		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALG6	Congenital disorder of glycosylation, type Ic, 603147		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALG8	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALG9	Congenital disorder of glycosylation, type Il, 608776;Gillessen-Kaesbach-Nishimura syndrome, 263210		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALMS1	Alstrom syndrome, 203800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALOX12B	Ichthyosis, congenital, autosomal recessive 2, 242100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALOXE3	Ichthyosis, congenital, autosomal recessive 3, 606545		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALPL	Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300;Hypophosphatasia, infantile, 241500;Odontohypophosphatasia, 146300		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALS2	Amyotrophic lateral sclerosis 2, juvenile, 205100;Primary lateral sclerosis, juvenile, 606353;Spastic paralysis, infantile onset ascending, 607225		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALX1	?Frontonasal dysplasia 3, 613456		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALX3	Frontonasal dysplasia 1, 136760		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ALX4	Parietal foramina 2, 609597;Frontonasal dysplasia 2, 613451		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AMELX	Amelogenesis imperfecta, type 1E, 301200		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
AMER1	Osteopathia striata with cranial sclerosis, 300373		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
AMN	Megaloblastic anemia-1, Norwegian type, 261100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AMT	Glycine encephalopathy, 605899		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ANK1	Spherocytosis, type 1, 182900		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ANKH	Craniometaphyseal dysplasia, 123000;Chondrocalcinosis 2, 118600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ANKRD11	KBG syndrome, 148050		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ANKRD26	Thrombocytopenia 2, 188000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ANO10	Spinocerebellar ataxia, autosomal recessive 10, 613728		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ANO5	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307;Miyoshi muscular dystrophy 3, 613319;Gnathodiaphyseal dysplasia, 166260		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ANO6	Scott syndrome, 262890		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ANTXR1	GAPO syndrome, 230740		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ANTXR2	Hyaline fibromatosis syndrome, 228600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AP1S1	MEDNIK syndrome, 609313		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AP1S2	Mental retardation, X-linked syndromic 5, 304340		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
AP3B1	Hermansky-Pudlak syndrome 2, 608233		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AP3B2	Epileptic encephalopathy, early infantile, 48, 617276		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AP4B1	Spastic paraplegia 47, autosomal recessive, OMIM:614066;Hereditary spastic paraplegia 47, MONDO:0013551		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AP4E1	Spastic paraplegia 51, autosomal recessive, 613744;Stuttering, familial persistent, 1, 184450		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AP4M1	Spastic paraplegia 50, autosomal recessive, 612936		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AP4S1	Spastic paraplegia 52, autosomal recessive, 614067		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AP5Z1	Spastic paraplegia 48, autosomal recessive, 613647		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
APC	Brain tumor-polyposis syndrome 2, 175100;Adenomatous polyposis coli, 175100;Desmoid disease, hereditary, 135290;Gardner syndrome, 175100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
APOA1	Amyloidosis, 3 or more types, 105200;ApoA-I and apoC-III deficiency, combined, 618463;Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
APOA5	Hyperchylomicronemia, late-onset, 144650		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
APOB	Hypobetalipoproteinemia, 615558;Hypercholesterolemia, familial, 2, 144010		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
APOC2	Hyperlipoproteinemia, type Ib, 207750		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
APOE	Lipoprotein glomerulopathy, 611771;Hyperlipoproteinemia, type III, 617347;Sea-blue histiocyte disease, 269600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
APRT	Adenine phosphoribosyltransferase deficiency, 614723		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AR	Androgen insensitivity, OMIM:300068;Androgen insensitivity, partial, with or without breast cancer, OMIM:312300;Hypospadias 1, X-linked, OMIM:300633;Spinal and bulbar muscular atrophy of Kennedy, OMIM:313200		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ARFGEF2	Periventricular heterotopia with microcephaly, 608097		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ARG1	Argininemia, 207800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ARHGAP31	Adams-Oliver syndrome 1, 100300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ARID1A	Coffin-Siris syndrome 2, 614607		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ARID1B	Coffin-Siris syndrome 1, 135900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ARL13B	Joubert syndrome 8, 612291		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ARL6	Bardet-Biedl syndrome 3, 600151		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ARSA	Metachromatic leukodystrophy, 250100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ARSE	Chondrodysplasia punctata, X-linked recessive, 302950		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ARX	Epileptic encephalopathy, early infantile, 1, 308350;Partington syndrome, 309510;Proud syndrome, 300004;Lissencephaly, X-linked 2, 300215;Hydranencephaly with abnormal genitalia, 300215;Mental retardation, X-linked 29 and others, 300419		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ASAH1	Farber lipogranulomatosis, 228000;Spinal muscular atrophy with progressive myoclonic epilepsy, 159950		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ASCC1	Barrett esophagus/esophageal adenocarcinoma, 614266		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ASL	Argininosuccinic aciduria, 207900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ASNS	Asparagine synthetase deficiency, 615574		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ASPA	Canavan disease, 271900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ASPM	Microcephaly 5, primary, autosomal recessive, 608716		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ASS1	Citrullinemia, 215700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ASXL1	Bohring-Opitz syndrome, 605039		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ATCAY	Ataxia, cerebellar, Cayman type, 601238		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATL1	Neuropathy, hereditary sensory, type ID, 613708;Spastic paraplegia 3A, autosomal dominant, 182600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ATM	Ataxia-telangiectasia, 208900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATN1	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, OMIM:618494;Dentatorubral-pallidoluysian atrophy, OMIM:125370		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ATP13A2	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATP1A2	Alternating hemiplegia of childhood 1, 104290;Migraine, familial basilar, 602481;Migraine, familial hemiplegic, 2, 602481		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ATP1A3	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ATP2A1	Brody myopathy, 601003		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATP6AP2	Mental retardation, X-linked, syndromic, Hedera type, 300423		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA, 219200;Wrinkly skin syndrome, 278250		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATP6V1B1	Renal tubular acidosis with deafness, 267300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATP7A	Spinal muscular atrophy, distal, X-linked 3, 300489;Menkes disease, 309400;Occipital horn syndrome, 304150		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ATP7B	Wilson disease, 277900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATP8B1	Cholestasis, progressive familial intrahepatic 1, 211600;Cholestasis, benign recurrent intrahepatic, 243300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATR	Seckel syndrome 1, 210600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATRX	Alpha-thalassemia/mental retardation syndrome, 301040;Mental retardation-hypotonic facies syndrome, X-linked, 309580		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
ATXN1	Spinocerebellar ataxia 1, OMIM:164400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ATXN10	Spinocerebellar ataxia 10, 603516		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ATXN2	Spinocerebellar ataxia 2, OMIM:183090;{Amyotrophic lateral sclerosis, susceptibility to, 13}, OMIM:183090		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ATXN3	Machado-Joseph disease, 109150		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ATXN7	Spinocerebellar ataxia 7, 164500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
AUH	3-methylglutaconic aciduria, type I, 250950		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
AUTS2	Mental retardation, autosomal dominant 26, 615834		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
AVPR2	Nephrogenic syndrome of inappropriate antidiuresis, 300539;Diabetes insipidus, nephrogenic, 304800		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
B2M	?Amyloidosis, familial visceral;Immunodeficiency 43, 241600		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
B3GALT6	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
B4GALT1	Congenital disorder of glycosylation, type IId, 607091		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
B9D2	Joubert syndrome 34, OMIM:614175;Meckel syndrome 10, OMIM:614175;Meckel syndrome, type 10, MONDO:0013609		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BAAT	Hypercholanemia, familial, 607748		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BAG3	Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
BBS1	Bardet-Biedl syndrome 1, 209900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BBS10	Bardet-Biedl syndrome 10, 615987		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BBS12	Bardet-Biedl syndrome 12, 615989		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BBS2	Bardet-Biedl syndrome 2, 615981;Retinitis pigmentosa 74, 616562		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BBS4	Bardet-Biedl syndrome 4, 615982		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BBS5	Bardet-Biedl syndrome 5, 615983		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BBS7	Bardet-Biedl syndrome 7, 615984		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BBS9	Bardet-Biedl syndrome 9, 615986		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BCAP31	Deafness, dystonia, and cerebral hypomyelination, 300475		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
BCKDHA	Maple syrup urine disease, type Ia, 248600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BCKDHB	Maple syrup urine disease, type Ib, 248600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BCL11A	Dias-Logan syndrome, 617101		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
BCOR	Microphthalmia, syndromic 2, 300166		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
BCS1L	Bjornstad syndrome, 262000;GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Leigh syndrome, 256000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BEAN1	Spinocerebellar ataxia 31, 117210		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
BFSP2	Cataract 12, multiple types, 611597		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
BHLHA9	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BIN1	Centronuclear myopathy 2, 255200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BLM	Bloom syndrome, 210900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BLOC1S3	Hermansky-Pudlak syndrome 8, 614077		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BLOC1S6	?Hermansky-pudlak syndrome 9, 614171		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BMP1	Osteogenesis imperfecta, type XIII, 614856		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BMP2	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877;Brachydactyly, type A2, 112600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
BMP4	Microphthalmia, syndromic 6, 607932;Orofacial cleft 11, 600625		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BMPER	Diaphanospondylodysostosis, 608022		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BMPR1A	Polyposis syndrome, hereditary mixed, 2, 610069;Juvenile polyposis syndrome, infantile form, 174900;Polyposis, juvenile intestinal, 174900		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BMPR1B	Acromesomelic dysplasia, Demirhan type, 609441;Brachydactyly, type A2, 112600;Brachydactyly, type A1, D, 616849		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BNC2	Lower urinary tract obstruction, congenital, 618612		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BRAF	Cardiofaciocutaneous syndrome, 115150;Noonan syndrome 7, 613706;LEOPARD syndrome 3, 613707		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498;Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BRCA1	Fanconi anemia, complementation group S, 617883		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BRCA2	Fanconi anemia, complementation group D1, 605724		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BRIP1	Fanconi anemia, complementation group J, 609054		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BRSK2			MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
BRWD3	Mental retardation, X-linked 93, 300659		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
BSCL2	Encephalopathy, progressive, with or without lipodystrophy, 615924;Silver spastic paraplegia syndrome, 270685;Lipodystrophy, congenital generalized, type 2, 269700;Neuropathy, distal hereditary motor, type VA, 600794		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BSND	Sensorineural deafness with mild renal dysfunction, 602522;Bartter syndrome, type 4a, 602522		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BTD	Biotinidase deficiency, 253260		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BTK	Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200;Agammaglobulinemia, X-linked 1, 300755		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
BUB1B	Mosaic variegated aneuploidy syndrome 1, 257300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C12orf57	Temtamy syndrome, 218340		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C12orf65	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C19orf12	?Spastic paraplegia 43, autosomal recessive, 615043;Neurodegeneration with brain iron accumulation 4, 614298		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C1QA	C1q deficiency, 613652		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C1QB	C1q deficiency, 613652		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C1QC	C1q deficiency, 613652		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C1R	Ehlers-Danlos syndrome, periodontal type, 1, 130080		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
C1S	Ehlers-Danlos syndrome, periodontal type, 2, 617174;C1s deficiency, 613783		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C2	C2 deficiency, 217000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C2orf71	Retinitis pigmentosa 54, 613428		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C3	C3 deficiency, 613779		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C4A	C4a deficiency, 614380		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C4B	C4B deficiency, 614379		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C5	C5 deficiency, 609536		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C5orf42	Joubert syndrome 17, 614615;Orofaciodigital syndrome VI, 277170		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C6	Combined C6/C7 deficiency;C6 deficiency, 612446		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C7	C7 deficiency, 610102		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C8A	C8 deficiency, type I, 613790		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C8B	C8 deficiency, type II, 613789		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C8orf37	Cone-rod dystrophy 16, 614500;Retinitis pigmentosa 64, 614500;Bardet-Biedl syndrome 21, 617406		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C9	C9 deficiency, 613825		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
C9orf72	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CACNA1A	Epileptic encephalopathy, early infantile, 42, 617106;Spinocerebellar ataxia 6, 183086;Migraine, familial hemiplegic, 1, 141500;Episodic ataxia, type 2, 108500;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CACNA1C	Brugada syndrome 3, 611875;Long QT syndrome 8, 618447;Timothy syndrome, 601005		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CACNA1E	Epileptic encephalopathy, early infantile, 69, 618285		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CACNA1F	Cone-rod dystrophy, X-linked, 3, 300476;Aland Island eye disease, 300600;Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
CACNA1G	Spinocerebellar ataxia 42, 616795;Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CACNA1S	Hypokalemic periodic paralysis, type 1, 170400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CACNB4	Episodic ataxia, type 5, 613855		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CANT1	Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CAPN3	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600;Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208

CARD14	Pityriasis rubra pilaris, OMIM:173200;Psoriasis 2, OMIM:602723		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CARD9	Candidiasis, familial, 2, autosomal recessive, 212050		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CASK	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749;FG syndrome 4, 300422;Mental retardation, with or without nystagmus, 300422		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
CASP10	Autoimmune lymphoproliferative syndrome, type II, 603909		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CASR	Hypercalcemia, familial, 239200;Hypocalciuria, autosomal dominant, with Bartter syndrome, 601198;Hypocalciuric hypercalcemia, type I, 145980;Hypocalciuria, autosomal dominant, 601198		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CAT	Acatalasemia, 614097		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CAV3	Myopathy, distal, Tateyama type, 614321;Long QT syndrome 9, 611818;Creatine phosphokinase, elevated serum, 123320;Cardiomyopathy, familial hypertrophic, 192600;Rippling muscle disease 2, 606072		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CBS	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CC2D1A	Mental retardation, autosomal recessive 9, 608443		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CC2D2A	Meckel syndrome 6, 612284;COACH syndrome, 216360;Joubert syndrome 9, 612285		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CCDC103	Ciliary dyskinesia, primary, 17, 614679		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CCDC22	Ritscher-Schinzel syndrome 2, 300963		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
CCDC39	Ciliary dyskinesia, primary, 14, 613807		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CCDC40	Ciliary dyskinesia, primary, 15, 613808		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CCDC8	3-M syndrome 3, 614205		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CCDC88C	Hydrocephalus, congenital, 1, 236600;?Spinocerebellar ataxia 40		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CCT5	Neuropathy, hereditary sensory, with spastic paraplegia, 256840		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD19	Immunodeficiency, common variable, 3, 613493		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD247	Immunodeficiency 25, 610163		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD27	Lymphoproliferative syndrome 2, 615122		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD3D	Immunodeficiency 19, 615617		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD3E	Immunodeficiency 18, SCID variant, 615615;Immunodeficiency 18, 615615		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD3G	Immunodeficiency 17, CD3 gamma deficient, 615607		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD40	Immunodeficiency with hyper-IgM, type 3, 606843		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD40LG	Immunodeficiency, X-linked, with hyper-IgM, 308230		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
CD55	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD59	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD79A	Agammaglobulinemia 3, 613501		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD79B	Agammaglobulinemia 6, 612692		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD81	Immunodeficiency, common variable, 6, 613496		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CD8A	CD8 deficiency, familial, 608957		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CDAN1	Dyserythropoietic anemia, congenital, type Ia, OMIM:224120;Anemia, congenital dyserythropoietic, type Ia, MONDO:0009135		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CDH1	Blepharocheiloodontic syndrome 1, 119580		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CDH15	Mental retardation, autosomal dominant 3, 612580		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CDH23	Usher syndrome, type 1D, 601067;Usher syndrome, type 1DF digenic, 601067;Deafness, autosomal recessive 12, 601386		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHAT	Myasthenic syndrome, congenital, 6, presynaptic, 254210		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHD2	Epileptic encephalopathy, childhood-onset, 615369		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CHD3	Snijders Blok-Campeau syndrome, 618205		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CHD7	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CHD8	Autism susceptibility;Overgrowth with intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CHKB	Muscular dystrophy, congenital, megaconial type, 602541		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHM	Choroideremia, 303100		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
CHMP2B	Dementia, familial, nonspecific, 600795;Amyotrophic lateral sclerosis 17, 614696		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CHRDL1	Megalocornea 1, X-linked, 309300		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
CHRNA1	Multiple pterygium syndrome, lethal type, 253290;Myasthenic syndrome, congenital, 1B, fast-channel, 608930;Myasthenic syndrome, congenital, 1A, slow-channel, 601462		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHRNA2	Epilepsy, nocturnal frontal lobe, type 4, 610353		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CHRNB1	Myasthenic syndrome, congenital, 2A, slow-channel, 616313;?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHRNB2	Epilepsy, nocturnal frontal lobe, 3, 605375		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CHRND	?Myasthenic syndrome, congenital, 3A, slow-channel, 616321;?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323;Multiple pterygium syndrome, lethal type, 253290;Myasthenic syndrome, congenital, 3B, fast-channel, 616322		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHRNE	Myasthenic syndrome, congenital, 4B, fast-channel, 616324;Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931;Myasthenic syndrome, congenital, 4A, slow-channel, 605809		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHRNG	Multiple pterygium syndrome, lethal type, 253290;Escobar syndrome, 265000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1, 601776		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHST6	Macular corneal dystrophy, 217800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHSY1	Tarsal-carpal brachydactyly syndrome, 605282		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CHUK	Cocoon syndrome, 613630		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CIB2	Usher syndrome, type IJ, 614869;Deafness, autosomal recessive 48, 609439		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CIC	Mental retardation, autosomal dominant 45, 617600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CIITA	Bare lymphocyte syndrome, type II, complementation group A, 209920		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CISD2	Wolfram syndrome 2, 604928		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CLCN2	Leukoencephalopathy with ataxia, 615651;Hyperaldosteronism, familial, type II, 605635;{Epilepsy, juvenile absence, susceptibility to, 2}, 607628;{Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628;{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CLCN5	Nephrolithiasis, type I, 310468;Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990;Hypophosphatemic rickets, 300554;Dent disease, 300009		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
CLCN7	Osteopetrosis, autosomal dominant 2, 166600;Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CLDN14	Deafness, autosomal recessive 29, 614035		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CLDN16	Hypomagnesemia 3, renal, 248250		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CLN5	Ceroid lipofuscinosis, neuronal, 5 OMIM:256731;neuronal ceroid lipofuscinosis 5 MONDO:0009745		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CLN6	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300;Ceroid lipofuscinosis, neuronal, 6, 601780		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CLN8	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CLRN1	Usher syndrome, type 3A, 276902;Retinitis pigmentosa 61, 614180		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CNBP	Myotonic dystrophy 2, OMIM:602668;Myotonic dystrophy type 2, MONDO:0011266		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CNGB3	Achromatopsia 3, 262300;Macular degeneration, juvenile, 248200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CNKSR2	Mental retardation, X-linked, syndromic, Houge type, 301008		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
CNNM2	Hypomagnesemia, seizures, and mental retardation, 616418;Hypomagnesemia 6, renal, 613882		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CNOT3	CNOT3 syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CNPY3	Epileptic encephalopathy, early infantile, 60, 617929		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome, 610042;Pitt-Hopkins like syndrome 1, 610042;{Autism susceptibility 15}, 612100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COCH	Deafness		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COG1	Congenital disorder of glycosylation, type IIg, 611209		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COG4	Congenital disorder of glycosylation, type IIi, 613489;Saul-Wilson syndrome, 618150		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COG5	Congenital disorder of glycosylation, type IIi, 613612		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COG6	Shaheen syndrome, 615328;Congenital disorder of glycosylation, type IIi, 614576		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COG7	Congenital disorder of glycosylation, type IIe, 608779		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COG8	Congenital disorder of glycosylation, type IIh, 611182		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL10A1	Metaphyseal chondrodysplasia, Schmid type, 156500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
COL11A1	Fibrochondrogenesis 1, 228520;?Deafness, autosomal dominant 37, 618533;Stickler syndrome, type II, 604841;Marshall syndrome, 154780		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL11A2	Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840;Deafness, autosomal recessive 53, 609706;Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL12A1	?Ullrich congenital muscular dystrophy 2, 616470;Bethlem myopathy 2, 616471		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL17A1	Epidermolysis bullosa, junctional, localisata variant, 226650;Epithelial recurrent erosion dystrophy, 122400;Epidermolysis bullosa, junctional, non-Herlitz type, 226650		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL18A1	Knobloch syndrome, type 1, 267750		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL1A1	Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060;Osteogenesis imperfecta, type I, 166200;Osteogenesis imperfecta, type III, 259420;Caffey disease, 114000;Osteogenesis imperfecta, type II, 166210;Osteogenesis imperfecta, type IV, 166220		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
COL1A2	Osteogenesis imperfecta, type III, 259420;Ehlers-Danlos syndrome, cardiac valvular type, 225320;Osteogenesis imperfecta, type I, 617820;Osteogenesis imperfecta, type IV, 166220		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL2A1	Achondrogenesis, type II or hypochondrogenesis, 200610;Spondyloepiheneal dysplasia, 271700;SMED Strudwick type, 184250;Stickler syndrome, type I, nonsyndromic ocular, 609508;Vitreoretinopathy with phalangeal epiphyseal dysplasia;Kniest dysplasia, 156550;SED congenita, 183900;Czech dysplasia, 609162;Platyspondylic skeletal dysplasia, Torrance type, 151210;Epiphy		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
COL3A1	Polymicrogyria with or without vascular-type EDS, 618343;Ehlers-Danlos syndrome, vascular type, 130050		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773;Microangiopathy and leukoencephalopathy, porencephaly of, 180000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
COL4A2	Brain small vessel disease 2, 614483		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
COL4A3	Alport syndrome 2, autosomal recessive, 203780;Alport syndrome 3, autosomal dominant, 104200;Hematuria, benign familial, 141200		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL4A4	Hematuria, familial benign, 141200;Alport syndrome 2, autosomal recessive, 203780		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL4A5	Alport syndrome 1, X-linked, 301050		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
COL5A1	Ehlers-Danlos syndrome, classic type, 1, 130000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
COL5A2	Ehlers-Danlos syndrome, classic type, 2, 130010		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
COL6A1	Bethlem myopathy 1, 158810;Ullrich congenital muscular dystrophy 1, 254090		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL6A2	Bethlem myopathy 1, 158810;?Myosclerosis, congenital, 255600;Ullrich congenital muscular dystrophy 1, 254090		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL6A3	Bethlem myopathy 1, 158810;Ullrich congenital muscular dystrophy 1, 254090;Dystonia 27, 616411		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL7A1	Epidermolysis bullosa dystrophica, AD, 131750;EBD inversa, 226600;EBD, localisata variant Epidermolysis bullosa pruriginosa, 604129;Toenail dystrophy, isolated, 607523;Epidermolysis bullosa dystrophica, AR, 226600;Epidermolysis bullosa, pretibial, 131850;Transient bullous of the newborn, 131705;EBD, Bart type, 132000		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL9A1	Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL9A2	?Stickler syndrome, type V, 614284;Epiphyseal dysplasia, multiple, 2, 600204		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
COLEC11	3MC syndrome 2, 265050		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COL0	Myasthenic syndrome, congenital, 5, 603034		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COMP	Pseudoachondroplasia, 177170;Epiphyseal dysplasia, multiple, 1, 132400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COQ4	Coenzyme Q10 deficiency, primary, 7, 616276		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COQ6	Coenzyme Q10 deficiency, primary, 6, 614650		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CORO1A	Immunodeficiency 8, 615401		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COX10	Leigh syndrome due to mitochondrial COX4 deficiency, 256000;Mitochondrial complex IV deficiency, 220110		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119;Leigh syndrome due to cytochrome c oxidase deficiency, 256000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COX6B1	Mitochondrial complex IV deficiency, nuclear type 7, OMIM 619051		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CP	Hemosiderosis, systemic, due to aceruloplasminemia, 604290;Cerebellar ataxia, 604290		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CPOX	Harderoporphyria, 121300;Coproporphyria, 121300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CPS1	Carbamoylphosphate synthetase I deficiency, 237300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CPT1A	CPT deficiency, hepatic, type IA, 255120		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CPT2	CPT II deficiency, myopathic, stress-induced, 255110;CPT II deficiency, infantile, 600649;CPT II deficiency, lethal neonatal, 608836		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CR2	Immunodeficiency, common variable, 7, 614699		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CRADD	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CRB1	Leber congenital amaurosis 8, 613835;Pigmented paravenous chorioretinal atrophy, 172870;Retinitis pigmentosa-12, 600105		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CREBBP	Rubinstein-Taubi syndrome 1, 180849;Menke-Hennekam syndrome 1, 618332		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CRLF1	Cold-induced sweating syndrome 1, 272430		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CRTAP	Osteogenesis imperfecta, type VII, 610682		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CRX	Cone-rod retinal dystrophy 2, 120970;Leber congenital amaurosis 7, 613829		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CRYAA	Cataract 9, multiple types, 604219		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CRYAB	Cataract 16, multiple types, 613763;Myopathy, myofibrillar, 2, 608810;Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869;Cardiomyopathy, dilated, 1II, 615184		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CRYBA1	Cataract 10, multiple types, 600881		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CRYBA4	Cataract 23, 610425		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CRYBB1	Cataract 17, multiple types, 611544		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CRYBB2	Cataract 3, multiple types, 601547		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CRYBB3	Cataract 22, 609741		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CRYGC	Cataract 2, multiple types, 604307		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CRYGD	Cataract 4, multiple types, 115700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820;Brain abnormalities, neurodegeneration, and dysostosis/sclerosis, 618476		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CSF2RA	Surfactant metabolism dysfunction, pulmonary, 4, 300770		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CSF2RB	Surfactant metabolism dysfunction, pulmonary, 5, 614370		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CSF3R	Neutropenia, severe congenital 7, autosomal recessive, 617014		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, 612199		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208

CTH	Homocysteine, total plasma, elevated;Cystathioninuria, 219500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CTLA4	Autoimmune lymphoproliferative syndrome, type V, 616100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CTNNB1	Exudative vitreoretinopathy 7, 617572;Neurodevelopmental disorder with spastic diplegia and visual defects, 615075		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CTNND1	Blepharocheilodontic syndrome 2, 617681		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CTNS	Cystinosis, atypical nephropathic, 219800;Cystinosis, late-onset juvenile or adolescent nephropathic, 219900;Cystinosis, ocular nonnephropathic, 219750;Cystinosis, nephropathic, 219800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CTSA	Galactosialidosis, 256540		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CTSC	Papillon-Lefevre syndrome, 245000;Periodontitis 1, juvenile, 170650;Haim-Munk syndrome, 245010		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CTSK	Pycnodysostosis, 265800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CUBN	Megaloblastic anemia-1, Finnish type, 261100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
CUL7	3-M syndrome 1, 273750		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CXCR4	Myelokathexis, isolated;WHIM syndrome, 193670		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CYB5R3	Methemoglobinemia, type I, 250800;Methemoglobinemia, type II, 250800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CYBB	Immunodeficiency 34, mycobacteriosis, X-linked, 300645;Chronic granulomatous disease, X-linked, 306400		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
CYCS	Thrombocytopenia 4, 612004		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CYP11B1	Aldosteronism, glucocorticoid-remediable, 103900;Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CYP17A1	17-alpha-hydroxylase/17,20-lyase deficiency, 202110;17,20-lyase deficiency, isolated, 202110		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CYP1B1	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300;Anterior segment dysgenesis 6, multiple subtypes, 617315		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CYP21A2	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910;Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CYP27A1	Cerebrotendinous xanthomatosis, 213700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CYP27B1	Vitamin D-dependent rickets, type I, 264700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CYP4F22	Ichthyosis, congenital, autosomal recessive 5, 604777		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CYP7B1	Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
D2HGDH	D-2-hydroxyglutaric aciduria, 600721		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DAG1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DBH	Orthostatic hypotension 1, due to DBH deficiency, 223360		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DBT	Maple syrup urine disease, type II, 248600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DCAF17	Woodhouse-Sakati syndrome, 241080		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DCC	Mirror movements 1 and/or agenesis of the corpus callosum, 157600;Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DCDC2	?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394;Nephronophthisis 19, 616217		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DCLRE1C	Omenn syndrome, 603554;Severe combined immunodeficiency, Athabascan type, 602450		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DCTN1	Perry syndrome, 168605;Neuropathy, distal hereditary motor, type VIIB, 607641		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DCX	Subcortical laminal heterotopia, X-linked, 300067;Lissencephaly, X-linked, 300067		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
DDB2	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DDC	Aromatic L-amino acid decarboxylase deficiency, OMIM:608643;Aromatic L-amino acid decarboxylase deficiency, MONDO:0012084		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DDR2	Warburg-Cinotti syndrome, 618175;Spondylometaepiphyseal dysplasia, short limb-hand type, 271665		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DDX11	Warsaw breakage syndrome, 613398		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171;Mental retardation, autosomal dominant 24, 615828		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DES	Myopathy, myofibrillar, 1, 601419;Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DFNA5	Deafness, autosomal dominant 5, 600994		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DFNB59	Deafness, autosomal recessive 59, 610220		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070;Portal hypertension, noncirrhotic, 617068		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DHCR24	Desmosterolosis, 602398		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DHCR7	Smith-Lemli-Opitz syndrome, 270400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DHDDS	?Congenital disorder of glycosylation, type 1bb, 613861;Developmental delay and seizures with or without movement abnormalities, 617836;Retinitis pigmentosa 59, 613861		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DHODH	Miller syndrome, 263750		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DIAPH1	Deafness, autosomal dominant 1, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DIS3L2	Perlman syndrome, 267000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DISP1	Holoprosencephaly		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DKC1	Dyskeratosis congenita, X-linked, 305000		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DLG3	Mental retardation, X-linked 90, 300850		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
DLL3	Spondylocostal dysostosis 1, autosomal recessive, 277300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DLX3	Trichodontoosseous syndrome, 190320;Amelogenesis imperfecta, type IV, 104510		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DLX5	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DMD	Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
DMP1	Hypophosphatemic rickets, AR, 241520		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DMPK	Myotonic dystrophy 1, OMIM:160900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DNAAF1	Ciliary dyskinesia, primary, 13, 613193		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DNAAF3	Ciliary dyskinesia, primary, 2, 606763		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DNAH11	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DNAH9	Ciliary dyskinesia, primary, 40, 618300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DNAJB6	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DNAJC19	3-methylglutaconic aciduria, type V, 610198		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DNASE2	Autoinflammatory-pancytopenia syndrome, OMIM:619858		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DNM1	Epileptic encephalopathy, early infantile, 31, 616346		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DNM1L	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DNM2	Lethal congenital contracture syndrome 5, 615368;Charcot-Marie-Tooth disease, dominant intermediate B, 606482;Charcot-Marie-Tooth disease, axonal type 2M, 606482;Centronuclear myopathy 1, 160150		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121;Neuropathy, hereditary sensory, type IE, 614116		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DNMT3A	Tatton-Brown-Rahman syndrome, 615879		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DOCK3	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia OMIM:618292;neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia MONDO:0032661		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DOCK6	Adams-Oliver syndrome 2, 614219		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DOK7	?Fetal akinesia deformation sequence 3, 618389;Myasthenic syndrome, congenital, 10, 254300		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DOLK	Congenital disorder of glycosylation, type Im, 610768		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DPAGT1	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DPM1	Congenital disorder of glycosylation, type Ie, 608799		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DPM3	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DPYD	5-fluorouracil toxicity, 274270;Dihydropyrimidine dehydrogenase deficiency, 274270		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DPYS	Dihydropyrimidinuria, 222748		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DSP	Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821;Skin fragility-woolly hair syndrome, 607655;Arrhythmogenic right ventricular dysplasia 8, 607450;Keratosis palmoplantaris striata II, 612908;Epidermolysis bullosa, lethal acantholytic, 609638		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DSPP	Dentinogenesis imperfecta, Shields type II, 125490;Dentinogenesis imperfecta, Shields type III, 125500;Deafness, autosomal dominant 39, with dentinogenesis, 605594;Dentin dysplasia, type II, 125420		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DTNBP1	Hermansky-Pudlak syndrome 7, 614076		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DUOX2	Thyroid dyshormonogenesis 6, 607200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DYM	Dyggve-Melchior-Clausen disease, 223800;Smith-McCort dysplasia, 607326		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DYNC1H1	Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600;Mental retardation, autosomal dominant 13, 614563;Charcot-Marie-Tooth disease, axonal, type 20, 614228		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DYRK1A	Mental retardation, autosomal dominant 7, 614104		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
DYSF	Myopathy, distal, with anterior tibial onset, 606768;Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601;Miyoshi muscular dystrophy 1, 254130		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EARS2	Combined oxidative phosphorylation deficiency 12, 614924		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EBP	Chondrodysplasia punctata, X-linked dominant, 302960;MEND syndrome, 300960		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
EDA	Tooth agenesis, selective, X-linked 1, 313500;Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
EDAR	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490;Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EDARADD	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941;Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EDNRA	Mandibulofacial dysostosis with alopecia, 616367		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
EDNRB	Waardenburg syndrome, type 4A, 277580;ABCD syndrome, 600501		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EFHC1	{Epilepsy, juvenile absence, susceptibility to, 1}, 607631;{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
EFNB1	Craniofrontonasal dysplasia, 304110		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type, 610536		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
EGR2	Charcot-Marie-Tooth disease, type 1D, 607678;Dejerine-Sottas disease, 145900;Hypomyelinating neuropathy, congenital, 1, 605253		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EHMT1	Kleefstra syndrome 1, 610253		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
EIF2AK3	Wolcott-Rallison syndrome, 226980		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EIF2B1	Leukoencephalopathy with vanishing white matter, 603896		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EIF2B2	Leukoencephalopathy with vanishing white matter, 603896;Ovarioleukodystrophy, 603896		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EIF2B3	Leukoencephalopathy with vanishing white matter, 603896		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EIF2B4	Leukoencephalopathy with vanishing white matter, 603896;Ovarioleukodystrophy, 603896		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EIF2B5	Leukoencephalopathy with vanishing white matter, 603896;Ovarioleukodystrophy, 603896		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ELAC2	Combined oxidative phosphorylation deficiency 17, 615440		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ELANE	Neutropenia, severe congenital 1, autosomal dominant, 202700;Neutropenia, cyclic, 162800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ELN	Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ELOVL4	Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and mental retardation, 614457;Spinocerebellar ataxia 34, 133190		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ELP2	Mental retardation, autosomal recessive 58, 617270		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
EMX2	Schizencephaly, 269160		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ENG	Telangiectasia, hereditary hemorrhagic, type 1, 187300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ENO3	Glycogen storage disease XIII, 612932		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ENPP1	Arterial calcification, generalized, of infancy, 1, 208000;Cole disease, 615522;Hypophosphatemic rickets, autosomal recessive, 2, 613312		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ENTPD1	Spastic paraplegia 64, autosomal recessive, 615683		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EP300	Rubinstein-Taybi syndrome 2, 613684;Menke-Hennekam syndrome 2, 618333		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EPB41	Elliptocytosis-1, 611804		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EPB42	Spherocytosis, type 5, 612690		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora), 254780		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ERCC1	Cerebrooculofacioskeletal syndrome 4, 610758		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ERCC2	?Cerebrooculofacioskeletal syndrome 2, 610756;Trichothiodystrophy 1, photosensitive, 601675;Xeroderma pigmentosum, group D, 278730		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ERCC3	Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ERCC4	Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, type F/Cockayne syndrome, 278760		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ERCC5	Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780;Xeroderma pigmentosum, group G, 278780		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ERCC6	Cerebrooculofacioskeletal syndrome 1, 214150;UV-sensitive syndrome 1, 600630;De Sanctis-Cacchione syndrome, 278800;Cockayne syndrome, type B, 133540;Premature ovarian failure 11, 616946		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ERCC8	Cockayne syndrome, type A, 216400;UV-sensitive syndrome 2, 614621		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ERLIN2	Spastic paraplegia 18, autosomal recessive, 611225		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ESCO2	SC phocomelia syndrome, 269000;Roberts syndrome, 268300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ESRRB	Deafness, autosomal recessive 35, 608565		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ETFA	Glutaric acidemia IIA, OMIM:231680;multiple acyl-CoA dehydrogenase deficiency, MONDO:0009282		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ETFB	Glutaric acidemia IIB, OMIM:231680;multiple acyl-CoA dehydrogenase deficiency, MONDO:0009282		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ETFDH	Glutaric acidemia IIC, 231680		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ETHE1	Ethylmalonic encephalopathy, 602473		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EVC	Ellis-van Creveld syndrome, 225500;?Weyers acrofacial dysostosis, 193530		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EVC2	Ellis-van Creveld syndrome, 225500;Weyers acrofacial dysostosis, 193530		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EXT1	Chondrosarcoma, 215300;Exostoses, multiple, type 1, 133700		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EXT2	Exostoses, multiple, type 2, 133701;Seizures, scoliosis, and macrocephaly syndrome, 616682		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
EYA1	?Otofaciocervical syndrome, 166780;Branchiootorenal syndrome 1, with or without cataracts, 113650;Anterior segment anomalies with or without cataract, 602588;Branchiootic syndrome 1, 602588		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
EYA4	?Cardiomyopathy, dilated, 1J, 605362;Deafness, autosomal dominant 10, 601316		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
EZH2	Weaver syndrome, 277590		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
F10	Factor X deficiency, 227600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
F11	Factor XI deficiency, autosomal recessive, 612416;Factor XI deficiency, autosomal dominant, 612416		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
F12	Angioedema, hereditary, 3, OMIM:610618;Factor XII deficiency, OMIM:234000		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
F13A1	Factor XIIIA deficiency, 613225		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
F13B	Factor XIIIB deficiency, 613235		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
F2	Dysprothrombinemia, 613679;Thrombophilia due to thrombin defect, 188050;Hypoprothrombinemia, 613679		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
F5	Factor V deficiency, 227400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
F7	Factor VII deficiency, 227500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
F8	Hemophilia A, 306700		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
F9	Thrombophilia, X-linked, due to factor IX defect, 300807;Hemophilia B, 306900		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
FA2H	Spastic paraplegia 35, autosomal recessive, 612319		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FADD	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FAH	Tyrosinemia, type I, 276700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FAM161A	Retinitis pigmentosa 28, 606068		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FAM20A	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FAM20C	Raine syndrome, 259775		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FAM58A	STAR syndrome, 300707		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
FANCA	Fanconi anemia, complementation group A, 227650		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FANCB	Fanconi anemia, complementation group B, 300514		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
FANCC	Fanconi anemia, complementation group C, 227645		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208

FANCD2	Fanconi anemia, complementation group D2, 227646		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FANCE	Fanconi anemia, complementation group E, 600901		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FANCF	Fanconi anemia, complementation group F, 603467		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FANCG	Fanconi anemia, complementation group G, 614082		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FANCI	Fanconi anemia, complementation group I, 609053		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FANCL	Fanconi anemia, complementation group L, 614083		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FARS2	Spastic paraplegia 77, autosomal recessive, 617046;Combined oxidative phosphorylation deficiency 14, 614946		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FAS	Autoimmune lymphoproliferative syndrome, type IA, 601859		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FASLG	Autoimmune lymphoproliferative syndrome, type IB, 601859		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FBLN5	Macular degeneration, age-related, 3, 608895;Cutis laxa, autosomal recessive, type IA, 219100;Neuropathy, hereditary, with or without age-related macular degeneration, 608895;?Cutis laxa, autosomal dominant 2, 614434		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FBN1	Weill-Marchesani syndrome 2, dominant, 608328;Ectopia lentis, familial, 129600;Geleophysic dysplasia 2, 614185;Marfan lipodystrophy syndrome, 616914;Stiff skin syndrome, 184900;Marfan syndrome, 154700;MASS syndrome, 604308;Acromicric dysplasia, 102370		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FBN2	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FBP1	Fructose-1,6-bisphosphatase deficiency, 229700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FBXO7	Parkinson disease 15, autosomal recessive, 260300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FCGR3A	Immunodeficiency 20, 615707		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FCGR3B	Neutropenia, alloimmune neonatal		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FCN3	Immunodeficiency due to ficolin 3 deficiency, 613860		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FECH	Protoporphyria, erythropoietic, 1, 177000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FERMT3	Leukocyte adhesion deficiency, type III, 612840		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FGA	Dysfibrinogenemia, congenital, 616004;Hypodysfibrinogenemia, congenital, 616004;Amyloidosis, familial visceral, 105200;Afibrinogenemia, congenital, 202400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FGB	Dysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400;Hypofibrinogenemia, congenital, 202400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FGD1	Mental retardation, X-linked syndromic 16, 305400;Aarskog-Scott syndrome, 305400		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
FGD4	Charcot-Marie-Tooth disease, type 4H, 609311		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FGF10	LADD syndrome, 149730;Aplasia of lacrimal and salivary glands, 180920		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FGF14	Spinocerebellar ataxia 27, 609307		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FGF23	Hypophosphatemic rickets, autosomal dominant, 193100;Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993;Osteomalacia, tumor-induced		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FGF8	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FGFR1	Pfeiffer syndrome, 101600;Osteoglophonic dysplasia, 166250;Hypogonadotropic hypogonadism 2 with or without anosmia, 147950;Hartsfield syndrome, 615465;Jackson-Weiss syndrome, 123150;Trigonocephaly 1, 190440		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FGFR2	Saethre-Chotzen syndrome, 101400;Apert syndrome, 101200;Beare-Stevenson cutis gyrata syndrome, 123790;Pfeiffer syndrome, 101600;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410;Crouzon syndrome, 123500;Scaphocephaly and Axenfeld-Rieger anomaly;Craniofacial-skeletal-dermatologic dysplasia, 101600;Bent bone dysplasia sy		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FGFR3	Thanatophoric dysplasia, type II, 187601;Muenke syndrome, 602849;SADDAN, 616482;Crouzon syndrome with acanthosis nigricans, 612247;CATSHL syndrome, 610474;Achondroplasia, 100800;LADD syndrome, 149730;Thanatophoric dysplasia, type I, 187600;Hypochondroplasia, 146000		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FGG	Dysfibrinogenemia, congenital, 616004;Hypodysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400;Hypofibrinogenemia, congenital, 202400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FH	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FHL1	?Uruguay faciocardiomusculoskeletal syndrome, 300280;Emery-Dreifuss muscular dystrophy 6, X-linked, 300696;Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717;Myopathy, X-linked, with postural muscle atrophy, 300696;Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718;Scapuloperoneal myopathy, X-linked d		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
FIG4	Charcot-Marie-Tooth disease, type 4J, 611228;Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FKBP10	Bruck syndrome 1, 259450;Osteogenesis imperfecta, type XI, 610968		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FKBP14	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FKTN	Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800;Cardiomyopathy, dilated, 1X, 611615;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FLCN	Pneumothorax, primary spontaneous, 173600;Birt-Hogg-Dube syndrome, 135150		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FLNA	Melnick-Needles syndrome, 309350;Congenital short bowel syndrome, 300048;Terminal osseous dysplasia, 300244;Intestinal pseudoobstruction, neuronal, 300048;?FG syndrome 2, 300321;Otopalatodigital syndrome, type II, 304120;Heterotopia, periventricular, 1, 300049;Cardiac valvular dysplasia, X-linked, 314400;Frontometaphyseal dysplasia 1, 305620;Otopalatodigital synd		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
FLNB	Spondylocarpotarsal synostosis syndrome, 272460;Atelosteogenesis, type III, 108721;Larsen syndrome, 150250;Boomerang dysplasia, 112310;Atelosteogenesis, type I, 108720		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FLNC	Cardiomyopathy, familial hypertrophic, 26, OMIM:617047;Cardiomyopathy, familial restrictive 5, OMIM:617047;Hypertrophic cardiomyopathy 26, MONDO:0014883;Myopathy, distal, 4, OMIM:614065;Distal myopathy with posterior leg and anterior hand involvement, MONDO:0013550;Myopathy, myofibrillar, 5, OMIM:609524;Myopathy, myofibrillar, 5, MONDO:0012289		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FLT4	Lymphatic malformation 1, 153100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FLVCR2	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FMN2	Mental retardation, autosomal recessive 47, 616193		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FMO3	Trimethylaminuria, 602079		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FMR1	Fragile X syndrome, 300624;Premature ovarian failure 1, 311360;Fragile X tremor/ataxia syndrome, 300623		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
FN1	Plasma fibronectin deficiency, 614101;Glomerulopathy with fibronectin deposits 2, 601894;Spondylometaphyseal dysplasia, corner fracture type, 184255		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FOXC1	Anterior segment dysgenesis 3, multiple subtypes, 601631;Axenfeld-Rieger syndrome, type 3, 602482		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FOXC2	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400;Lymphedema-distichiasis syndrome, 153400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FOXE1	Bamforth-Lazarus syndrome, 241850		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FOXE3	Cataract 34, multiple types, 612968;Anterior segment dysgenesis 2, multiple subtypes, 610256		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FOXG1	Rett syndrome, congenital variant, 613454		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FOXP1	Mental retardation with language impairment and with or without autistic features, 613670		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FOXP2	Speech-language disorder-1, 602081		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19, 618241		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FRAS1	Fraser syndrome 1, 219000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FREM1	Trigonocephaly 2, 614485;Manitoba oculotrichoanal syndrome, 248450;Bifid nose with or without anorectal and renal anomalies, 608980		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FREM2	Fraser syndrome 2, 617666;Cryptophthalmos, unilateral or bilateral, isolated, 123570		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FRMD7	Nystagmus 1, congenital, X-linked, 310700;Nystagmus, infantile periodic alternating, X-linked, 310700		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
FRMPD4	Mental retardation, X-linked 104, 300983		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
FTCD	Glutamate formiminotransferase deficiency, 229100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FTL	Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FTSJ1	Mental retardation, X-linked 9/44, 309549		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
FUCA1	Fucosidosis, 230000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FUT8	Congenital disorder of glycosylation with defective fucosylation 1, 618005		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FXN	Friedreich ataxia OMIM:229300;Friedreich ataxia with retained reflexes OMIM:229300;Friedreich ataxia 1 MONDO:0100340		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FYCO1	Cataract 18, autosomal recessive, 610019		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FZD6	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
G6PC	Glycogen storage disease Ia, 232200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
G6PC3	Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
G6PD	Hemolytic anemia, G6PD deficient (favism), 300908		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
GAA	Glycogen storage disease II, 232300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GABRA1	{Epilepsy, childhood absence, susceptibility to, 4}, 611136;Epileptic encephalopathy, early infantile, 19, 615744;{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GABRB3	Epileptic encephalopathy, early infantile, 43, 617113		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GABRG2	Epileptic encephalopathy, early infantile, 74, 618396;Epilepsy, generalized, with febrile seizures plus, type 3, 607681;Febrile seizures, familial, 8, 607681		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GALC	Krabbe disease, 245200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GALE	Galactose epimerase deficiency, 230350		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GALK1	Galactokinase deficiency with cataracts, 230200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GALNS	Mucopolysaccharidosis IVA, 253000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GALT	Galactosemia, 230400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GAMT	Cerebral creatine deficiency syndrome 2, 612736		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GAN	Giant axonal neuropathy-1, 256850		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GARS	Charcot-Marie-Tooth disease, type 2D, 601472;Neuropathy, distal hereditary motor, type VA, 600794		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GATA1	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835;Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367;Thrombocytopenia with beta-thalassemia, X-linked, 314050		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
GATA2	Immunodeficiency 21, 614172;Emberger syndrome, 614038		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GATA4	Tetralogy of Fallot, 187500;Atrial septal defect 2, 607941;?Testicular anomalies with or without congenital heart disease, 615542;Atrioventricular septal defect 4, 614430;Ventricular septal defect 1, 614429		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GATA6	Persistent truncus arteriosus, 217095;Tetralogy of Fallot, 187500;Atrioventricular septal defect 5, 614474;Atrial septal defect 9, 614475;Pancreatic agenesis and congenital heart defects, 600001		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GATM	Cerebral creatine deficiency syndrome 3, 612718		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GBA	Gaucher disease, perinatal lethal, 608013;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, type IIIC, 231005;Gaucher disease, type II, 230900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GBE1	Polyglucosan body disease, adult form, 263570;Glycogen storage disease IV, 232500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GCDH	Glutaricaciduria, type I, 231670		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GCK	Diabetes mellitus, permanent neonatal, 606176;MODY, type II, 125851;Hyperinsulinemic hypoglycemia, familial, 3, 602485;Diabetes mellitus, noninsulin-dependent, late onset, 125853		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GCLC	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GDAP1	Charcot-Marie-Tooth disease, axonal, type 2K, OMIM:607831;Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, OMIM:607706;Charcot-Marie-Tooth disease, recessive intermediate, A, OMIM:608340;Charcot-Marie-Tooth disease, type 4A, OMIM:214400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GDF5	Du Pan syndrome, 228900;Symphalangism, proximal, 1B, 615298;Chondrodysplasia, Grebe type, 200700;Brachydactyly, type A1, C, 615072;?Acromesomelic dysplasia, Hunter-Thompson type, 201250;Multiple synostoses syndrome 2, 610017;Brachydactyly, type A2, 112600;Brachydactyly, type C, 113100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GDF6	Leber congenital amaurosis 17, 615360;Multiple synostoses syndrome 4, 617898;Microphthalmia, isolated 4, 613094;Klippel-Feil syndrome 1, autosomal dominant, 118100;Microphthalmia with coloboma 6, digenic, 613703		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GDI1	Mental retardation, X-linked 41, 300849		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
GFAP	Alexander disease, 203450		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GFI1B	Bleeding disorder, platelet-type, 17, 187900		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GFM1	Combined oxidative phosphorylation deficiency 1, 609060		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GFPT1	Myasthenia, congenital, 12, with tubular aggregates, 610542		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GGCX	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842;Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GH1	Growth hormone deficiency, isolated, type IA, 262400;Kowarski syndrome, 262650;Growth hormone deficiency, isolated, type IB, 612781;Growth hormone deficiency, isolated, type II, 173100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GHR	Laron dwarfism, 262500;Increased responsiveness to growth hormone, 604271;Growth hormone insensitivity, partial, 604271		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GHRHR	Growth hormone deficiency, isolated, type IV, 618157		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GIF	Intrinsic factor deficiency, 261000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GIPC3	Deafness, autosomal recessive 15, 601869		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GJA1	Oculodentodigital dysplasia, autosomal recessive, 257850;Syndactyly, type III, 186100;Craniometaphyseal dysplasia, autosomal recessive, 218400;Palmoplantar keratoderma with congenital alopecia, 104100;Atrioventricular septal defect 3, 600309;Erythrokeratodermia variabilis et progressiva 3, 617525;Hypoplastic left heart syndrome 1, 241550;Oculodentodigital dysplasia, 16		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GJA3	Cataract 14, multiple types, 601885		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GJA8	Cataract 1, multiple types, 116200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
GJB2	Deafness, autosomal recessive 1A, 220290;Keratoderma, palmoplantar, with deafness, 148350;Vohwinkel syndrome, 124500;Deafness, autosomal dominant 3A, 601544;Hystrix-like ichthyosis with deafness, 602540;Bart-Pumphrey syndrome, 149200;Keratitis-ichthyosis-deafness syndrome, 148210		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GJB3	Deafness, autosomal dominant, with peripheral neuropathy;Deafness, autosomal recessive;Deafness, digenic, GJB2/GJB3, 220290;Erythrokeratodermia variabilis et progressiva 1, 133200;Deafness, autosomal dominant 2B, 612644		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GJC2	Spastic paraplegia 44, autosomal recessive, 613206;Lymphatic malformation 3, 613480;Leukodystrophy, hypomyelinating, 2, 608804		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GK	Glycerol kinase deficiency, 307030		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
GLA	Fabry disease, cardiac variant, 301500;Fabry disease, 301500		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
GLB1	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GLDC	Glycine encephalopathy, 605899		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GLE1	Congenital arthrogryposis with anterior horn cell disease, 611890;Lethal congenital contracture syndrome 1, 253310		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GLI1	Polydactyly, preaxial I, 174400;Polydactyly, postaxial, type A8, 618123		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GLI2	Holoprosencephaly 9, 610829;Culler-Jones syndrome, 615849		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GLI3	Pallister-Hall syndrome, 146510;Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, preaxial, type IV, 174700;Polydactyly, postaxial, types A1 and B, 174200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GLMN	Glomuvenous malformations, 138000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GLRA1	Hyperekplexia 1, 149400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GLRB	Hyperekplexia 2, 614619		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GLRX5	Spasticity, childhood-onset, with hyperglycinemia, 616859;Anemia, sideroblastic, 3, pyridoxine-refractory, 616860		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GLUD1	Hyperinsulinism-hyperammonemia syndrome, 606762		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GLUL	Glutamine deficiency, congenital, 610015		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GLYCTK	D-glyceric aciduria, 220120		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GM2A	GM2-gangliosidosis, AB variant, 272750		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GNAS	Pseudopseudohypoparathyroidism, 612463;Pseudohypoparathyroidism Ia, 103580;Osseous heteroplasia, progressive, 166350;Pseudohypoparathyroidism Ic, 612462;ACTH-independent macronodular adrenal hyperplasia, 219080;Pseudohypoparathyroidism Ib, 603233		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GNB5	Intellectual developmental disorder with cardiac arrhythmia, 617173;Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GNE	Sialuria, 269921;Nonaka myopathy, 605820		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GNMT	Glycine N-methyltransferase deficiency, 606664		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GNPTAB	Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GNPTG	Mucolipidosis III gamma, 252605		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GNS	Mucopolysaccharidosis type IIID, 252940		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GORAB	Geroderma osteodysplasticum, 231070		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GOSR2	Epilepsy, progressive myoclonic 6, 614018		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GP1BA	Bernard-Soulier syndrome, type A2 (dominant), 153670;Bernard-Soulier syndrome, type A1 (recessive), 231200;von Willebrand disease, platelet-type, 177820		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GP1BB	Giant platelet disorder, isolated, 231200;Bernard-Soulier syndrome, type B, 231200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GP6	Bleeding disorder, platelet-type, 11, 614201		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GP9	Bernard-Soulier syndrome, type C, 231200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
GPC6	Omodysplasia 1, 258315		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GPD1	Hypertriglyceridemia, transient infantile, 614480		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GPHN	Molybdenum cofactor deficiency C, 615501		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GPR143	Ocular albinism, type I, Nettleship-Falls type, 300500;Nystagmus 6, congenital, X-linked, 300814		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
GPR179	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GPSM2	Chudley-McCullough syndrome, 604213		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GRHPR	Hyperoxaluria, primary, type II, 260000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GRIA3	Mental retardation, X-linked 94, 300699		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
GRIK2	Mental retardation, autosomal recessive, 6, 611092		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139;Mental retardation, autosomal dominant 6, 613970		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
GRIP1	Fraser syndrome 3, 617667		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GRM1	Spinocerebellar ataxia 44, 617691;Spinocerebellar ataxia, autosomal recessive 13, 614831		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GRM6	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GRN	Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706;Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GSC	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GSS	Hemolytic anemia due to glutathione synthetase deficiency, 231900;Glutathione synthetase deficiency, 266130		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208

GTF2H5	Trichothiodystrophy 3, photosensitive, 616395		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GUCY2C	Diarrhea 6, 614616;Meconium ileus, 614665		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GUSB	Mucopolysaccharidosis VII, 253220		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GYG1	Polyglucosan body myopathy 2, 616199;?Glycogen storage disease XV, 613507		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GYS1	Glycogen storage disease 0, muscle, 611556		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GYS2	Glycogen storage disease 0, liver, 240600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
H19	Silver-Russell syndrome, OMIM:180860;Wilms tumor 2, OMIM:194071;Beckwith-Wiedemann syndrome, OMIM:130650		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures, 616756		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530;Hyperinsulinemic hypoglycemia, familial, 4, 609975		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HADHA	Fatty liver, acute, of pregnancy, 609016;LCHAD deficiency, 609016;Trifunctional protein deficiency, 609015;HELLP syndrome, maternal, of pregnancy, 609016		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HADHB	Trifunctional protein deficiency, 609015		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HAMP	Hemochromatosis, type 2B, 613313		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HAX1	Neutropenia, severe congenital 3, autosomal recessive, 610738		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HBA1	Erythrocytosis, 7, 617981;Hemoglobin H disease, nondeletional, 613978;Heinz body anemias, alpha-, 140700;Thalassemias, alpha-, 604131;Methemoglobinemia, alpha type, 617973		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HBA2	Erythrocytosis 7, 617981;Hemoglobin H disease, deletional and nondeletional, 613978;Thalassemia, alpha-, 604131;Heinz body anemia, 140700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HBB	Delta-beta thalassemia, 141749;Methmoglobinemia, beta type, 617971;Heinz body anemia, 140700;Thalassemia, beta, 613985;Thalassemia-beta, dominant inclusion-body, 603902;Hereditary persistence of fetal hemoglobin, 141749;Erythrocytosis 6, 617980;Sickle cell anemia, 603903		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
HCN1	Epileptic encephalopathy, early infantile, 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HCN2	Febrile seizures		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HDAC4	Brachydactyly-mental retardation syndrome, 600430		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HDAC8	Cornelia de Lange syndrome 5, 300882		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926;Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HES7	Spondylocostal dysostosis 4, autosomal recessive, 613686		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HESX1	Pituitary hormone deficiency, combined, 5, 182230;Growth hormone deficiency with pituitary anomalies, 182230;Septooptic dysplasia, 182230		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HEXA	[Hex A pseudodeficiency], 272800;GM2-gangliosidosis, several forms, 272800;Tay-Sachs disease, 272800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HFE	Hemochromatosis, 235200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HFE2	Hemochromatosis, type 2A, 602390		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HGD	Alkaptonuria, 203500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HIBCH	3-hydroxyisobutyrl-CoA hydrolase deficiency, 250620		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HK1	Retinitis pigmentosa 79, 617460;Hemolytic anemia due to hexokinase deficiency, 235700;Neurodevelopmental disorder with visual defects and brain anomalies, 618547;Neuropathy, hereditary motor and sensory, Russe type, 605285		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HLCS	Holocarboxylase synthetase deficiency, 253270		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HMBS	Porphyria, acute intermittent, 176000;Porphyria, acute intermittent, nonerythroid variant, 176000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HMGCL	HMG-CoA lyase deficiency, 246450		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HMGCS2	HMG-CoA synthase-2 deficiency, 605911		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HNF1B	Diabetes mellitus, noninsulin-dependent, 125853;Renal cysts and diabetes syndrome, 137920		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HNF4A	MODY, type I, 125850;Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HNRNPU	Epileptic encephalopathy, early infantile, 54, 617391		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HOGA1	Hyperoxaluria, primary, type III, 613616		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HOXA1	Athabaskan brainstem dysgenesis syndrome, 601536;Bosley-Salih-Alorainy syndrome, 601536		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HOXA11	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HOXA13	Hand-foot-uterus syndrome, 140000;?Guttmacher syndrome, 176305		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HOXD13	?Brachydactyly-syndactyly syndrome, 610713;Synpolydactyly 1, 186000;Syndactyly, type V, 186300;Brachydactyly, type E, 113300;Brachydactyly, type D, 113200		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HPD	Tyrosinemia, type III, 276710;Hawkinsinuria, 140350		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HPGD	Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100;Cranioosteoarthropathy, 259100;Digital clubbing, isolated congenital, 119900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HPRT1	Lesch-Nyhan syndrome, 300322;HPRT-related gout, 300323		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
HPS1	Hermansky-Pudlak syndrome 1, 203300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HPS3	Hermansky-Pudlak syndrome 3, 614072		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HPS4	Hermansky-Pudlak syndrome 4, 614073		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HPS5	Hermansky-Pudlak syndrome 5, 614074		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HPS6	Hermansky-Pudlak syndrome 6, 614075		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HPSE2	Urofacial syndrome 1, 236730		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HR	Alopecia universalis, 203655;Atrichia with papular lesions, 209500;Hypotrichosis 4, 146550		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HRAS	Congenital myopathy with excess of muscle spindles, 218040;Costello syndrome, 218040		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HSD17B10	HSD10 mitochondrial disease, 300438		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
HSD17B3	Pseudohermaphroditism, male, with gynecomastia, 264300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HSD17B4	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HSD3B7	Bile acid synthesis defect, congenital, 1, 607765		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HSF4	Cataract 5, multiple types, 116800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HSPB1	Neuropathy, distal hereditary motor, type IIB, 608634;Charcot-Marie-Tooth disease, axonal, type 2F, 606595		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HSPB8	Charcot-Marie-Tooth disease, axonal, type 2L, 608673;Neuropathy, distal hereditary motor, type IIA, 158590		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HSPD1	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HSPG2	Schwartz-Jampel syndrome, type 1, OMIM:255800;Schwartz-Jampel syndrome, MONDO:0009717;Dyssegmental dysplasia, Silverman-Handmaker type, OMIM:224410;Silverman-Handmaker type dyssegmental dysplasia, MONDO:0009140		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HTRA1	CARASIL syndrome, 600142;Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HTRA2	3-methylglutaconic aciduria, type VIII, 617248		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HUWE1	Mental retardation, X-linked syndromic, Turner type, 309590		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
HYDIN	Ciliary dyskinesia, primary, 5, 608647		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HYLS1	Hydrolethalus syndrome, 236680		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ICK	{Epilepsy, juvenile myoclonic, susceptibility to, 10}, OMIM:617924;Endocrine-cerebroosteodysplasia, OMIM:612651;Endocrine-cerebro-osteodysplasia syndrome, MONDO:0012980		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ICOS	Immunodeficiency, common variable, 1, 607594		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IDH2	D-2-hydroxyglutaric aciduria 2, 613657		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
IDS	Mucopolysaccharidosis II, 309900		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
IDUA	Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih, 607014		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IFIH1	Singleton-Merten syndrome 1, 182250;Aicardi-Goutieres syndrome 7, 615846		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
IFITM5	Osteogenesis imperfecta, type V, 610967		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
IFNGR1	Immunodeficiency 27A, mycobacteriosis, AR, 209950;Immunodeficiency 27B, mycobacteriosis, AD, 615978		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IFNGR2	Immunodeficiency 28, mycobacteriosis, 614889		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IFT122	Cranioectodermal dysplasia 1, 218330		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IFT43	Short-rib thoracic dysplasia 18 with polydactyly, 617866;?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IGF1R	Insulin-like growth factor I, resistance to, 270450		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IGF2	Beckwith-Wiedemann Syndrome;Chromosome 11p15.5-Related Russell-Silver Syndrome		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
IGFALS	Acid-labile subunit, deficiency of, 615961		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IGHM	Agammaglobulinemia 1, 601495		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IGHMBP2	Neuronopathy, distal hereditary motor, type VI, 604320;Charcot-Marie-Tooth disease, axonal, type 2S, 616155		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IGLL1	Agammaglobulinemia 2, 613500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IHH	Acrocapitofemoral dysplasia, 607778;Brachydactyly, type A1, 112500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IKBKG	Invasive pneumococcal disease, recurrent isolated, 2, 300640;Incontinentia pigmenti, 308300;Immunodeficiency 33, 300636;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency, isolated, 300584;Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
IL10RA	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL10RB	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL11RA	Craniosynostosis and dental anomalies, 614188		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL12B	Immunodeficiency 29, mycobacteriosis, 614890		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL12RB1	Immunodeficiency 30, 614891		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL17RA	Immunodeficiency 51, 613953		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL1RAPL1	Mental retardation, X-linked 21/34, 300143		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
IL1RN	Interleukin 1 receptor antagonist deficiency, OMIM:612852		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL21R	Immunodeficiency 56, 615207		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL2RA	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL2RG	Combined immunodeficiency, X-linked, moderate, 312863;Severe combined immunodeficiency, X-linked, 300400		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
IL36RN	Psoriasis 14, pustular, OMIM:614204		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ILDR1	Deafness, autosomal recessive 42, 609646		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IMPAD1	Chondrodysplasia with joint dislocations, GPAPP type, 614078		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
INF2	Charcot-Marie-Tooth disease, dominant intermediate E, 614455;Glomerulosclerosis, focal segmental, 5, 613237		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
INSR	Leprechaunism, 246200;Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549;Rabson-Mendenhall syndrome, 262190;Hyperinsulinemic hypoglycemia, familial, 5, 609968		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
INVS	Nephronophthisis 2, infantile, 602088		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IQCB1	Senior-Loken syndrome 5, 609254		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IQSEC2	Mental retardation, X-linked 1/78, 309530		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
IRAK4	Invasive pneumococcal disease, recurrent isolated, 1, 610799;IRAK4 deficiency, 607676		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IRF6	van der Woude syndrome, 119300;Popliteal pterygium syndrome 1, 119500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
IRF8	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990;Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IRX5	Hamamy syndrome, 611174		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ISCU	Myopathy with lactic acidosis, hereditary, 255125		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ISPD	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITCH	Autoimmune disease, multisystem, with facial dysmorphism, 613385		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITGA2B	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800;Thrombocytopenia, neonatal alloimmune, BAK antigen related;Glanzmann thrombasthenia, 273800		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITGB2	Leukocyte adhesion deficiency, 116920		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITGB3	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800;Purpura, posttransfusion;Glanzmann thrombasthenia, 273800;Thrombocytopenia, neonatal alloimmune		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia, 226730;Epidermolysis bullosa of hands and feet, 131800;Epidermolysis bullosa, junctional, non-Herlitz type, 226650		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITK	Lymphoproliferative syndrome 1, 613011		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITPA	[Inosine triphosphatase deficiency], 613850;Epileptic encephalopathy, early infantile, 35, 616647		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITPR1	Spinocerebellar ataxia 15, 606658;Gillespie syndrome, 206700;Spinocerebellar ataxia 29, congenital nonprogressive, 117360		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ITSN2	Nephrotic syndrome		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IVD	Isovaleric acidemia, 243500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
JAG1	Alagille syndrome 1, 118450;?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Tetralogy of Fallot, 187500		MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	Research panel - Severe Paediatric Disorders			1.208
JAK3	SCID, autosomal recessive, T-negative/B-positive type, 600802		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
JPH2	Cardiomyopathy, hypertrophic, 17, 613873		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
JPH3	Huntington disease-like 2, OMIM:606438		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
JUP	Arrhythmogenic right ventricular dysplasia 12, 611528;Naxos disease, 601214		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KANSL1	Koolen-De Vries syndrome, 610443		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KARS	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641;Deafness, autosomal recessive 89, 613916		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KAT6B	Genitopatellar syndrome, 606170;SBBYSS syndrome, 603736		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KBTBD13	Nemaline myopathy 6, autosomal dominant, 609273		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KCNA1	Episodic ataxia/myokymia syndrome, 160120		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KCNC3	Spinocerebellar ataxia 13, 605259		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KCND3	Brugada syndrome 9, 616399;Spinocerebellar ataxia 19, 607346		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KCNE1	Long QT syndrome 5, 613695;Jervell and Lange-Nielsen syndrome 2, 612347		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KCNJ1	Bartter syndrome, type 2, 241200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KCNJ11	Diabetes mellitus, transient neonatal, 3, 610582;Maturity-onset diabetes of the young, type 13, 616329;Diabetes, permanent neonatal, with or without neurologic features, 606176;Hyperinsulinemic hypoglycemia, familial, 2, 601820		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KCNJ2	Short QT syndrome 3, OMIM:609622;Short QT syndrome type 3, MONDO:0012314;Atrial fibrillation, familial, 9, OMIM:613980;Atrial fibrillation, familial, 9, MONDO:0013513;Andersen syndrome, OMIM:170390;Andersen-Tawil syndrome, MONDO:0008222		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KCNJ6	Keppen-Lubinsky syndrome, 614098		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KCNK9	Birk-Barel mental retardation dysmorphism syndrome, 612292		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KCNMA1	Cerebellar atrophy, developmental delay, and seizures, OMIM:617643;Cerebellar atrophy, developmental delay, and seizures, MONDO:0060551;Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, OMIM:609446;Generalized epilepsy-paroxysmal dyskinesia syndrome, MONDO:0012276;Liang-Wang syndrome, OMIM:618729;Liang-Wang syndrome, MON		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KCNQ1	Short QT syndrome 2, 609621;Atrial fibrillation, familial, 3, 607554;Long QT syndrome 1, 192500;Jervell and Lange-Nielsen syndrome, 220400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KCNQ2	Epileptic encephalopathy, early infantile, 7, 613720;Seizures, benign neonatal, 1, 121200;Myokymia, 121200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KCNQ3	Seizures, benign neonatal, 2, 121201		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KCNQ4	Deafness, autosomal dominant 2A, 600101		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
KDM6A	Kabuki syndrome 2, 300867		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KIF1A	Mental retardation, autosomal dominant 9, 614255;Spastic paraplegia 30, autosomal recessive, 610357;Neuropathy, hereditary sensory, type IIC, 614213		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KIF21A	Fibrosis of extraocular muscles, congenital, 1, 135700;Fibrosis of extraocular muscles, congenital, 3B, 135700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KIF5A	Spastic paraplegia 10, autosomal dominant, 604187;Myoclonus, intractable, neonatal, 617235		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KIF7	Acrocallosal syndrome, 200990;?Al-Gazali-Bakalinova syndrome, 607131;Joubert syndrome 12, 200990;?Hydrolethalus syndrome 2, 614120		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KIT	Piebaldism, 172800;Mastocytosis, cutaneous, 154800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KLF1	Blood group--Lutheran inhibitor, 111150;Dyserythropoietic anemia, congenital, type IV, 613673		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KLHL7	Retinitis pigmentosa 42, 612943;Cold-induced sweating syndrome 3, 617055		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KLKB1	Fletcher factor (prekallikrein) deficiency, 612423		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KMT2C	Kleefstra syndrome 2, 617768		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KMT2D	Kabuki syndrome 1, 147920		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KMT2E	O'Donnell-Luria-Rodan syndrome, 618512		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KNG1	Kininogen deficiency, 228960		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KRAS	Leukemia, acute myeloid, 601626;Noonan syndrome 3, 609942;RAS-associated autoimmune leukoproliferative disorder, 614470		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KRIT1	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860;Cerebral cavernous malformations-1, 116860;Cavernous malformations of CNS and retina, 116860		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KRT14	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800;Dermatopathia pigmentosa reticularis, 125595;Epidermolysis bullosa simplex, Koebner type, 131900;Naegeli-Franceschetti-Jadassohn syndrome, 161000;Epidermolysis bullosa simplex, Dowling-Meara type, 131760;Epidermolysis bullosa simplex, recessive 1, 601001		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208

KRT16	Palmoplantar keratoderma, nonepidermolytic, focal, 613000;Pachyonychia congenita 1, 167200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KRT17	Pachyonychia congenita 2, 167210;Steatocystoma multiplex, 184500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KRT5	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800;Epidermolysis bullosa simplex, Koebner type, 131900;Epidermolysis bullosa simplex-MCR, 609352;Dowling-Degos disease 1, 179850;Epidermolysis bullosa simplex, Dowling-Meara type, 131760;Epidermolysis bullosa simplex-MP, 131960;Epidermolysis bullosa simplex, recessive 1, 601001		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
KRT6A	Pachyonychia congenita 3, 615726		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
KYNU	?Hydroxykynureninuria, 236800;Vertebral, cardiac, renal, and limb defects syndrome 2, 617661		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
L1CAM	Hydrocephalus with Hirschsprung disease, 307000;Corpus callosum, partial agenesis of, 304100;MASA syndrome, 303350;Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000;CRASH syndrome, 303350;Hydrocephalus due to aqueductal stenosis, 307000		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
L2HGDH	L-2-hydroxyglutaric aciduria, 236792		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LAMA1	Poretti-Boltshauser syndrome, 615960		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LAMA2	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LAMA3	Epidermolysis bullosa, junctional, Herlitz type, 226700;Epidermolysis bullosa, generalized atrophic benign, 226650;Laryngoonychocutaneous syndrome, 245660		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LAMB1	Lissencephaly 5, 615191		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LAMB2	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199;Pierson syndrome, 609049		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LAMB3	Amelogenesis imperfecta, type IA, 104530;Epidermolysis bullosa, junctional, Herlitz type, 226700;Epidermolysis bullosa, junctional, non-Herlitz type, 226650		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LAMC2	Epidermolysis bullosa, junctional, Herlitz type, 226700;Epidermolysis bullosa, junctional, non-Herlitz type, 226650		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LAMC3	Cortical malformations, occipital, 614115		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LAMP2	Danon disease, 300257		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
LAMTOR2	Immunodeficiency due to defect in MAPBP-interacting protein, 610798		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LARS2	Perrault syndrome 4, OMIM:615300;Hydrops, lactic acidosis, and sideroblastic anemia, OMIM:617021		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LBR	Pelger-Huet anomaly with mild skeletal anomalies, 618019;Pelger-Huet anomaly, 169400;?Reynolds syndrome, 613471;Greenberg skeletal dysplasia, 215140		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LCAT	Fish-eye disease, 136120;Norum disease, 245900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LCT	Lactase deficiency, congenital, 223000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LDB3	Myopathy, myofibrillar, 4, 609452;Left ventricular noncompaction 3, 601493;Cardiomyopathy, dilated, 1C, with or without LVNC, 601493;Cardiomyopathy, hypertrophic, 24, 601493		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
LDHA	Glycogen storage disease XI, 612933		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LDLR	LDL cholesterol level QTL2, 143890;Hypercholesterolemia, familial, 1, 143890		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
LDLRAP1	Hypercholesterolemia, familial, 4, 603813		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LEMD3	Buschke-Ollendorff syndrome, 166700;Osteopoikilosis with or without melorheostosis, 166700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
LEPR	Obesity, morbid, due to leptin receptor deficiency, 614963		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LFNG	Spondylocostal dysostosis 3, autosomal recessive, 609813		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LGI1	Epilepsy, familial temporal lobe, 1, 600512		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
LHFPL5	Deafness, autosomal recessive 67, 610265		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LHX3	Pituitary hormone deficiency, combined, 3, 221750		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LHX4	Pituitary hormone deficiency, combined, 4, 262700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
LIAS	Hyperglycinemia, lactic acidosis, and seizures, 614462		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LIG4	LIG4 syndrome, 606593		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LIPA	Wolman disease, 278000;Cholesteryl ester storage disease, 278000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LITAF	Charcot-Marie-Tooth disease, type 1C, 601098		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
LMAN1	Combined factor V and VIII deficiency, 227300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LMBR1	Syndactyly, type IV, 186200;Hypoplastic or aplastic tibia with polydactyly, 188740;Triphalangeal thumb, type I, 174500;Triphalangeal thumb-polysyndactyly syndrome, 174500;Polydactyly, preaxial type II, 174500;Laurin-Sandrow syndrome, 135750;Acheiropody, 200500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type, 277380		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LMNA	Malouf syndrome, 212112;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Muscular dystrophy, congenital, 613205;Restric		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LMNB1	Leukodystrophy, adult-onset, autosomal dominant, 169500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
LMX1B	Nail-patella syndrome, 161200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
LOXHD1	Deafness, autosomal recessive 77, 613079		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LPIN1	Myoglobinuria, acute recurrent, autosomal recessive, 268200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LPIN2	Majeed syndrome, OMIM:609628		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LPL	Lipoprotein lipase deficiency, 238600;Combined hyperlipidemia, familial, 144250		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LRP2	Donnai-Barrow syndrome, 222448		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LRP4	?Myasthenic syndrome, congenital, 17, 616304;Cenani-Lenz syndactyly syndrome, 212780;Sclerosteosis 2, 614305		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LRP5	van Buchem disease, type 2, 607636;Osteosclerosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;Osteopetrosis, autosomal dominant 1, 607634;Exudative vitreoretinopathy 4, 601813;Polycystic liver disease 4 with or without kidney cysts, 617875;Hyperostosis, endosteal, 144750		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LRPPRC	Leigh syndrome, French-Canadian type, 220111		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LRRC6	Ciliary dyskinesia, primary, 19, 614935		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LRRK2	Parkinson disease 8, Autosomal Dominant, 607060		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
LRSAM1	Charcot-Marie-Tooth disease, axonal, type 2P, 614436		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LRTOMT	Deafness, autosomal recessive 63, 611451		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LTBP2	?Weill-Marchesani syndrome 3, recessive, 614819;Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750;Glaucoma 3, primary congenital, D, 613086		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LTBP3	Geleophysic dysplasia 3, 617809;Dental anomalies and short stature, 601216		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LTBP4	Cutis laxa, autosomal recessive, type IC, 613177		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LYST	Chediak-Higashi syndrome, 214500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LZTFL1	Bardet-Biedl syndrome 17, 615994		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MAF	Ayme-Gripp syndrome, 601088;Cataract 21, multiple types, 610202		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MAFB	Multicentric carpotarsal osteolysis syndrome, 166300;Duane retraction syndrome 3, 617041		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MAGEL2	Schaaf-Yang syndrome, 615547		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MAGI2	Nephrotic syndrome, type 15, 617609		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853;Congenital disorder of glycosylation, type Icc, 301031		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
MAN1B1	Mental retardation, autosomal recessive 15, 614202		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MAN2B1	Mannosidosis, alpha-, types I and II, 248500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MANBA	Mannosidosis, beta, 248510		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MAOA	Brunner syndrome, 300615		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MAP3K1	46XY sex reversal 6, 613762		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MAPT	Pick disease, 172700;Supranuclear palsy, progressive, 601104;Supranuclear palsy, progressive atypical, 260540;Dementia, frontotemporal, with or without parkinsonism, 600274		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MARS2	?Combined oxidative phosphorylation deficiency 25, 616430;Spastic ataxia 3, autosomal recessive, 611390		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MARVELD2	Deafness, autosomal recessive 49, 610153		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MASP1	3MC syndrome 1, 257920		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MASP2	MASP2 deficiency, 613791		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MAT1A	Methionine adenosyltransferase deficiency, autosomal recessive, 250850;Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MATN3	Epiphyseal dysplasia, multiple, 5, 607078;?Spondyloepimetaphyseal dysplasia, 608728		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MATR3	Amyotrophic lateral sclerosis 21, 606070		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MBD5	Mental retardation, autosomal dominant 1, 156200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MBTPS2	Keratosis follicularis spinulosa decalvans, X-linked, 308800;Osteogenesis imperfecta, type XIX, 301014;?Olmsted syndrome, X-linked, 300918;IFAP syndrome with or without BRESHECK syndrome, 308205		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MCEE	Methylmalonyl-CoA epimerase deficiency, 251120		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MCFD2	Factor V and factor VIII, combined deficiency of, 613625		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MCM3AP	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MCM4	Immunodeficiency 54, 609981		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MCOLN1	Mucolipidosis IV, 252650		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MCPH1	Microcephaly 1, primary, autosomal recessive, 251200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MECP2	Rett syndrome, 312750;Mental retardation, X-linked syndromic, Lubs type, 300260;Rett syndrome, atypical, 312750;Rett syndrome, preserved speech variant, 312750;Encephalopathy, neonatal severe, 300673;Mental retardation, X-linked, syndromic 13, 300055		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
MED12	Ohdo syndrome, X-linked, 300895;Opitz-Kaveggia syndrome, 305450;Lujan-Fryns syndrome, 309520		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
MED13L	Mental retardation and distinctive facial features with or without cardiac defects, 616789;Transposition of the great arteries, dextro-looped 1, 608808		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MED23	Mental retardation, autosomal recessive 18, 614249		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MEF2C	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443;Chromosome 5q14.3 deletion syndrome, 613443		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MEFV	Familial Mediterranean fever, AD, OMIM:134610;Familial Mediterranean fever, AR, OMIM:249100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399;Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MEIS2	Cleft palate, cardiac defects, and mental retardation, OMIM:600987;Cardiac malformation, cleft lip/palate, microcephaly, and digital anomalies, MONDO:0010970		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MESP2	Spondylocostal dysostosis 2, autosomal recessive, 608681		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MFF	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MFN2	Hereditary motor and sensory neuropathy VIA, 601152;Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260;Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MFRP	Microphthalmia, isolated 5, 611040;Nanophthalmos 2, 609549		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MFSD2A	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain imaging abnormalities, 616486		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951;Macular dystrophy with central cone involvement, 616170		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MGAT2	Congenital disorder of glycosylation, type IIa, 212066		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MGP	Keutel syndrome, 245150		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MID1	Opitz GBBB syndrome, type I, 300000		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
MITF	Waardenburg syndrome, type 2A, 193510;Tietz albinism-deafness syndrome, 103500;Waardenburg syndrome/ocular albinism, digenic, 103470;COMMAD syndrome, 617306		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MKKS	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MKS1	Bardet-Biedl syndrome 13, 615990;Joubert syndrome 28, 617121;Meckel syndrome 1, 249000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MMADHC	Homocystinuria, cblD type, variant 1, 277410;Methylmalonic aciduria, cblD type, variant 2, 277410;Methylmalonic aciduria and homocystinuria, cblD type, 277410		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MMP13	Metaphyseal dysplasia, Spahr type, 250400;Spondyloepimetaphyseal dysplasia, Missouri type, 602111;Metaphyseal anadysplasia 1, 602111		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MMP2	Multicentric osteolysis, nodulosis, and arthropathy, 259600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MNX1	Currarino syndrome, 176450		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MOCS1	Molybdenum cofactor deficiency A, 252150		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MOCS2	Molybdenum cofactor deficiency B, 252160		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MOGS	Congenital disorder of glycosylation, type IIb, 606056		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MPDU1	Congenital disorder of glycosylation, type If, 609180		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MPI	Congenital disorder of glycosylation, type Ib, OMIM:602579;MPI-CDG, MONDO:0011257		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MPL	Thrombocytopenia, congenital amegakaryocytic, 604498;Thrombocythemia 2, 601977		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MPLKIP	Trichothiodystrophy 4, nonphotosensitive, 234050		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MPO	Myeloperoxidase deficiency, 254600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MPV17	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MPZ	Charcot-Marie-Tooth disease, type 2I, 607677;Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 2J, 607736;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, type 1B, 118200;Hypomyelinating neuropathy, congenital, 2, 618184;Charcot-Marie-Tooth disease, dominant intermediate D, 607791		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MRPL3	Combined oxidative phosphorylation deficiency 9, 614582		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MRPS22	Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MS4A1	Immunodeficiency, common variable, 5, 613495		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MSH6	Mismatch repair cancer syndrome, 276300;Colorectal cancer, hereditary nonpolyposis, type 5, 614350		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis, OMIM:616834;Microcephaly-congenital cataract-psoriasiform dermatitis syndrome, MONDO:0014793		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MSX1	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600;Ectodermal dysplasia 3, Witkop type, 189500;Orofacial cleft 5, 608874		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MSX2	Parietal foramina with cleidocranial dysplasia, 168550;Craniosynostosis 2, 604757;Parietal foramina 1, 168500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MTFMT	Combined oxidative phosphorylation deficiency 15 OMIM:614947;combined oxidative phosphorylation defect type 15 MONDO:0013987;Mitochondrial complex I deficiency, nuclear type 27 OMIM:618248;mitochondrial complex 1 deficiency, nuclear type 27 MONDO:0032631		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MTHFR	Homocystinuria due to MTHFR deficiency, 236250		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MTM1	Myotubular myopathy, X-linked, 310400		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
MTMR2	Charcot-Marie-Tooth disease, type 4B1, 601382		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MTO1	Combined oxidative phosphorylation deficiency 10, 614702		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MTTP	Abetalipoproteinemia, 200100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MUC1	Medullary cystic kidney disease 1, 174000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MUSK	Fetal akinesia deformation sequence 1, 208150;Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MUT	Methylmalonic aciduria, mut(0) type, 251000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MVK	Hyper-IgD syndrome, OMIM:260920;Mevalonic aciduria, OMIM:610377;Porokeratosis 3, multiple types, OMIM:175900		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYBPC1	Arthrogryposis, distal, type 1B, 614335;Lethal congenital contracture syndrome 4, 614915;Myopathy, congenital, with tremor, 618524		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYCN	Feingold syndrome 1, 164280		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MYD88	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYH14	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369;Deafness, autosomal dominant 4A, 600652		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MYH2	Proximal myopathy and ophthalmoplegia, OMIM:605637;Myopathy, proximal, and ophthalmoplegia, MONDO:0011577		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYH3	Contractures, pterygia, and variable skeletal fusions syndrome 1B, 618469;Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436;Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700;Contractures, pterygia, and variable skeletal fusions syndrome 1A, 178110		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYH6	Cardiomyopathy, hypertrophic, 14, 613251;Cardiomyopathy, dilated, 1EE, 613252;Atrial septal defect 3, 614089		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MYH7	Laing distal myopathy, 160500;Myopathy, myosin storage, autosomal dominant, 608358;Myopathy, myosin storage, autosomal recessive, 255160;Cardiomyopathy, hypertrophic, 1, 192600;Left ventricular noncompaction 5, 613426;Scapuloperoneal syndrome, myopathic type, 181430;Cardiomyopathy, dilated, 1S, 613426		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYH8	Carney complex variant, 608837;Trismus-pseudocamptodactyly syndrome, 158300		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYH9	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MYO15A	Deafness, autosomal recessive 3, 600316		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYO18B	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYO1E	Glomerulosclerosis, focal segmental, 6, 614131		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYO3A	Deafness, autosomal recessive 30, 607101		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYO5A	Griscelli syndrome, type 1, 214450		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYO5B	Microvillus inclusion disease, 251850		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYO6	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYO7A	Usher syndrome, type 1B, 276900;Deafness, autosomal dominant 11, 601317;Deafness, autosomal recessive 2, 600060		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MYOT	Myopathy, spheroid body, 182920;Myopathy, myofibrillar, 3, 609200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MYT1	OAVS/Goldenhar syndrome;Oculo-auriculo-vertebral spectrum (OAVS)		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MYT1L	Mental retardation, autosomal dominant 39, 616521		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NAA10	?Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
NAGA	Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NAGS	N-acetylglutamate synthase deficiency, 237310		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208

NARS2	?Deafness, autosomal recessive 94, 618434;Combined oxidative phosphorylation deficiency 24, 616239		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NBAS	Infantile liver failure syndrome 2, 616483;Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NBEA	Seizures;Global developmental delay;Intellectual disability		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NBEAL2	Gray platelet syndrome, 139090		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NBN	Nijmegen breakage syndrome, 251260;Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NCF1	Chronic granulomatous disease due to deficiency of NCF-1, 233700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NCF2	Chronic granulomatous disease due to deficiency of NCF-2, 233710		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDE1	Lissencephaly 4 (with microcephaly), 614019;?Microhydranencephaly, 605013		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDP	Norrie disease, 310600;Exudative vitreoretinopathy 2, X-linked, 305390		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
NDRG1	Charcot-Marie-Tooth disease, type 4D, 601455		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDST1	Mental retardation, autosomal recessive 46, 616116		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFA1	Mitochondrial complex I deficiency, nuclear type 12, 301020		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
NDUFA10	Mitochondrial complex I deficiency, nuclear type 22, 618243		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFA11	Mitochondrial complex I deficiency, nuclear type 14, 618236		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFA4	Isolated complex IV deficiency		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFA6	Mitochondrial complex I deficiency, nuclear type 33, 618253		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFA9	Mitochondrial complex I deficiency, nuclear type 26, 618247		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFAF1	Mitochondrial complex I deficiency, nuclear type 11, 618234		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10, 618233		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFAF3	Mitochondrial complex I deficiency, nuclear type 18, 618240		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFAF4	Mitochondrial complex I deficiency, nuclear type 15, 618237		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16, 618238		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFAF6	Mitochondrial complex I deficiency, nuclear type 17, 618239		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFB3	Mitochondrial complex I deficiency, nuclear type 25, 618246		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFS1	Mitochondrial complex I deficiency, nuclear type 5, 618226		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFS2	Mitochondrial complex I deficiency, nuclear type 6, 618228		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFS3	Mitochondrial complex I deficiency, nuclear type 8, 618230		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1, 252010		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFS6	Mitochondrial complex I deficiency, nuclear type 9, 618232		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFS7	Mitochondrial complex I deficiency, nuclear type 3, 618224		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFS8	Mitochondrial complex I deficiency, nuclear type 2, 618222		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4, 618225		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFV2	Mitochondrial complex I deficiency, nuclear type 7, 618229		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NEB	Nemaline myopathy 2, autosomal recessive, 256030		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NEDD4L	Periventricular nodular heterotopia 7, 617201		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NEFH	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NEFL	Charcot-Marie-Tooth disease, dominant intermediate G, 617882;Charcot-Marie-Tooth disease, type 2E, 607684;Charcot-Marie-Tooth disease, type 1F, 607734		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NEK1	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NEK8	?Nephronophthisis 9, 613824;Renal-hepatic-pancreatic dysplasia 2, 615415		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NEU1	Sialidosis, type I, 256550;Sialidosis, type II, 256550		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NF1	Watson syndrome, 193520;Neurofibromatosis-Noonan syndrome, 601321;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Leukemia, juvenile myelomonocytic, 607785		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NF2	Neurofibromatosis, type 2, 101000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NFE2L2	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NFIA	Brain malformations with or without urinary tract defects, 613735		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NFIX	Sotos syndrome 2, 614753;Marshall-Smith syndrome, 602535		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NFKB1	Immunodeficiency, common variable, 12, 616576		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NFKBIA	Ectodermal dysplasia and immunodeficiency 2, 612132		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NFU1	Multiple mitochondrial dysfunctions syndrome 1, 605711		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora), 254780		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NHP2	Dyskeratosis congenita, autosomal recessive 2, 613987		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NHS	Nance-Horan syndrome, 302350;Cataract 40, X-linked, 302200		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
NIPA1	Spastic paraplegia 6, autosomal dominant, 600363		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NIPAL4	Ichthyosis, congenital, autosomal recessive 6, 612281		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NIPBL	Cornelia de Lange syndrome 1, 122470		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NKX2-1	Chorea, hereditary benign, 118700;Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NKX2-5	Tetralogy of Fallot, 187500;Conotruncal heart malformations, variable, 217095;Ventricular septal defect 3, 614432;Hypothyroidism, congenital nongoitrous, 5, 225250;Hypoplastic left heart syndrome 2, 614435;Atrial septal defect 7, with or without AV conduction defects, 108900		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NLGN3	{Autism susceptibility, X-linked 1}, 300425;{Asperger syndrome susceptibility, X-linked 1}, 300494		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
NLRP12	Familial cold autoinflammatory syndrome 2, OMIM:611762		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NLRP3	CINCA syndrome, OMIM:607115;Deafness, autosomal dominant 34, with or without inflammation, OMIM:617772;Familial cold inflammatory syndrome 1, OMIM:120100;Muckle-Wells syndrome, OMIM:191900;Keratoendothelitis fugax hereditaria, OMIM:148200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NMNAT1	Leber congenital amaurosis 9, 608553		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NOD2	Blau syndrome, OMIM:186580;{Inflammatory bowel disease 1, Crohn disease}, OMIM:266600;{Yao syndrome}, OMIM:617321		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NODAL	Heterotaxy, visceral, 5, 270100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NOG	Tarsal-carpal coalition syndrome, 186570;Symphalangism, proximal, 1A, 185800;Brachydactyly, type B2, 611377;Stapes ankylosis with broad thumbs and toes, 184460;Multiple synostoses syndrome 1, 186500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NOP10	Dyskeratosis congenita, autosomal recessive 1, 224230		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NOP56	Spinocerebellar ataxia 36, 614153		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NOTCH1	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NOTCH2	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NOTCH3	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310;Lateral meningocele syndrome, 130720;?Myofibromatosis, infantile 2, 615293		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NPC1	Niemann-Pick disease, type D, 257220;Niemann-Pick disease, type C1, 257220		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NPC2	Niemann-pick disease, type C2, 607625		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NPHP1	Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900;Joubert syndrome 4, 609583		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NPHP3	Meckel syndrome 7, 267010;Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NPHP4	Nephronophthisis 4, 606966;Senior-Loken syndrome 4, 606996		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NPHS1	Nephrotic syndrome, type 1, 256300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NPHS2	Nephrotic syndrome, type 2, 600995		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NPR2	Short stature with nonspecific skeletal abnormalities, 616255;Epiphyseal chondrodysplasia, Miura type, 615923;Acromesomelic dysplasia, Maroteaux type, 602875		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NR0B1	46XY sex reversal 2, dosage-sensitive, 300018;Adrenal hypoplasia, congenital, 300200		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
NR1H4	Cholestasis, progressive familial intrahepatic, 5, 617049		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NR5A1	Premature ovarian failure 7, 612964;46, XX sex reversal 4, 617480;Spermatogenic failure 8, 613957;Adrenocortical insufficiency, 612964;46XY sex reversal 3, 612965		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NRAS	Noonan syndrome 6, 613224		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NRXN1	Pitt-Hopkins-like syndrome 2, 614325		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NSD1	Sotos syndrome 1, 117550;Leukemia, acute myeloid, 601626		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NSDHL	CK syndrome, 300831;CHILD syndrome, 308050		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
NSUN2	Mental retardation, autosomal recessive 5, 611091		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency, 266120		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800;Medullary thyroid carcinoma, familial, 155240		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NTRK2	Obesity, hyperphagia, and developmental delay, 613886;Epileptic encephalopathy, early infantile, 58, 617830		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NUBPL	Mitochondrial complex I deficiency, nuclear type 21, 618242		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NYX	Night blindness, congenital stationary (complete), 1A, X-linked, 310500		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
OBSL1	3-M syndrome 2, 612921		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
OCA2	Albinism, oculocutaneous, type II, 203200;Albinism, brown oculocutaneous, 203200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
OCLN	Pseudo-TORCH syndrome 1, 251290		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
OCRL	Dent disease 2, 300555;Lowe syndrome, 309000		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ODC1	Global developmental delay;Ectodermal dysplasia;Alopecia;Intellectual disability;Macrocephaly		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
OFD1	Joubert syndrome 10, 300804;?Retinitis pigmentosa 23, 300424;Simpson-Golabi-Behmel syndrome, type 2, 300209;Orofaciodigital syndrome I, 311200		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
OPA1	Behr syndrome, 210000;Optic atrophy 1, 165500;?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896;Optic atrophy plus syndrome, 125250		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
OPA3	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ORAI1	Immunodeficiency 9, 612782;Myopathy, tubular aggregate, 2, 615883		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ORC1	Meier-Gorlin syndrome 1, 224690		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ORC4	Meier-Gorlin syndrome 2, 613800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ORC6	Meier-Gorlin syndrome 3, 613803		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
OSMR	Amyloidosis, primary localized cutaneous, 1, 105250		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
OSTM1	Osteopetrosis, autosomal recessive 5, 259720		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
OTC	Ornithine transcarbamylase deficiency, 311250		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
OTOA	Deafness, autosomal recessive 22, 607039		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
OTOF	Deafness, autosomal recessive 9, 601071;Auditory neuropathy, autosomal recessive, 1, 601071		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
OTX2	Microphthalmia, syndromic 5, 610125;Pituitary hormone deficiency, combined, 6, 613986;Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
P2RY12	Bleeding disorder, platelet-type, 8, 609821		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PAFAH1B1	Lissencephaly 1, 607432;Subcortical laminar heterotopia, 607432		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PAH	Phenylketonuria, 261600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PAK3	Mental retardation, X-linked 30/47, 300558		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
PALB2	Fanconi anemia, complementation group N, 610832		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PANK2	Neurodegeneration with brain iron accumulation 1, 234200;HARP syndrome, 607236		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PAPSS2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PARK7	Parkinson disease 7, autosomal recessive early-onset, 606324		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PAX2	Glomerulosclerosis, focal segmental, 7, 616002;Papillorenal syndrome, 120330		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PAX3	Rhabdomyosarcoma 2, alveolar, 268220;Waardenburg syndrome, type 3, 148820;Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 1, 193500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PAX6	Anterior segment dysgenesis 5, multiple subtypes, 604229;Keratitis, 148190;Cataract with late-onset corneal dystrophy, 106210;?Coloboma, ocular, 120200;Optic nerve hypoplasia, 165550;?Coloboma of optic nerve, 120430;Foveal hypoplasia 1, 136520;Aniridia, 106210;?Morning glory disc anomaly, 120430		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PAX9	Tooth agenesis, selective, 3, 604625		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PC	Pyruvate carboxylase deficiency, 266150		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PCBD1	Hyperphenylalaninemia, BH4-deficient, D, 264070		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PCCA	Propionicacidemia, 606054		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PCCB	Propionicacidemia, 606054		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PCDH15	Deafness, autosomal recessive 23, 609533;Usher syndrome, type 1F, 602083;Usher syndrome, type 1D/F digenic, 601067		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PCSK9	Hypercholesterolemia, familial, 3, 603776		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PDCD10	Cerebral cavernous malformations 3, 603285		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PDE10A	Dyskinesia, limb and orofacial, infantile-onset, 616921;Striatal degeneration, autosomal dominant, 616922		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PDE4D	Acrodysostosis 2, with or without hormone resistance, 614613		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PDE6G	Retinitis pigmentosa 57, 613582		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PDGFB	Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483;Dermatofibrosarcoma protuberans, 607907		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
PDHB	Pyruvate dehydrogenase E1-beta deficiency, 614111		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PDHX	Lacticacidemia due to PDX1 deficiency, 245349		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PDYN	Spinocerebellar ataxia 23, 610245		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PEPD	Prolidase deficiency, 170100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX1	Peroxisome biogenesis disorder 1A (Zellweger), 214100;Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859;Peroxisome biogenesis disorder 3B, 266510		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX13	Peroxisome biogenesis disorder 11B, 614885;Peroxisome biogenesis disorder 11A (Zellweger), 614883		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX14	Peroxisome biogenesis disorder 13A (Zellweger), 614887		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX16	Peroxisome biogenesis disorder 8A (Zellweger), 614876;Peroxisome biogenesis disorder 8B, 614877		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX2	Peroxisome biogenesis disorder 5B, 614867;Peroxisome biogenesis disorder 5A (Zellweger), 614866		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872;Peroxisome biogenesis disorder 7B, 614873		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX3	?Peroxisome biogenesis disorder 10B, 617370;Peroxisome biogenesis disorder 10A (Zellweger), 614882		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716;Peroxisome biogenesis disorder 2B, 202370		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX6	Heimler syndrome 2, 616617;Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX7	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PFKM	Glycogen storage disease VII, 232800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PGAM2	Glycogen storage disease X, 261670		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PGK1	Phosphoglycerate kinase 1 deficiency, 300653		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
PGM1	Congenital disorder of glycosylation, type It, 614921		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PHEX	Hypophosphatemic rickets, X-linked dominant, 307800		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
PHF8	Mental retardation syndrome, X-linked, Siderius type, 300263		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
PHGDH	Phosphoglycerate dehydrogenase deficiency, 601815;Neu-Laxova syndrome 1, 256520		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PHIP	Developmental delay, intellectual disability, obesity, and dysmorphism, 617991		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PHKA1	Muscle glycogenosis, 300559		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
PHKA2	Glycogen storage disease, type IXa1, 306000;Glycogen storage disease, type IXa2, 306000		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
PHKB	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PHKG2	Glycogen storage disease IXc, 613027;Cirrhosis due to liver phosphorylase kinase deficiency		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PHOX2B	Neuroblastoma with Hirschsprung disease, 613013;Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PHYH	Refsum disease, 266500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PIEZO2	?Marden-Walker syndrome, 248700;Arthrogryposis, distal, type 3, 114300;Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208

PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
PIGL	CHIME syndrome, 280000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PIK3CA	PIK3CA-related Overgrowth Spectrum		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PIK3CD	Immunodeficiency 14, 615513		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PIK3R1	Immunodeficiency 36, 616005;SHORT syndrome, 269880;?Agammaglobulinemia 7, autosomal recessive, 615214		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PINK1	Parkinson disease 6, early onset, 605909		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PITX1	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800;Liebenberg syndrome, 186550		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PITX2	Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600;Ring dermoid of cornea, 180550		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PITX3	Cataract 11, multiple types, 610623;Anterior segment dysgenesis 1, multiple subtypes, 107250;Cataract 11, syndromic, autosomal recessive, 610623		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PKD1	Polycystic kidney disease 1, 173900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PKD1L1	Heterotaxy, visceral, 8, autosomal, 617205		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PKD2	Polycystic kidney disease 2, 613095		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PKHD1	Polycystic kidney disease 4, with or without hepatic disease, 263200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PKLR	Adenosine triphosphate, elevated, of erythrocytes, 102900;Pyruvate kinase deficiency, 266200		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PLA2G4A	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600;Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PLAU	Quebec platelet disorder, 601709		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PLCE1	Nephrotic syndrome, type 3, 610725		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PLCG2	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, OMIM:614878;Familial cold autoinflammatory syndrome 3, OMIM:614468		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PLEC	Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723;Epidermolysis bullosa simplex with muscular dystrophy, 226670;?Epidermolysis bullosa simplex with nail dystrophy, 616487;Epidermolysis bullosa simplex, Ogna type, 131950;Epidermolysis bullosa simplex with pyloric atresia, 612138		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PLG	Plasminogen deficiency, type I, 217090;Dysplasminogenemia, 217090		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PLOD2	Bruck syndrome 2, 609220		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PLP1	Pelizaeus-Merzbacher disease, 312080;Spastic paraplegia 2, X-linked, 312920		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
PMM2	Congenital disorder of glycosylation, type Ia, 212065		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PMP22	Charcot-Marie-Tooth disease, type 1E, 118300;?Neuropathy, inflammatory demyelinating, 139393;Dejerine-Sottas disease, 145900;Roussy-Levy syndrome, 180800;Neuropathy, recurrent, with pressure palsies, 162500;Charcot-Marie-Tooth disease, type 1A, 118220		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PMPCA	Spinocerebellar ataxia, autosomal recessive 2, 213200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PMS2	Intellectual disability, developmental delay		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PNKD	Paroxysmal nonkinesigenic dyskinesia 1, 118800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PNKP	Microcephaly, seizures, and developmental delay, 613402;Ataxia-oculomotor apraxia 4, 616267		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PNPLA6	?Laurence-Moon syndrome, 245800;Spastic paraplegia 39, autosomal recessive, 612020;Boucher-Neuhauser syndrome, 215470;Oliver-McFarlane syndrome, 275400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency, 610090		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POGZ	White-Sutton syndrome, 616364		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
POLD1	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
POLG	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal recessive 1, 258450;Progressive external ophthalmoplegia, autosomal dominant 1, 157640		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POLG2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131;Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POLH	Xeroderma pigmentosum, variant type, 278750		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POLR1C	Treacher Collins syndrome 3, 248390;Leukodystrophy, hypomyelinating, 11, 616494		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POLR1D	Treacher Collins syndrome 2, 613717		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694;Wiedemann-Rautenstrauch syndrome, 264090		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157;Retinitis pigmentosa 76, 617123		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POMT1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POMT2	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POP1	Anauxetic dysplasia 2, 617396		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POR	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571;Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PORCN	Focal dermal hypoplasia, 305600		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
POU1F1	Pituitary hormone deficiency, combined, 1, 613038		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
POU3F4	Deafness, X-linked 2, 304400		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
POU4F3	Deafness, autosomal dominant 15, 602459		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PPIB	Osteogenesis imperfecta, type IX, 259440		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PPM1D	Jansen de Vries syndrome, 617450		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PPOX	Porphyria variegata, 176200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PPP2R1A	Mental retardation, autosomal dominant 36, 616362		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PPP2R2B	Spinocerebellar ataxia 12, 604326		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PQBP1	Renpenning syndrome, 309500		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Lymphoma, non-Hodgkin, 605027;Aplastic anemia, 609135		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PRICKLE1	Epilepsy, progressive myoclonic 1B, 612437		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PRKAG2	Glycogen storage disease of heart, lethal congenital, 261740;Cardiomyopathy, hypertrophic 6, 600858;Wolff-Parkinson-White syndrome, 194200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PRKAR1A	Pigmented nodular adrenocortical disease, primary, 1, 610489;Carney complex, type 1, 160980;Acrodysostosis 1, with or without hormone resistance, 101800;Myxoma, intracardiac, 255960		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PRKCG	Spinocerebellar ataxia 14, 605361		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PRKDC	Immunodeficiency 26, with or without neurologic abnormalities, 615966		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PRKRA	Dystonia 16, 612067		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PROC	Thrombophilia due to protein C deficiency, autosomal recessive, 612304;Thrombophilia due to protein C deficiency, autosomal dominant, 176860		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PRODH	Hyperprolinemia, type I, OMIM:239500;hyperprolinemia type 1, MONDO:0009400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PROKR2	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PROP1	Pituitary hormone deficiency, combined, 2, 262600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PROS1	Thrombophilia due to protein S deficiency, autosomal dominant, 612336;Thrombophilia due to protein S deficiency, autosomal recessive, 614514		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PRPS1	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Phosphoribosylpyrophosphate synthetase superactivity, 300661;Gout, PRPS-related, 300661;Arts syndrome, 301835		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
PRRT2	Episodic kinesigenic dyskinesia 1, 128200;Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066;Seizures, benign familial infantile, 2, 605751		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PRSS12	Mental retardation, autosomal recessive 1, 249500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PRSS56	Microphthalmia, isolated 6, 613517		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PRX	Charcot-Marie-Tooth disease, type 4F, 614895;Dejerine-Sottas disease, 145900		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PSAP	Combined SAP deficiency, 611721;Metachromatic leukodystrophy due to SAP-b deficiency, 249900;Gaucher disease, atypical, 610539;Krabbe disease, atypical, 611722		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PSAT1	Neu-Laxova syndrome 2, 616038;?Phosphoserine aminotransferase deficiency, 610992		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PSEN1	?Acne inversa, familial, 3, 613737;Alzheimer disease, type 3, 607822;Pick disease, 172700;Cardiomyopathy, dilated, 1U, 613694;Dementia, frontotemporal, 600274;Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822;Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PSMB8	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, OMIM:256040		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PSPH	Phosphoserine phosphatase deficiency, 614023		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PSTPIP1	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, OMIM:604416		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PTCH1	Basal cell nevus syndrome, 109400;Holoprosencephaly 7, 610828		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PTEN	Macrocephaly/autism syndrome, 605309;Cowden syndrome 1, 158350;Lhermitte-Duclos syndrome, 158350		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PTF1A	Pancreatic agenesis 2, 615935;Pancreatic and cerebellar agenesis, 609069		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PTH1R	Eiken syndrome, 600002;Failure of tooth eruption, primary, 125350;Metaphyseal chondrodysplasia, Murk Jansen type, 156400;Chondrodysplasia, Blomstrand type, 215045		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PTHLH	Brachydactyly, type E2, 613382		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PTPN11	Metachondromatosis, 156250;Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PTPRC	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PYCR1	Cutis laxa, autosomal recessive, type IIB, 612940;Cutis laxa, autosomal recessive, type IIIB, 614438		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PYGL	Glycogen storage disease VI, 232700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PYGM	McArdle disease, 232600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RAB18	Warburg micro syndrome 3, 614222		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RAB23	Carpenter syndrome, 201000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RAB27A	Griscelli syndrome, type 2, 607624		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RAB39B	Mental retardation, X-linked 72, 300271;Waisman syndrome, 311510		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
RAB3GAP1	Warburg micro syndrome 1, 600118		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RAB3GAP2	Warburg micro syndrome 2, 614225;Martsolf syndrome, 212720		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RAB7A	Charcot-Marie-Tooth disease, type 2B, 600882		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RAC1	Mental retardation, autosomal dominant 48, 617751		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RAC2	Neutrophil immunodeficiency syndrome, 608203		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RAD21	?Mungan syndrome, 611376;Cornelia de Lange syndrome 4, 614701		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RAF1	Noonan syndrome 5, 611553;Cardiomyopathy, dilated, 1NN, 615916;LEOPARD syndrome 2, 611554		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RAG1	Omenn syndrome, 603554;Combined cellular and humoral immune defects with granulomas, 233650;Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889;Severe combined immunodeficiency, B cell-negative, 601457		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RAG2	Omenn syndrome, 603554;Combined cellular and humoral immune defects with granulomas, 233650;Severe combined immunodeficiency, B cell-negative, 601457		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RAI1	Smith-Magenis syndrome, 182290		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RAPSN	Fetal akinesia deformation sequence 2, 618388;Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RARS2	Pontocerebellar hypoplasia, type 6, 611523		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RASA1	Capillary malformation-arteriovenous malformation 1, 608354		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RASGRP2	Bleeding disorder, platelet-type, 18, 615888		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RAX	Microphthalmia, isolated 3, 611038		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RBBP8	Pancreatic carcinoma, somatic;Jawad syndrome, 251255;Seckel syndrome 2, 606744		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RBM10	TARP syndrome, 311900		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
RBM8A	Thrombocytopenia-absent radius syndrome, 274000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RECQL4	RAPADILINO syndrome, 266280;Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2,, 268400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
REEP1	?Neuronopathy, distal hereditary motor, type VB, 614751;Spastic paraplegia 31, autosomal dominant, 610250		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RELN	Lissencephaly 2 (Norman-Roberts type), 257320		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
REN	Hyperuricemic nephropathy, familial juvenile 2, 613092;Renal tubular dysgenesis, 267430		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RET	Central hypoventilation syndrome, congenital, 209880		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RFT1	Congenital disorder of glycosylation, type In, 612015		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RFX5	Bare lymphocyte syndrome, type II, complementation group E, 209920;Bare lymphocyte syndrome, type II, complementation group C, 209920		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RFX6	Mitchell-Riley syndrome, 615710		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RFXANK	MHC class II deficiency, complementation group B, 209920		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RFXAP	Bare lymphocyte syndrome, type II, complementation group D, 209920		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RHAG	Overhydrated hereditary stomatocytosis, 185000;Anemia, hemolytic, Rh-null, regulator type, 268150		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RIPK4	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650;CHAND syndrome, 214350		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RMRP	Cartilage-hair hypoplasia, 250250;Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RNF168	RIDDLE syndrome, 611943		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RNF170	Ataxia, sensory, 1, autosomal dominant, 608984		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RNU4ATAC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710;Roifman syndrome, 616651		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ROBO1	tetralogy of Fallot and septal defects		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ROBO3	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ROGDI	Kohlschutter-Tonz syndrome, 226750		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ROR2	Robinow syndrome, autosomal recessive, 268310;Brachydactyly, type B1, 113000		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RORA	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RPE65	Leber congenital amaurosis 2, 204100;Retinitis pigmentosa 20, 613794		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RPGR	Retinitis pigmentosa 3, 300029;Macular degeneration, X-linked atrophic, 300834;Cone-rod dystrophy, X-linked, 1, 304020;Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
RPGRIP1	Leber congenital amaurosis 6, 613826;Cone-rod dystrophy 13, 608194		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RPGRIP1L	Meckel syndrome 5, 611561;COACH syndrome, 216360;Joubert syndrome 7, 611560		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RPIA	Ribose 5-phosphate isomerase deficiency, 608611		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RPL10	Mental retardation, X-linked, syndromic, 35, 300998		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
RPL11	Diamond-Blackfan anemia 7, 612562		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RPL35A	Diamond-Blackfan anemia 5, 612528		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RPL5	Diamond-Blackfan anemia 6, 612561		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RPS10	Diamond-Blackfan anemia 9, 613308		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RPS17	Diamond-Blackfan anemia 4, 612527		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RPS19	Diamond-Blackfan anemia 1, 105650		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RPS24	Diamond-blackfan anemia 3, 610629		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RPS26	Diamond-Blackfan anemia 10, 613309		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RPS6KA3	Coffin-Lowry syndrome, 303600;Mental retardation, X-linked 19, 300844		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
RPS7	Diamond-Blackfan anemia 8, 612563		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075;Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RS1	Retinoschisis, 312700		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
RSPH4A	Ciliary dyskinesia, primary, 11, 612649		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RSPH9	Ciliary dyskinesia, primary, 12, 612650		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RSPO4	Anonychia congenita, 206800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RTN2	Spastic paraplegia 12, autosomal dominant, 604805		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RUNX1	Leukemia, acute myeloid, 601626;Platelet disorder, familial, with associated myeloid malignancy, 601399		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RUNX2	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510;Cleidocranial dysplasia, 119600;Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600;Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RYR1	Central core disease, 117000;King-Denborough syndrome, 145600;Minicore myopathy with external ophthalmoplegia, 255320;Neuromuscular disease, congenital, with uniform type 1 fiber, 117000		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RYR2	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772;Arrhythmogenic right ventricular dysplasia 2, 600996		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SACS	Spastic ataxia, Charlevoix-Saguenay type, 270550		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SALL1	Townes-Brocks branchiootorenal-like syndrome, 107480;Townes-Brocks syndrome 1, 107480		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208

SALL4	IVIC syndrome, 147750;Duane-radial ray syndrome, 607323		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SAMD9	Tumoral calcinosis, familial, normophosphatemic, 610455;MIRAGE syndrome, 617053		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SAMHD1	Aicardi-Goutieres syndrome 5, 612952;?Chilblain lupus 2, 614415		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SAR1B	Chylomicron retention disease, 246700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SATB2	Glass syndrome, 612313		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SBDS	Shwachman-Diamond syndrome, 260400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SBF2	Charcot-Marie-Tooth disease, type 4B2, 604563		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SCARF2	Van den Ende-Gupta syndrome, 600920		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403;Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208;Migraine, familial hemiplegic, 3, 609634;Febrile seizures, familial, 3A, 604403		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SCN1B	Epilepsy, generalized, with febrile seizures plus, type 1, 604233;Cardiac conduction defect, nonspecific, 612838;Epileptic encephalopathy, early infantile, 52, 617350;Atrial fibrillation, familial, 13, 615377;Brugada syndrome 5, 612838		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721;Seizures, benign familial infantile, 3, 607745		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SCN3A	Epilepsy, familial focal, with variable foci 4, 617935;Epileptic encephalopathy, early infantile, 62, 617938		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SCN4A	Hyperkalemic periodic paralysis, type 2, 170500;Myasthenic syndrome, congenital, 16, 614198;Myotonia congenita, atypical, acetazolamide-responsive, 608390;Hypokalemic periodic paralysis, type 2, 613345;Paramyotonia congenita, 168300		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SCN8A	Epileptic encephalopathy, early infantile, 13, 614558;?Myoclonus, familial, 2, 618364;Cognitive impairment with or without cerebellar ataxia, 614306;Seizures, benign familial infantile, 5, 617080		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SCN9A	Insensitivity to pain, congenital, 243000;Febrile seizures, familial, 3B, 613863;Paroxysmal extreme pain disorder, 167400;Epilepsy, generalized, with febrile seizures plus, type 7, 613863;Small fiber neuropathy, 133020;HSAN2D, autosomal recessive, 243000;Erythermalgia, primary, 133020		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SCNN1A	Bronchiectasis with or without elevated sweat chloride 2, 613021;?Liddle syndrome 3, 618126;Pseudohypoaldosteronism, type I, 264350		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SCNN1B	Liddle syndrome 1, 177200;Bronchiectasis with or without elevated sweat chloride 1, 211400;Pseudohypoaldosteronism, type I, 264350		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SCO1	Mitochondrial complex IV deficiency, nuclear type 4, OMIM:619048		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SCO2	Myopia 6, 608908;Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SDCCAG8	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SDHA	Paragangliomas 5, 614165;Leigh syndrome, 256000;Mitochondrial respiratory chain complex II deficiency, 252011;Cardiomyopathy, dilated, 1GG, 613642		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SDHAF1	Mitochondrial complex II deficiency, 252011		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SDHB	Paraganglioma and gastric stromal sarcoma, 606864;Gastrointestinal stromal tumor, 606764;Paragangliomas 4, 115310;Pheochromocytoma, 171300		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SDHD	Mitochondrial complex II deficiency, 252011;Paragangliomas 1, with or without deafness, 168000;Paraganglioma and gastric stromal sarcoma, 606864;Pheochromocytoma, 171300		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SEC23B	?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SEPT9	Leukemia, acute myeloid, therapy-related;Amyotrophy, hereditary neuralgic, 162100;Ovarian carcinoma		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SERPINE1	Plasminogen activator inhibitor-1 deficiency, 613329		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SERPINF1	Osteogenesis imperfecta, type VI, 613982		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SERPINF2	Alpha-2-plasmin inhibitor deficiency, 262850		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SERPING1	Complement component 4, partial deficiency of, 120790;Angioedema, hereditary, types I and II, 106100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SERPINH1	Osteogenesis imperfecta, type X, 613848		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SETBP1	Mental retardation, autosomal dominant 29, 616078;Schinzel-Giedion midface retraction syndrome, 269150		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SETD2	Luscan-Lumish syndrome, 616831		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SETX	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002;Amyotrophic lateral sclerosis 4, juvenile, 602433		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SF3B4	Acrofacial dysostosis 1, Nager type, 154400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SFTPB	Surfactant metabolism dysfunction, pulmonary, 1, 265120		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SGCA	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SGCB	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SGCD	Cardiomyopathy, dilated, 1L, 606685;Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SGCE	Dystonia-11, myoclonic, 159900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SGCG	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SH2D1A	Lymphoproliferative syndrome, X-linked, 1, 308240		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
SH3BP2	Cherubism, OMIM:118400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SH3PXD2B	Frank-ter Haar syndrome, 249420		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SH3TC2	Charcot-Marie-Tooth disease, type 4C, 601596;Mononeuropathy of the median nerve, mild, 613353		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SHANK2	{Autism susceptibility 17}, 613436		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SHANK3	Phelan-McDermid syndrome, 606232		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SHH	Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia with coloboma 5, 611638;Schizencephaly, 269160		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SHOC2	Noonan syndrome-like with loose anagen hair, 607721		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SHOX	Leri-Weill dyschondrosteosis, 127300;Short stature, idiopathic familial, 300582;Langer mesomelic dysplasia, 249700		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SI	Sucrase-isomaltase deficiency, congenital, 222900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SIL1	Marinesco-Sjogren syndrome, 248800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SIX1	Branchiootic syndrome 3, 608389;Deafness, autosomal dominant 23, 605192		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SIX3	Holoprosencephaly 2, 157170;Schizencephaly, 269160		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SIX5	Branchiootorenal syndrome 2, 610896		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SKI	Shprintzen-Goldberg syndrome, 182212		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SKIV2L	Trichohepatoenteric syndrome 2, 614602		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC11A2	Anemia, hypochromic microcytic, with iron overload 1, 206100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC12A1	Bartter syndrome, type 1, OMIM:601678;Bartter disease type 1, MONDO:0100344		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC12A3	Gitelman syndrome, OMIM: 263800;Gitelman syndrome, MONDO:0009904		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, 218000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC16A1	Erythrocyte lactate transporter defect, 245340;Hyperinsulinemic hypoglycemia, familial, 7, 610021;Monocarboxylate transporter 1 deficiency, 616095		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC16A2	Allan-Herndon-Dudley syndrome, 300523		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
SLC17A5	Sialic acid storage disorder, infantile, 269920;Salla disease, 604369		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome, 249270		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC1A2	Epileptic encephalopathy, early infantile, 41, 617105		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SLC1A3	Episodic ataxia, type 6, 612656		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SLC20A2	Basal ganglia calcification, idiopathic, 1, 213600		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SLC22A5	Carnitine deficiency, systemic primary, OMIM:212140;systemic primary carnitine deficiency disease, MONDO:0008919		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC25A12	Epileptic encephalopathy, early infantile, 39, 612949		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC25A13	Citrullinemia, adult-onset type II, 603471;Citrullinemia, type II, neonatal-onset, 605814		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC25A19	Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC25A20	Carnitine-acylcarnitine translocase deficiency, 212138		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC25A3	Mitochondrial phosphate carrier deficiency, 610773		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC25A38	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC25A4	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283;Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC26A2	Diastrophic dysplasia, 222600;Epiphyseal dysplasia, multiple, 4, 226900;Achondrogenesis Ib, 600972;De la Chapelle dysplasia, 256050;Diastrophic dysplasia, broad bone-platyspondylic variant, 222600;Atelosteogenesis, type II, 256050		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC26A3	Diarrhea 1, secretory chloride, congenital, 214700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791;Pendred syndrome, 274600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC27A4	Ichthyosis prematurity syndrome, 608649		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome, 602782		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC2A1	Stomatin-deficient cryohydrocytosis with neurologic defects, 608885;{Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847;Dystonia 9, 601042;GLUT1 deficiency syndrome 2, childhood onset, 612126;GLUT1 deficiency syndrome 1, infantile onset, severe, 606777		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC2A10	Arterial tortuosity syndrome, 208050		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC2A2	Fanconi-Bickel syndrome, 227810		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC30A10	Hypermanganesemia with dystonia 1, 613280		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482;Spastic paraplegia 42, autosomal dominant, 612539		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC34A2	Pulmonary alveolar microlithiasis, 265100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC34A3	Hypophosphatemic rickets with hypercalciuria, 241530		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC35D1	Schneckenbecken dysplasia, 269250		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC37A4	Glycogen storage disease Ib, 232220;Glycogen storage disease Ic, 232240		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC39A13	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC39A4	Acrodermatitis enteropathica, 201100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC3A1	Cystinuria, 220100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC40A1	Hemochromatosis, type 4, 606069		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SLC45A2	Albinism, oculocutaneous, type IV, 606574		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC46A1	Folate malabsorption, hereditary, 229050		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC4A1	Renal tubular acidosis, distal, AR, 611590;Cryohydrocytosis, 185020;Spherocytosis, type 4, 612653;Ovalocytosis, SA type, 166900;Renal tubular acidosis, distal, AD, 179800		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC4A11	Corneal dystrophy, Fuchs endothelial, 4, 613268;Corneal endothelial dystrophy, autosomal recessive, 217700;Corneal endothelial dystrophy and perceptive deafness, 217400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities, 604278		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC52A3	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC5A1	Glucose/galactose malabsorption, 606824		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC5A2	Renal glucosuria, 233100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC5A5	Thyroid dyshormonogenesis 1, 274400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC5A7	Neuronopathy, distal hereditary motor, type VIIA, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC6A1	Myoclonic-atonic epilepsy, 616421		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SLC6A19	Iminoglycinuria, digenic, 242600;Hyperglycinuria, 138500;Hartnup disorder, 234500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC6A3	Parkinsonism-dystonia, infantile, 1, 613135		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC6A5	Hyperekplexia 3, 614618		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
SLC7A7	Lysinuric protein intolerance, 222700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC7A9	Cystinuria, 220100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLC9A6	Mental retardation, X-linked syndromic, Christianson type, 300243		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SLX4	Fanconi anemia, complementation group P, 613951		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SMAD3	Loeys-Dietz syndrome 3, 613795		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050;Polyposis, juvenile intestinal, 174900;Myhre syndrome, 139210		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SMARCA2	Nicolaides-Baraitser syndrome, 601358		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SMARCA4	Coffin-Siris syndrome 4, 614609		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SMARCAL1	Schimke immunoosseous dysplasia, 242900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SMARCB1	Coffin-Siris syndrome 3, 614608;Rhabdoid tumors, somatic, 609322		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SMARCE1	Coffin-Siris syndrome 5, 616938		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SMC1A	Cornelia de Lange syndrome 2, OMIM:300590;Cornelia de Lange syndrome 2, MONDO:0010370;Developmental and epileptic encephalopathy 85, with or without midline brain defects, OMIM:301044;Developmental and epileptic encephalopathy, 85, with or without midline brain defects, MONDO:0026771		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
SMC3	Cornelia de Lange syndrome 3, 610759		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SMN1	Spinal muscular atrophy-3, 253400;Spinal muscular atrophy-4, 271150;Spinal muscular atrophy-2, 253550;Spinal muscular atrophy-1, 253300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SMOC1	Microphthalmia with limb anomalies, 206920		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SMPD1	Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SMPX	Deafness, X-linked 4, 300066		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
SMS	Mental retardation, X-linked, Snyder-Robinson type, 309583		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SNCA	Parkinson disease 1, 168601;Dementia, Lewy body, 127750;Parkinson disease 4, 605543		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SNX10	Osteopetrosis, autosomal recessive 8, 615085		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SOS1	Noonan syndrome 4, 610733		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SOST	Craniodiaphyseal dysplasia, autosomal dominant, 122860;Sclerosteosis 1, 269500;Van Buchem disease, 239100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SOX10	PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584;Waardenburg syndrome, type 4C, 613266		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SOX17	Vesicoureteral reflux 3, 613674		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SOX2	Microphthalmia, syndromic 3, 206900;Optic nerve hypoplasia and abnormalities of the central nervous system, 206900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency, OMIM:300123;Intellectual disability, X-linked, with panhypopituitarism, MONDO:0010252;Panhypopituitarism, X-linked, OMIM:312000;Panhypopituitarism, X-linked, MONDO:0010712		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
SOX5	Lamb-Shaffer syndrome, 616803		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SOX9	Acampomelic campomelic dysplasia, 114290;Campomelic dysplasia, 114290;Campomelic dysplasia with autosomal sex reversal, 114290		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SP110	Hepatic venoocclusive disease with immunodeficiency, 235550		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SPAST	Spastic paraplegia 4, autosomal dominant, OMIM:182601;hereditary spastic paraplegia 4, MONDO:0008438		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SPECC1L	Opitz GBBB syndrome, type II, 145410;?Facial clefting, oblique, 1, 600251;Hypertelorism, Teebi type, 145420		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SPG11	Spastic paraplegia 11, autosomal recessive, 604360;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Amyotrophic lateral sclerosis 5, juvenile, 602099		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SPG21	Mast syndrome, 248900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SPG7	Spastic paraplegia 7, autosomal recessive, OMIM:607259;hereditary spastic paraplegia 7, MONDO:0011803		BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SPINK5	Netherton syndrome, 256500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SPRED1	Legius syndrome, 611431		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SPTA1	Elliptocytosis-2, 130600;Spherocytosis, type 3, 270970;Pyropoikilocytosis, 266140		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SPTAN1	Developmental and epileptic encephalopathy 5, OMIM:613477;developmental and epileptic encephalopathy, 5, MONDO:0013277		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SPTB	Spherocytosis, type 2, 616649;Anemia, neonatal hemolytic, fatal or near-fatal, 617948;Elliptocytosis-3, 617948		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SPTBN2	Spinocerebellar ataxia 5, 600224;Spinocerebellar ataxia, autosomal recessive 14, 615386		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SPTLC1	Neuropathy, hereditary sensory and autonomic, type IA, 162400		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SPTLC2	Neuropathy, hereditary sensory and autonomic, type IC, 613640		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SRC	Colon cancer, advanced, somatic, 114500;?Thrombocytopenia 6, 616937		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SRCAP	Floating-Harbor syndrome, 136140		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SRD5A2	Pseudovaginal perineoscrotal hypospadias, 264600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SRD5A3	Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SRY	46XX sex reversal 1, 400045;46XY sex reversal 1, 400044		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
ST3GAL3	Mental retardation, autosomal recessive 12, 611090;?Epileptic encephalopathy, early infantile, 15, 615006		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ST3GAL5	Salt and pepper developmental regression syndrome, 609056		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
STAR	Lipoid adrenal hyperplasia, 201710		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
STAT1	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796;Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892;Immunodeficiency 31C, autosomal dominant, 614162		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
STAT3	Hyper-IgE recurrent infection syndrome, 147060;Autoimmune disease, multisystem, infantile-onset, 1, 615952		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
STAT5B	Growth hormone insensitivity with immunodeficiency, 245590		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
STIL	Microcephaly 7, primary, autosomal recessive, 612703		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
STIM1	Immunodeficiency 10, 612783;Stormorken syndrome, 185070;Myopathy, tubular aggregate, 1, 160565		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208

STK11	Peutz-Jeghers syndrome, 175200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
STRA6	Microphthalmia, isolated, with coloboma 8, 601186;Microphthalmia, syndromic 9, 601186		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
STRC	Deafness, autosomal recessive 16, 603720		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
STS	Ichthyosis, X-linked, 308100		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
STX11	Hemophagocytic lymphohistiocytosis, familial, 4, 603552		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
STXBP1	Developmental and epileptic encephalopathy 4, OMIM:612164;developmental and epileptic encephalopathy, 4, MONDO:0012812		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5, 613101		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SUMF1	Multiple sulfatase deficiency, 272200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SUOX	Sulfite oxidase deficiency, 272300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684;Leigh syndrome, due to COX IV deficiency, 256000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
SYNE1	Spinocerebellar ataxia, autosomal recessive 8, 610743;Arthrogryposis multiplex congenita, myogenic type, 618484;Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SYNGAP1	Mental retardation, autosomal dominant 5, 612621		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SYP	Mental retardation, X-linked 96, 300802		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
TAB2	Congenital heart defects, nonsyndromic, 2, 614980		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TACO1	Mitochondrial complex IV deficiency, 220110		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TAF1	Mental retardation, X-linked, syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
TALDO1	Transaldolase deficiency, 606003		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TAP1	Bare lymphocyte syndrome, type I, 604571		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TAP2	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TAPBP	Bare lymphocyte syndrome, type I, 604571		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TARDBP	Amyotrophic lateral sclerosis 10, with or without FTD, 612069;Frontotemporal lobar degeneration, TARDBP-related, 612069		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TAT	Tyrosinemia, type II, 276600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TAZ	Barth syndrome, 302060		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
TBC1D23	Pontocerebellar hypoplasia, type 11, 617695		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TBC1D24	DOORS syndrome, 220500;Deafness , autosomal recessive 86, 614617;Deafness, autosomal dominant 65, 616044;Epileptic encephalopathy, early infantile, 16, 615338;Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105;Myoclonic epilepsy, infantile, familial, 605021		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TBCE	Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TBK1	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TBL1XR1	Mental retardation, autosomal dominant 41, 616944;Pierpont syndrome, 602342		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TBP	Spinocerebellar ataxia 17, 607136		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TBX1	DiGeorge syndrome, 188400;Tetralogy of Fallot, 187500;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TBX15	Cousin syndrome, 260660		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TBX20	Atrial septal defect 4, 611363		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TBX22	?Abruzzo-Erickson syndrome, 302905;Cleft palate with ankyloglossia, 303400		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
TBX3	Ulnar-mammary syndrome, 181450		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TBX4	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TBX5	Holt-Oram syndrome, 142900		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TBX6	Spondylocostal dysostosis 5, 122600		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TBXA2R	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TBXAS1	Ghosal hematodiaphyseal syndrome, 231095		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TCAP	Cardiomyopathy, hypertrophic, 25, 607487;Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TCF4	Pitt-Hopkins syndrome, 610954;Corneal dystrophy, Fuchs endothelial, 3, 613267		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TCIRG1	Osteopetrosis, autosomal recessive 1, 259700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TCN2	Transcobalamin II deficiency, 275350		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TCOF1	Treacher Collins syndrome 1, 154500		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TCTN1	Joubert syndrome 13, 614173		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TCTN2	Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TECTA	Deafness, autosomal recessive 21, 603629;Deafness, autosomal dominant 8/12, 601543		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TEK	Glaucoma 3, primary congenital, E, 617272;Venous malformations, multiple cutaneous and mucosal, 600195		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TERC	Dyskeratosis congenita, autosomal dominant 1, 127550		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TERT	{Dyskeratosis congenita, autosomal dominant 2}, 613989;{Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742;{Dyskeratosis congenita, autosomal recessive 4}, 613989		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TF	Atransferrinemia, 209300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TFAP2A	Branchiooculofacial syndrome, 113620		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TFAP2B	Patent ductus arteriosus 2, 617035;Char syndrome, 169100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TFR2	Hemochromatosis, type 3, 604250		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TG	Thyroid dyshormonogenesis 3, 274700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TGFB1	Camurati-Engelmann disease, 131300;Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TGFB2	Loeys-Dietz syndrome 4, 614816		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TGFB3	Loeys-Dietz syndrome 5, 615582;Arrhythmogenic right ventricular dysplasia 1, 107970		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TGFBR1	Loeys-Dietz syndrome 1, 609192		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TGFBR2	Loeys-Dietz syndrome 2, 610168		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TGIF1	Holoprosencephaly 4, 142946		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TGM1	Ichthyosis, congenital, autosomal recessive 1, 242300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TGM5	Peeling skin syndrome 2, 609796		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TGM6	Spinocerebellar ataxia 35, 613908		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TH	Segawa syndrome, recessive, 605407		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
THAP1	Dystonia 6, torsion, 602629		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
THBD	Thrombophilia due to thrombomodulin defect, 614486		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
THPO	Thrombocythemia 1, 187950		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
THRA	Hypothyroidism, congenital, nongoitrous, 6, 614450		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
THRB	Thyroid hormone resistance, selective pituitary, 145650;Thyroid hormone resistance, 188570;Thyroid hormone resistance, autosomal recessive, 274300		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TICAM1	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TIMM8A	Mohr-Tranebjaerg syndrome, 304700		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
TINF2	Revesz syndrome, 268130;Dyskeratosis congenita, autosomal dominant 3, 613990		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TJP2	Hypercholanemia, familial, 607748;Cholestasis, progressive familial intrahepatic 4, 615878		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560;?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TLR3	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMC1	Deafness, autosomal dominant 36, 606705;Deafness, autosomal recessive 7, 600974		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMC6	Epidermodysplasia verruciformis, 226400		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMC8	Epidermodysplasia verruciformis 2, 618231		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMEM138	Joubert syndrome 16, 614465		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMEM165	Congenital disorder of glycosylation, type IIk, 614727		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMEM173	STING-associated vasculopathy, infantile-onset, OMIM:615934		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TMEM216	Meckel syndrome 2, 603194;Joubert syndrome 2, 608091		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMEM237	Joubert syndrome 14, 614424		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMEM43	Arrhythmogenic right ventricular dysplasia 5, 604400;Emery-Dreifuss muscular dystrophy 7, AD, 614302		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TMEM67	Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;Nephronophthisis 11, 613550;?RHYNS syndrome, 602152;COACH syndrome, 216360		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMIE	Deafness, autosomal recessive 6, 600971		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMPRSS3	Deafness, autosomal recessive 8/10, 601072		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TMPRSS6	Iron-refractory iron deficiency anemia, 206200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TNFAIP3	Autoinflammatory syndrome, familial, Behcet-like, OMIM:616744		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TNFRSF11A	Osteolysis, familial expansile, 174810;Osteopetrosis, autosomal recessive 7, 612301		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TNFRSF11B	Paget disease of bone 5, juvenile-onset, 239000		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TNFRSF13B	Immunoglobulin A deficiency 2, 609529;Immunodeficiency, common variable, 2, 240500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TNFRSF13C	Immunodeficiency, common variable, 4, 613494		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TNFRSF1A	Periodic fever, familial, OMIM:142680		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TNFSF11	Osteopetrosis, autosomal recessive 2, 259710		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TNNI2	Arthrogryposis, distal, type 2B1, 601680		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TNNT1	Nemaline myopathy 5, Amish type, 605355		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TNNT3	Arthrogryposis, distal, type 2B2, OMIM:618435;Arthrogryposis, distal, type 2B2, MONDO:0032750		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TOR1A	Arthrogryposis multiplex congenita 5, OMIM:618947;Arthrogryposis multiplex congenita 5, MONDO:0100218;Dystonia-1, torsion, OMIM:128100;Dystonic disorder, MONDO:0003441		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TP53	Bone marrow failure syndrome 5, 618165;Li-Fraumeni syndrome, 151623		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TP53RK	Galloway-Mowat syndrome 4, 617730		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TP63	Orofacial cleft 8, 618149;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292;Hay-Wells syndrome, 106260;Split-hand/foot malformation 4, 605289;ADULT syndrome, 103285;Limb-mammary syndrome, 603543;Rapp-Hodgkin syndrome, 129400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TPM2	CAP myopathy 2, 609285;Arthrogryposis, distal, type 2B4, 108120;Nemaline myopathy 4, autosomal dominant, 609285;Arthrogryposis, distal, type 1A, 108120		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TPM3	Nemaline myopathy 1, autosomal dominant or recessive, 609284;CAP myopathy 1, 609284;Myopathy, congenital, with fiber-type disproportion, 255310		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TPO	Thyroid dyshormonogenesis 2A, 274500		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TPP2	TPP2 deficiency		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRAC	Immunodeficiency 7, TCR-alpha/beta deficient, 615387		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRAF3IP1	Senior-Loken syndrome 9, 616629		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRAPPC2	Spondyloepiphyseal dysplasia tarda, 313400		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
TRAPPC9	Mental retardation, autosomal recessive 13, 613192		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TREM2	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;Chilblain lupus, 610448;Vasculopathy, retinal, with cerebral leukodystrophy, 192315		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRIM32	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRIM37	Mulibrey nanism, 253250		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRIO	Mental retardation, autosomal dominant 44, 617061		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TRIOBP	Deafness, autosomal recessive 28, 609823		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRIP11	Osteochondrodysplasia, 184260;Achondrogenesis, type IA, 200600		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRIP12	Mental retardation, autosomal dominant 49, 617752		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TRMT1	Mental retardation, autosomal recessive 68, 618302		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRMU	Liver failure, transient infantile, 613070		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRPC6	Glomerulosclerosis, focal segmental, 2, 603965		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TRPM1	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRPM4	Erythrokeratodermia veriabilis et progressiva 6, 618531;Progressive familial heart block, type IB, 604559		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TRPM6	Hypomagnesemia 1, intestinal, 602014		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRPS1	Trichorhinophalangeal syndrome, type I, 190350;Trichorhinophalangeal syndrome, type III, 190351		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TRPV4	Digital arthropathy-brachydactyly, familial, 606835;Metatropic dysplasia, 156530;Spinal muscular atrophy, distal, congenital nonprogressive, 600175;Brachyolmia type 3, 113500;Hereditary motor and sensory neuropathy, type IIc, 606071;Scapuloperoneal spinal muscular atrophy, 181405;[Sodium serum level QTL 1], 613508;Spondylometaphyseal dysplasia, Kozlowski type, 1842		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRRAP	Developmental delay with or without dysmorphic facies and autism, 618454		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TSC1	Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TSC2	Tuberous sclerosis-2, 613254		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TSEN2	Pontocerebellar hypoplasia type 2B, 612389		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TSEN54	Pontocerebellar hypoplasia type 4, 225753;Pontocerebellar hypoplasia type 2A, 277470;?Pontocerebellar hypoplasia type 5, 610204		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TSFM	Combined oxidative phosphorylation deficiency 3, 610505		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TSHB	Hypothyroidism, congenital, nongoitrous 4, 275100		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TSHR	Hyperthyroidism, nonautoimmune, 609152;Hyperthyroidism, familial gestational, 603373;Hypothyroidism, congenital, nongoitrous, 1, 275200		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TSPAN7	Mental retardation, X-linked 58, 300210		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
TTBK2	Spinocerebellar ataxia 11, 604432		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TTC21B	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819;Nephronophthisis 12, 613820		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TTC37	Trichohepatoenteric syndrome 1, 222470		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TTC8	?Retinitis pigmentosa 51, 613464;Bardet-Biedl syndrome 8, 615985		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TTI2	Mental retardation, autosomal recessive 39, 615541		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TTN	Tibial muscular dystrophy, tardive, 600334;Salih myopathy, 611705;Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807;Myopathy, myofibrillar, 9, with early respiratory failure, 603689;Cardiomyopathy, familial hypertrophic, 9, 613765;Cardiomyopathy, dilated, 1G, 604145		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TTPA	Ataxia with isolated vitamin E deficiency, 277460		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TTR	Carpal tunnel syndrome, familial, 115430;Amyloidosis, hereditary, transthyretin-related, 105210		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TUBA1A	Lissencephaly 3, 611603		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TUBA8	Cortical dysplasia, complex, with other brain malformations 8, 613180		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TUBB1	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TUBB2B	Cortical dysplasia, complex, with other brain malformations 7, 610031		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TUBB3	Fibrosis of extraocular muscles, congenital, 3A, 600638;Cortical dysplasia, complex, with other brain malformations 1, 614039		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TUFM	Combined oxidative phosphorylation deficiency 4, 610678		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TUSC3	Mental retardation, autosomal recessive 7, 611093		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TWIST1	Saethre-Chotzen syndrome with or without eyelid anomalies, 101400;Sweeney-Cox syndrome, 617746;Robinow-Sorauf syndrome, 180750;Craniosynostosis 1, 123100		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TWIST2	Ablepharon-macrostomia syndrome, 200110;Focal facial dermal dysplasia 3, Setleis type, 227260;Barber-Say syndrome, 209885		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TYK2	Immunodeficiency 35, 611521		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TYR	Albinism, oculocutaneous, type IA, 203100;Albinism, oculocutaneous, type IB, 606952;Waardenburg syndrome/albinism, digenic, 103470		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TYROBP	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TYRP1	Albinism, oculocutaneous, type III, 203290		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UBA1	Spinal muscular atrophy, X-linked 2, infantile, 301830		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
UBE2A	Mental retardation, X-linked syndromic, Nascimento-type, 300860		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
UBE3A	Angelman syndrome, 105830		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
UBR1	Johanson-Blizzard syndrome, 243800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UCHL1	Spastic paraplegia 79B, autosomal recessive, OMIM:615491;early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome, MONDO:0014209;Spastic paraplegia 79A, autosomal dominant, OMIM:620221		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208

UGT1A1	Crigler-Najjar syndrome, type I, 218800;Crigler-Najjar syndrome, type II, 606785;Hyperbilirubinemia, familial transient neonatal, 237900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UMOD	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886;Medullary cystic kidney disease 2, 603860;Hyperuricemic nephropathy, familial juvenile 1, 162000		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
UMPS	Orotic aciduria, 258900		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3, 608898		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UNC93B1	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UNG	Immunodeficiency with hyper IgM, type 5, 608106		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UPF3B	Mental retardation, X-linked, syndromic 14, 300676		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
UQCRB	Mitochondrial complex III deficiency, nuclear type 3, 615158		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UROD	Porphyria cutanea tarda, 176100;Porphyria, hepatoerythropoietic, 176100		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UROS	Porphyria, congenital erythropoietic, 263700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
USB1	Poikiloderma with neutropenia, 604173		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
USH1C	Deafness, autosomal recessive 18A, 602092;Usher syndrome, type 1C, 276904		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
USH1G	Usher syndrome, type 1G, 606943		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
USH2A	Usher syndrome, type 2A, 276901;Retinitis pigmentosa 39, 613809		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
USP9X	Mental retardation, X-linked 99, syndromic, female-restricted, 300968;Mental retardation, X-linked 99, 300919		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
UVSSA	UV-sensitive syndrome 3, 614640		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VCAN	Wagner syndrome 1, 143200		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
VCP	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954;Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320;Charcot-Marie-Tooth disease, type 2Y, 616687		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VDR	Rickets, vitamin D-resistant, type IIA, 277440		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VHL	Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Pheochromocytoma, 171300		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473;Warfarin resistance, 122700		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VMA21	Myopathy, X-linked, with excessive autophagy, 310440		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
VPS13A	Choreoacanthocytosis, 200150		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VPS13B	Cohen syndrome, 216550		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VRK1	Pontocerebellar hypoplasia type 1A, 607596		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VSX2	Microphthalmia with coloboma 3, 610092;Microphthalmia, isolated 2, 610093		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VWF	von Willibrand disease, type 3, 277480;von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554;von Willebrand disease, type 1, 193400		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WAS	Thrombocytopenia, X-linked, intermittent, 313900;Neutropenia, severe congenital, X-linked, 300299;Wiskott-Aldrich syndrome, 301000;Thrombocytopenia, X-linked, 313900		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
WDR11	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
WDR19	Senior-Loken syndrome 8, 616307;?Cranioectodermal dysplasia 4, 614378;Nephronophthisis 13, 614377;?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WDR35	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WDR4	Microcephaly, growth deficiency, seizures, and brain malformations, 618346;Galloway-Mowat syndrome 6, 618347		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WDR45	Neurodegeneration with brain iron accumulation 5, 300894		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
WDR45B	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WDR81	Hydrocephalus, congenital, 3, with brain anomalies, 617967;Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WFS1	Wolfram-like syndrome, autosomal dominant, 614296;?Cataract 41, 116400;Wolfram syndrome 1, 222300;Deafness, autosomal dominant 6/14/38, 600965		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood, 208230;Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WNT10A	Tooth agenesis, selective, 4, 150400;Schopf-Schulz-Passarge syndrome, 224750;Odontoonychodermal dysplasia, 257980		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WNT10B	Split-hand/foot malformation 6, 225300;Tooth agenesis, selective, 8, 617073		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WNT5A	Robinow syndrome, autosomal dominant 1, 180700		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
WNT7A	Ulna and fibula, absence of, with severe limb deficiency, 276820;Fuhrmann syndrome, 228930		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WRAP53	Dyskeratosis congenita, autosomal recessive 3, 613988		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WRN	Werner syndrome, 277700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WT1	Wilms tumor, type 1, 194070;Denys-Drash syndrome, 194080;Frasier syndrome, 136680;Meacham syndrome, 608978;Nephrotic syndrome, type 4, 256370		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
WWOX	Epileptic encephalopathy, early infantile, 28, 616211;Spinocerebellar ataxia, autosomal recessive 12, 614322		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
XDH	Xanthinuria, type I, 278300		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
XIAP	Lymphoproliferative syndrome, X-linked, 2, 300635		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
XPA	Xeroderma pigmentosum, group A, 278700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
XPC	Xeroderma pigmentosum, group C, 278720		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
XRCC4	Short stature, microcephaly, and endocrine dysfunction, 616541		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
XYLT1	Desbuquois dysplasia 2, OMIM:615777;Desbuquois dysplasia 2, MONDO:0014343		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
XYLT2	Spondyloocular syndrome, 605822		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
YY1	Gabriele-de Vries syndrome, 617557		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ZAP70	Autoimmune disease, multisystem, infantile-onset, 2, 617006;Immunodeficiency 48, 269840		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ZBTB18	Mental retardation, autosomal dominant 22, 612337		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type, 300799		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ZEB2	Mowat-Wilson syndrome, 235730		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ZFP57	Diabetes mellitus, transient neonatal, 1, 601410		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ZFYVE26	Spastic paraplegia 15, autosomal recessive, 270700		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ZIC1	Craniosynostosis 6, 616602		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ZIC2	Holoprosencephaly 5, 609637		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ZIC3	VACTERL association, X-linked, 314390;Heterotaxy, visceral, 1, X-linked, 306955;Congenital heart defects, nonsyndromic, 1, X-linked, 306955		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy, lethal, 275210		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ZMYND11	Mental retardation, autosomal dominant 30, 616083		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ZNF469	Brittle cornea syndrome 1, 229200		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ZNF711	Mental retardation, X-linked 97, 300803		X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Research panel - Severe Paediatric Disorders			1.208
ADAM17	?Inflammatory skin and bowel disease, neonatal, 1, 614328		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
APOL1	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551;{End-stage renal disease, nondiabetic, susceptibility to}, 612551		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
ATP8A2	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATPAF2	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BCL10	?Immunodeficiency 37, 616098		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
BLNK	?Agammaglobulinemia 4, 613502		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CASP8	?Autoimmune lymphoproliferative syndrome, type IIB, 607271		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CDC6	?Meier-Gorlin syndrome 5, 613805		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CFB	?Complement factor B deficiency, 615561		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
COX14	?Mitochondrial complex IV deficiency, 220110		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DDOST	?Congenital disorder of glycosylation, type Ir, 614507		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DPP6	Mental retardation, autosomal dominant 33, 616311		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FASTKD2	?Mitochondrial complex IV deficiency, 220110		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GAD1	?Cerebral palsy, spastic quadriplegic, 1, 603513		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GFI1	?Neutropenia, severe congenital 2, autosomal dominant, 613107;?Neutropenia, nonimmune chronic idiopathic, of adults, 607847		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HARS2	?Perrault syndrome 2, 614926		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HTT	Lopes-Maciel-Rodan syndrome, OMIM:617435;Lopes-Maciel-Rodan syndrome, MONDO:0054573		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
HYAL1	?Mucopolysaccharidosis type IX, 601492		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL17F	?Candidiasis, familial, 6, autosomal dominant, 613956		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
IL21	?Immunodeficiency, common variable, 11, 615767		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IRF7	?Immunodeficiency 39, 616345		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
LIG1	DNA ligase I deficiency		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MTPAP	?Spastic ataxia 4, autosomal recessive, 613672		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NCF4	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
NDUFA2	?Mitochondrial complex I deficiency, nuclear type 13, 618235		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PCK1	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PEX11B	?Peroxisome biogenesis disorder 14B, 614920		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
PTCHD1	?Bleeding disorder, platelet-type, 19, 616176		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
RB1	{Autism, susceptibility to, X-linked 4}, 300830		X-LINKED: hemizygous mutation in males, biallelic mutations in females	Research panel - Severe Paediatric Disorders			1.208
SCP2	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SEMA3E	?Leukoencephalopathy with dystonia and motor neuropathy, 613724		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
SIGMAR1	?CHARGE syndrome, 214800		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SQSTM1	Paget disease of bone 3, 167250;Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145;Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437;Myopathy, distal, with rimmed vacuoles, 617158		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRAF3			MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
TSEN34	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UNC119	?Pontocerebellar hypoplasia type 2C, 612390		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
UROC1	?Immunodeficiency 13, 615518;?Cone-rod dystrophy		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WDPCP	?Urocanase deficiency, 276880		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WIPF1	?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085;?Bardet-Biedl syndrome 15, 615992		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
WNT3	?Wiskott-Aldrich syndrome 2, 614493		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
ATG16L1			MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
CD46	{Inflammatory bowel disease (Crohn disease) 10}, 611081		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CFHR1	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CFHR2	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400;{Macular degeneration, age-related, reduced risk of}, 603075		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CFHR3			BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CFHR4	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400;{Macular degeneration, age-related, reduced risk of}, 603075		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
CPA6	Epilepsy, familial temporal lobe, 5, 614417;Febrile seizures, familial, 11, 614418		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DCXR			BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
DRD2	[Pentosuria], 260800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
FPR1			BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
GNAO	Periodontitis, susceptibility to		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
HMGA2	Silver-Russell syndrome 5, OMIM:618908;Silver-Russell syndrome 5, MONDO:0020795		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
IDH1	Leiomyoma, uterine, somatic, 150699		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
IL10	{Glioma, susceptibility to, somatic}, 137800		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
IL17A	{HIV-1, susceptibility to}, 609423;{Graft-versus-host disease, protection against}, 614395;{Rheumatoid arthritis, progression of}, 180300		Unknown	Research panel - Severe Paediatric Disorders			1.208
ITGAM	AutoAb Chronic Mucocutaneous Candidiasis.		Unknown	Research panel - Severe Paediatric Disorders			1.208
KIRREL3	Systemic lupus erythematous, suscpetibility to		Unknown	Research panel - Severe Paediatric Disorders			1.208
MBL2	Mental retardation		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
MEN1	{Chronic infections, due to MBL deficiency}, 614372		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
MUTYH	Angiofibroma, somatic;Carcinoid tumor of lung;Multiple endocrine neoplasia 1, 131100;Lipoma, somatic;Adrenal adenoma, somatic;Parathyroid adenoma, somatic		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
NRXN3	Spinocerebellar ataxia 4, kidney papillary necrosis, IBD, recurrent sinopulmolnary infections, NFAT5 haploinsufficieny		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PRICKLE2	Autism		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
PRNP	Creutzfeldt-Jakob disease, 123400;Huntington disease-like 1, 603218;Insomnia, fatal familial, 600072;Prion disease with protracted course, 606688;Cerebral amyloid angiopathy, PRNP-related, 137440;Gerstmann-Straussler disease, 137440		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RNF135	Epilepsy, progressive myoclonic 5, 613832		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
RPS15	Polyglucosan body myopathy, early-onset, with or without immunodeficiency		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SART3			MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SIM1	Porokeratosis		MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown	Research panel - Severe Paediatric Disorders			1.208
SLC6A20	Iminoglycinuria, digenic, 242600;Hyperglycinuria, 138500		BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TNFRSF4	Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
TRAF3IP2	Immunodeficiency, common variable with lack of anti-pneumococcal antibody		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208
VPS35	?Candidiasis, familial, 8, 615527		BIALLELIC, autosomal or pseudoautosomal	Research panel - Severe Paediatric Disorders			1.208