PRE CONCEPTION SCREENING GENE PANEL DG 3.5.0 (2337 genes)

Releasedate: 05-12-2022

Gene	TWIST X2 covered >10x	TWIST X2 covered >20x	Associated Phenotype description and OMIM disease ID
AAAS	100%	100%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS1	100%	100%	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287 ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 Trichothiodystrophy 8, nonphotosensitive, 619691
AARS2	100%	100%	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
AASS	100%	100%	Hyperlysinemia, 238700
ABAT	100%	100%	GABA-transaminase deficiency, 613163
ABCA1	100%	100%	Tangier disease, 205400 HDL deficiency, familial, 1, 604091
ABCA3	100%	100%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	100%	100%	Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Stargardt disease 1, 248200
ABCB11	100%	100%	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	100%	100%	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347
ABCC2	100%	100%	Dubin-Johnson syndrome, 237500
ABCC6	100%	100%	Pseudoxanthoma elasticum, 264800 Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	100%	100%	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Hypoglycemia of infancy, leucine-sensitive, 240800 Hyperinsulinemic hypoglycemia, familial, 1, 256450
ABCD4	100%	100%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	100%	100%	Sitosterolemia 2, 618666
ABCG8	100%	100%	Sitosterolemia 1, 210250

ABHD12	*100%*	*100%*	*Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674*
ABHD16A	*100%*	*100%*	*Spastic paraplegia 86, autosomal recessive, 619735*
ABHD5	*100%*	*100%*	*Chanarin-Dorfman syndrome, 275630*
ACACA	*100%*	*100%*	*No OMIM disease ID*
ACAD8	*100%*	*100%*	*Isobutyryl-CoA dehydrogenase deficiency, 611283*
ACAD9	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 20, 611126*
ACADM	*100%*	*100%*	*Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450*
ACADS	*100%*	*100%*	*Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470*
ACADSB	*100%*	*100%*	*2-methylbutyrylglycinuria, 610006*
ACADVL	*100%*	*100%*	*VLCAD deficiency, 201475*
ACAN	*99%*	*99%*	*?Spondyloepiphyseal dysplasia, Kimberley type, 608361* *Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800* *Spondyloepimetaphyseal dysplasia, aggrecan type, 612813*
ACAT1	*100%*	*100%*	*Alpha-methylacetoacetic aciduria, 203750*
ACE	*100%*	*100%*	*Renal tubular dysgenesis, 267430*
ACER3	*100%*	*100%*	*?Leukodystrophy, progressive, early childhood-onset, 617762*
ACO2	*100%*	*100%*	*Optic atrophy 9, 616289* *Infantile cerebellar-retinal degeneration, 614559*
ACOX1	*100%*	*100%*	*Mitchell syndrome, 618960* *Peroxisomal acyl-CoA oxidase deficiency, 264470*
ACOX2	*100%*	*100%*	*Bile acid synthesis defect, congenital, 6, 617308*
ACP5	*100%*	*100%*	*Spondyloenchondrodysplasia with immune dysregulation, 607944*
ACSF3	*100%*	*100%*	*Combined malonic and methylmalonic aciduria, 614265*
ACTA1	*100%*	*100%*	*?Myopathy, scapulohumeroperoneal, 616852* *Nemaline myopathy 3, autosomal dominant or recessive, 161800* *Myopathy, actin, congenital, with excess of thin myofilaments, 161800* *Myopathy, actin, congenital, with cores, 161800* *Myopathy, congenital, with fiber-type disproportion 1, 255310*
ACTL6B	*100%*	*100%*	*Developmental and epileptic encephalopathy 76, 618468* *Intellectual developmental disorder with severe speech and ambulation defects, 618470*
ACY1	*100%*	*100%*	*Aminoacylase 1 deficiency, 609924*
ADA	*100%*	*100%*	*Adenosine deaminase deficiency, partial, 102700* *Severe combined immunodeficiency due to ADA deficiency, 102700*

ADA2	*100%*	*100%*	*Sneddon syndrome, 182410* *Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688*
ADAM17	*100%*	*100%*	*?Inflammatory skin and bowel disease, neonatal, 1, 614328*
ADAM22	*100%*	*100%*	*Developmental and epileptic encephalopathy 61, 617933*
ADAM9	*100%*	*100%*	*Cone-rod dystrophy 9, 612775*
ADAMTS10	*100%*	*100%*	*Weill-Marchesani syndrome 1, recessive, 277600*
ADAMTS13	*100%*	*100%*	*Thrombotic thrombocytopenic purpura, hereditary, 274150*
ADAMTS17	*100%*	*100%*	*Weill-Marchesani 4 syndrome, recessive, 613195*
ADAMTS18	*100%*	*100%*	*Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458*
ADAMTS2	*98%*	*98%*	*Ehlers-Danlos syndrome, dermatosparaxis type, 225410*
ADAMTS3	*99%*	*99%*	*Hennekam lymphangiectasia-lymphedema syndrome 3, 618154*
ADAMTSL2	*100%*	*100%*	*Geleophysic dysplasia 1, 231050*
ADAMTSL4	*100%*	*100%*	*Ectopia lentis et pupillae, 225200* *Ectopia lentis, isolated, autosomal recessive, 225100*
ADAR	*100%*	*100%*	*Dyschromatosis symmetrica hereditaria, 127400* *Aicardi-Goutieres syndrome 6, 615010*
ADARB1	*95%*	*95%*	*Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862*
ADAT3	*100%*	*100%*	*Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286*
ADCY6	*100%*	*100%*	*Lethal congenital contracture syndrome 8, 616287*
ADD3	*100%*	*100%*	*Cerebral palsy, spastic quadriplegic, 3, 617008*
ADGRG1	*100%*	*100%*	*Polymicrogyria, bilateral frontoparietal, 606854* *Polymicrogyria, bilateral perisylvian, 615752*
ADGRG6	*100%*	*100%*	*Lethal congenital contracture syndrome 9, 616503*
ADGRV1	*100%*	*100%*	*Usher syndrome, type 2C, 605472* *Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472* *?Febrile seizures, familial, 4, 604352*
ADK	*91%*	*91%*	*Hypermethioninemia due to adenosine kinase deficiency, 614300*
ADPRS	*100%*	*100%*	*Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170*
ADSL	*100%*	*100%*	*Adenylosuccinase deficiency, 103050*
ADSS1	*100%*	*100%*	*Myopathy, distal, 5, 617030*
AEBP1	*100%*	*100%*	*Ehlers-Danlos syndrome, classic-like, 2, 618000*
AFG3L2	*100%*	*100%*	*Spastic ataxia 5, autosomal recessive, 614487* *Optic atrophy 12, 618977* *Spinocerebellar ataxia 28, 610246*

AGA	*100%*	*100%*	*Aspartylglucosaminuria, 208400*
AGBL5	*100%*	*100%*	*Retinitis pigmentosa 75, 617023*
AGK	*92%*	*92%*	*Cataract 38, autosomal recessive, 614691* *Sengers syndrome, 212350*
AGL	*100%*	*100%*	*Glycogen storage disease IIIa, 232400* *Glycogen storage disease IIIb, 232400*
AGPAT2	*100%*	*100%*	*Lipodystrophy, congenital generalized, type 1, 608594*
AGPS	*100%*	*100%*	*Rhizomelic chondrodysplasia punctata, type 3, 600121*
AGRN	*100%*	*100%*	*Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120*
AGT	*100%*	*100%*	*Renal tubular dysgenesis, 267430*
AGTPBP1	*100%*	*100%*	*Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276*
AGTR1	*100%*	*100%*	*Renal tubular dysgenesis, 267430*
AGXT	*100%*	*100%*	*Hyperoxaluria, primary, type 1, 259900*
AHCY	*100%*	*100%*	*Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752*
AHI1	*100%*	*100%*	*Joubert syndrome 3, 608629*
AHR	*100%*	*100%*	*?Retinitis pigmentosa 85, 618345*
AICDA	*100%*	*100%*	*Immunodeficiency with hyper-IgM, type 2, 605258*
AIMP1	*100%*	*100%*	*Leukodystrophy, hypomyelinating, 3, 260600*
AIMP2	*100%*	*100%*	*Leukodystrophy, hypomyelinating, 17, 618006*
AIPL1	*100%*	*100%*	*Leber congenital amaurosis 4, 604393* *Retinitis pigmentosa, juvenile, 604393* *Cone-rod dystrophy, 604393*
AIRE	*100%*	*100%*	*Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300*
AK1	*100%*	*100%*	*Hemolytic anemia due to adenylate kinase deficiency, 612631*
AK2	*100%*	*100%*	*Reticular dysgenesis, 267500*
AKR1D1	*100%*	*100%*	*Bile acid synthesis defect, congenital, 2, 235555*
ALAD	*100%*	*100%*	*Porphyria, acute hepatic, 612740*
ALB	*100%*	*100%*	*Analbuminemia, 616000*
ALDH18A1	*100%*	*100%*	*Spastic paraplegia 9A, autosomal dominant, 601162* *Cutis laxa, autosomal recessive, type IIIA, 219150* *Spastic paraplegia 9B, autosomal recessive, 616586* *Cutis laxa, autosomal dominant 3, 616603*
ALDH1A3	*100%*	*100%*	*Microphthalmia, isolated 8, 615113*
ALDH3A2	*94%*	*94%*	*Sjogren-Larsson syndrome, 270200*

ALDH4A1	*100%*	*100%*	*Hyperprolinemia, type II, 239510*
ALDH5A1	*100%*	*100%*	*Succinic semialdehyde dehydrogenase deficiency, 271980*
ALDH6A1	*100%*	*100%*	*Methylmalonate semialdehyde dehydrogenase deficiency, 614105*
ALDH7A1	*100%*	*100%*	*Epilepsy, pyridoxine-dependent, 266100*
ALDOA	*100%*	*100%*	*Glycogen storage disease XII, 611881*
ALDOB	*100%*	*100%*	*Fructose intolerance, hereditary, 229600*
ALG1	*100%*	*100%*	*Congenital disorder of glycosylation, type Ik, 608540*
ALG11	*96%*	*96%*	*Congenital disorder of glycosylation, type Ip, 613661*
ALG12	*100%*	*100%*	*Congenital disorder of glycosylation, type Ig, 607143*
ALG14	*100%*	*100%*	*Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031* *Myopathy, epilepsy, and progressive cerebral atrophy, 619036* *?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227*
ALG2	*100%*	*100%*	*Congenital disorder of glycosylation, type Ii, 607906* *Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228*
ALG3	*100%*	*100%*	*Congenital disorder of glycosylation, type Id, 601110*
ALG6	*100%*	*100%*	*Congenital disorder of glycosylation, type Ic, 603147*
ALG8	*96%*	*96%*	*Congenital disorder of glycosylation, type Ih, 608104* *Polycystic liver disease 3 with or without kidney cysts, 617874*
ALG9	*100%*	*100%*	*Gillessen-Kaesbach-Nishimura syndrome, 263210* *Congenital disorder of glycosylation, type Il, 608776*
ALKBH8	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 71, 618504*
ALMS1	*100%*	*100%*	*Alstrom syndrome, 203800*
ALOX12B	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 2, 242100*
ALOXE3	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 3, 606545*
ALPK3	*100%*	*100%*	*Cardiomyopathy, familial hypertrophic 27, 618052*
ALPL	*100%*	*100%*	*Odontohypophosphatasia, 146300* *Hypophosphatasia, infantile, 241500* *Hypophosphatasia, childhood, 241510* *Hypophosphatasia, adult, 146300*
ALS2	*100%*	*100%*	*Primary lateral sclerosis, juvenile, 606353* *Spastic paralysis, infantile onset ascending, 607225* *Amyotrophic lateral sclerosis 2, juvenile, 205100*
ALX1	*100%*	*100%*	*Frontonasal dysplasia 3, 613456*
ALX3	*100%*	*100%*	*Frontonasal dysplasia 1, 136760*

ALX4	*100%*	*100%*	*Parietal foramina 2, 609597* *Frontonasal dysplasia 2, 613451*
AMACR	*100%*	*100%*	*Alpha-methylacyl-CoA racemase deficiency, 614307* *Bile acid synthesis defect, congenital, 4, 214950*
AMBN	*100%*	*100%*	*Amelogenesis imperfecta, type IF, 616270*
AMN	*100%*	*100%*	*Imerslund-Grasbeck syndrome 2, 618882*
AMPD1	*100%*	*100%*	*Myopathy due to myoadenylate deaminase deficiency, 615511*
AMPD2	*100%*	*100%*	*?Spastic paraplegia 63, 615686* *Pontocerebellar hypoplasia, type 9, 615809*
AMT	*100%*	*100%*	*Glycine encephalopathy, 605899*
ANAPC1	*100%*	*100%*	*Rothmund-Thomson syndrome, type 1, 618625*
ANGPTL3	*100%*	*100%*	*Hypobetalipoproteinemia, familial, 2, 605019*
ANK1	*100%*	*100%*	*Spherocytosis, type 1, 182900*
ANK3	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 37, 615493*
ANKH	*100%*	*100%*	*Chondrocalcinosis 2, 118600* *Craniometaphyseal dysplasia, 123000*
ANKLE2	*100%*	*100%*	*Microcephaly 16, primary, autosomal recessive, 616681*
ANKS6	*100%*	*99%*	*Nephronophthisis 16, 615382*
ANO10	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive 10, 613728*
ANO5	*100%*	*100%*	*Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307* *Miyoshi muscular dystrophy 3, 613319* *Gnathodiaphyseal dysplasia, 166260*
ANO6	*100%*	*100%*	*Scott syndrome, 262890*
ANTXR1	*100%*	*100%*	*GAPO syndrome, 230740*
ANTXR2	*100%*	*100%*	*Hyaline fibromatosis syndrome, 228600*
AP1B1	*100%*	*100%*	*Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150*
AP1S1	*100%*	*100%*	*MEDNIK syndrome, 609313*
AP3B1	*100%*	*100%*	*Hermansky-Pudlak syndrome 2, 608233*
AP3B2	*100%*	*100%*	*Developmental and epileptic encephalopathy 48, 617276*
AP3D1	*100%*	*100%*	*?Hermansky-Pudlak syndrome 10, 617050*
AP4B1	*100%*	*100%*	*Spastic paraplegia 47, autosomal recessive, 614066*
AP4E1	*100%*	*100%*	*Stuttering, familial persistent, 1, 184450* *Spastic paraplegia 51, autosomal recessive, 613744*
AP4M1	*100%*	*100%*	*Spastic paraplegia 50, autosomal recessive, 612936*

AP4S1	*87%*	*87%*	*Spastic paraplegia 52, autosomal recessive, 614067*
AP5Z1	*100%*	*100%*	*Spastic paraplegia 48, autosomal recessive, 613647*
APC2	*100%*	*100%*	*Cortical dysplasia, complex, with other brain malformations 10, 618677* *Intellectual developmental disorder, autosomal recessive 74, 617169*
APOC2	*100%*	*100%*	*Hyperlipoproteinemia, type Ib, 207750*
APOE	*100%*	*100%*	*Alzheimer disease 2, 104310* *Sea-blue histiocyte disease, 269600* *Lipoprotein glomerulopathy, 611771* *Hyperlipoproteinemia, type III, 617347*
APRT	*100%*	*100%*	*Adenine phosphoribosyltransferase deficiency, 614723*
APTX	*100%*	*100%*	*Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920*
AQP2	*100%*	*100%*	*Diabetes insipidus, nephrogenic, 2, 125800*
ARFGEF2	*100%*	*100%*	*Periventricular heterotopia with microcephaly, 608097*
ARG1	*93%*	*93%*	*Argininemia, 207800*
ARHGDIA	*100%*	*100%*	*Nephrotic syndrome, type 8, 615244*
ARHGEF18	*100%*	*100%*	*Retinitis pigmentosa 78, 617433*
ARHGEF2	*100%*	*100%*	*?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523*
ARL13B	*100%*	*100%*	*Joubert syndrome 8, 612291*
ARL2BP	*100%*	*100%*	*Retinitis pigmentosa with or without situs inversus, 615434*
ARL3	*100%*	*100%*	*Retinitis pigmentosa 83, 618173* *Joubert syndrome 35, 618161*
ARL6	*100%*	*100%*	*Retinitis pigmentosa 55, 613575* *Bardet-Biedl syndrome 3, 600151*
ARL6IP1	*100%*	*100%*	*?Spastic paraplegia 61, autosomal recessive, 615685*
ARMC9	*100%*	*100%*	*Joubert syndrome 30, 617622*
ARNT2	*100%*	*100%*	*?Webb-Dattani syndrome, 615926*
ARPC1B	*100%*	*100%*	*Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718*
ARSA	*100%*	*100%*	*Metachromatic leukodystrophy, 250100*
ARSB	*100%*	*100%*	*Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200*
ARSG	*100%*	*100%*	*Usher syndrome, type IV, 618144*
ARV1	*100%*	*100%*	*Developmental and epileptic encephalopathy 38, 617020*
ASAH1	*100%*	*100%*	*Spinal muscular atrophy with progressive myoclonic epilepsy, 159950* *Farber lipogranulomatosis, 228000*

ASCC1	*87%*	*87%*	*Spinal muscular atrophy with congenital bone fractures 2, 616867* *Barrett esophagus/esophageal adenocarcinoma, 614266*
ASL	*100%*	*100%*	*Argininosuccinic aciduria, 207900*
ASNS	*100%*	*100%*	*Asparagine synthetase deficiency, 615574*
ASPA	*100%*	*100%*	*Canavan disease, 271900*
ASPH	*100%*	*100%*	*Traboulsi syndrome, 601552*
ASPM	*100%*	*100%*	*Microcephaly 5, primary, autosomal recessive, 608716*
ASS1	*100%*	*100%*	*Citrullinemia, 215700*
ATAD1	*100%*	*100%*	*Hyperekplexia 4, 618011*
ATAD3A	*100%*	*100%*	*Harel-Yoon syndrome, 617183* *Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810*
ATCAY	*100%*	*100%*	*Ataxia, cerebellar, Cayman type, 601238*
ATF6	*100%*	*100%*	*Achromatopsia 7, 616517*
ATG5	*100%*	*100%*	*?Spinocerebellar ataxia, autosomal recessive 25, 617584*
ATIC	*100%*	*100%*	*AICA-ribosiduria due to ATIC deficiency, 608688*
ATM	*100%*	*100%*	*Ataxia-telangiectasia, 208900* *Lymphoma, B-cell non-Hodgkin, somatic,* *T-cell prolymphocytic leukemia, somatic,* *Lymphoma, mantle cell, somatic,*
ATOH7	*100%*	*100%*	*Persistent hyperplastic primary vitreous, autosomal recessive, 221900*
ATP13A2	*100%*	*100%*	*Spastic paraplegia 78, autosomal recessive, 617225* *Kufor-Rakeb syndrome, 606693*
ATP2A1	*100%*	*100%*	*Brody myopathy, 601003*
ATP5F1A	*100%*	*100%*	*?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228* *?Combined oxidative phosphorylation deficiency 22, 616045*
ATP5F1D	*100%*	*100%*	*Mitochondrial complex V (ATP synthase) deficiency, 618120*
ATP5F1E	*100%*	*100%*	*?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053*
ATP6V0A2	*100%*	*100%*	*Wrinkly skin syndrome, 278250* *Cutis laxa, autosomal recessive, type IIA, 219200*
ATP6V0A4	*100%*	*100%*	*Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722*
ATP6V1A	*100%*	*100%*	*Cutis laxa, autosomal recessive, type IID, 617403* *Developmental and epileptic encephalopathy 93, 618012*
ATP6V1B1	*100%*	*100%*	*Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300*
ATP6V1E1	*100%*	*100%*	*Cutis laxa, autosomal recessive, type IIC, 617402*

ATP7B	*100%*	*100%*	*Wilson disease, 277900*
ATP8A2	*100%*	*100%*	*?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268*
ATP8B1	*100%*	*100%*	*Cholestasis, progressive familial intrahepatic 1, 211600* *Cholestasis, intrahepatic, of pregnancy, 1, 147480* *Cholestasis, benign recurrent intrahepatic, 243300*
ATPAF2	*100%*	*100%*	*?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273*
ATR	*100%*	*100%*	*Seckel syndrome 1, 210600* *?Cutaneous telangiectasia and cancer syndrome, familial, 614564*
AUH	*100%*	*100%*	*3-methylglutaconic aciduria, type I, 250950*
AURKC	*100%*	*100%*	*Spermatogenic failure 5, 243060*
B2M	*100%*	*100%*	*?Amyloidosis, familial visceral, 105200* *Immunodeficiency 43, 241600*
B3GALNT2	*92%*	*92%*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181*
B3GALT6	*100%*	*98%*	*Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349* *Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640* *Al-Gazali syndrome, 609465*
B3GAT3	*95%*	*94%*	*Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600*
B3GLCT	*100%*	*100%*	*Peters-plus syndrome, 261540*
B4GALNT1	*100%*	*100%*	*Spastic paraplegia 26, autosomal recessive, 609195*
B4GALT1	*100%*	*100%*	*Congenital disorder of glycosylation, type IId, 607091*
B4GALT7	*100%*	*100%*	*Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070*
B4GAT1	*100%*	*100%*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287*
B9D1	*100%*	*100%*	*?Meckel syndrome 9, 614209* *Joubert syndrome 27, 617120*
B9D2	*100%*	*100%*	*?Meckel syndrome 10, 614175* *Joubert syndrome 34, 614175*
BAAT	*100%*	*100%*	*Bile acid conjugation defect 1, 619232*
BANF1	*100%*	*100%*	*Nestor-Guillermo progeria syndrome, 614008*
BBS1	*100%*	*100%*	*Bardet-Biedl syndrome 1, 209900*
BBS10	*100%*	*100%*	*Bardet-Biedl syndrome 10, 615987*
BBS12	*100%*	*100%*	*Bardet-Biedl syndrome 12, 615989*
BBS2	*100%*	*100%*	*Retinitis pigmentosa 74, 616562* *Bardet-Biedl syndrome 2, 615981*
BBS4	*100%*	*100%*	*Bardet-Biedl syndrome 4, 615982*
BBS5	*100%*	*100%*	*Bardet-Biedl syndrome 5, 615983*

BBS7	*100%*	*100%*	*Bardet-Biedl syndrome 7, 615984*
BBS9	*96%*	*96%*	*Bardet-Biedl syndrome 9, 615986*
BCAS3	*100%*	*100%*	*Hengel-Maroofian-Schols syndrome, 619641*
BCKDHA	*100%*	*100%*	*Maple syrup urine disease, type Ia, 248600*
BCKDHB	*100%*	*100%*	*Maple syrup urine disease, type Ib, 248600*
BCKDK	*100%*	*100%*	*Branched-chain keto acid dehydrogenase kinase deficiency, 614923*
BCL10	*100%*	*100%*	*?Immunodeficiency 37, 616098* *Lymphoma, MALT, somatic, 137245*
BCS1L	*100%*	*100%*	*GRACILE syndrome, 603358* *Mitochondrial complex III deficiency, nuclear type 1, 124000* *Bjornstad syndrome, 262000*
BFSP1	*100%*	*100%*	*Cataract 33, multiple types, 611391*
BFSP2	*100%*	*100%*	*Cataract 12, multiple types, 611597*
BHLHA9	*100%*	*100%*	*?Camptosynpolydactyly, complex, 607539* *Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432*
BIN1	*100%*	*100%*	*Centronuclear myopathy 2, 255200*
BLM	*100%*	*100%*	*Bloom syndrome, 210900*
BLOC1S3	*100%*	*100%*	*Hermansky-Pudlak syndrome 8, 614077*
BLOC1S6	*100%*	*100%*	*?Hermansky-Pudlak syndrome 9, 614171*
KIAA1109	*100%*	*100%*	*Alkuraya-Kucinskas syndrome, 617822*
BLVRA	*100%*	*100%*	*Hyperbiliverdinemia, 614156*
BMP1	*100%*	*100%*	*Osteogenesis imperfecta, type XIII, 614856*
BMPER	*100%*	*100%*	*Diaphanospondylodysostosis, 608022*
BMPR1B	*100%*	*100%*	*Acromesomelic dysplasia 3, 609441* *Brachydactyly, type A2, 112600* *Brachydactyly, type A1, D, 616849*
BOLA3	*100%*	*100%*	*Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299*
BPGM	*100%*	*100%*	*Erythrocytosis, familial, 8, 222800*
IMPAD1	*100%*	*100%*	*Chondrodysplasia with joint dislocations, GPAPP type, 614078*
BRAT1	*100%*	*100%*	*Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056* *Rigidity and multifocal seizure syndrome, lethal neonatal, 614498*
BRCA1	*100%*	*100%*	*Fanconi anemia, complementation group S, 617883*
BRCA2	*100%*	*100%*	*Fanconi anemia, complementation group D1, 605724* *Wilms tumor, 194070*

BRF1	*100%*	*100%*	*Cerebellofaciodental syndrome, 616202*
BRIP1	*100%*	*100%*	*Fanconi anemia, complementation group J, 609054*
BSCL2	*100%*	*100%*	*Lipodystrophy, congenital generalized, type 2, 269700* *Neuropathy, distal hereditary motor, type VC, 619112* *Silver spastic paraplegia syndrome, 270685* *Encephalopathy, progressive, with or without lipodystrophy, 615924*
BSND	*100%*	*100%*	*Sensorineural deafness with mild renal dysfunction, 602522* *Bartter syndrome, type 4a, 602522*
BTD	*94%*	*94%*	*Biotinidase deficiency, 253260*
BUB1B	*100%*	*100%*	*Colorectal cancer, somatic, 114500* *Mosaic variegated aneuploidy syndrome 1, 257300*
BVES	*100%*	*100%*	*Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812*
C12orf4	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 66, 618221*
C12orf57	*100%*	*100%*	*Temtamy syndrome, 218340*
C19orf12	*100%*	*100%*	*Neurodegeneration with brain iron accumulation 4, 614298* *?Spastic paraplegia 43, autosomal recessive, 615043*
C1QA	*100%*	*100%*	*C1q deficiency, 613652*
C1QB	*100%*	*100%*	*C1q deficiency, 613652*
C1QBP	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 33, 617713*
C1QC	*100%*	*100%*	*C1q deficiency, 613652*
C1S	*100%*	*99%*	*C1s deficiency, 613783* *Ehlers-Danlos syndrome, periodontal type, 2, 617174*
C2CD3	*96%*	*96%*	*Orofaciodigital syndrome XIV, 615948*
C2orf69	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 53, 619423*
C3	*100%*	*100%*	*C3 deficiency, 613779*
C4A	*100%*	*99%*	*C4a deficiency, 614380*
C4B	*100%*	*100%*	*C4B deficiency, 614379*
C5	*100%*	*100%*	*C5 deficiency, 609536*
C8A	*100%*	*100%*	*C8 deficiency, type I, 613790*
C8B	*100%*	*100%*	*C8 deficiency, type II, 613789*
C9	*100%*	*100%*	*C9 deficiency, 613825*
CA12	*100%*	*100%*	*Hyperchlorhidrosis, isolated, 143860*
CA2	*100%*	*100%*	*Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730*
CA5A	*100%*	*100%*	*Hyperammonemia due to carbonic anhydrase VA deficiency, 615751*

CA8	*100%*	*100%*	*Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227*
CABP2	*100%*	*100%*	*Deafness, autosomal recessive 93, 614899*
CABP4	*100%*	*100%*	*Cone-rod synaptic disorder, congenital nonprogressive, 610427*
CACNA1B	*100%*	*100%*	*Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497*
CACNA1D	*100%*	*100%*	*Primary aldosteronism, seizures, and neurologic abnormalities, 615474* *Sinoatrial node dysfunction and deafness, 614896*
CACNA2D2	*100%*	*100%*	*Cerebellar atrophy with seizures and variable developmental delay, 618501*
CAD	*100%*	*100%*	*Developmental and epileptic encephalopathy 50, 616457*
CAMK2A	*100%*	*100%*	*Intellectual developmental disorder, autosomal dominant 53, 617798* *?Intellectual developmental disorder, autosomal recessive 63, 618095*
CANT1	*100%*	*100%*	*Desbuquois dysplasia 1, 251450* *Epiphyseal dysplasia, multiple, 7, 617719*
CAPN1	*100%*	*100%*	*Spastic paraplegia 76, autosomal recessive, 616907*
CAPN10	*100%*	*100%*	*No OMIM disease ID*
CAPN3	*100%*	*100%*	*Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600* *Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129*
CARD11	*100%*	*100%*	*B-cell expansion with NFKB and T-cell anergy, 616452* *Immunodeficiency 11B with atopic dermatitis, 617638* *Immunodeficiency 11A, 615206*
CARD9	*100%*	*100%*	*Immunodeficiency 103, susceptibility to fungal infection, 212050*
CARS2	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 27, 616672*
CASP14	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 12, 617320*
CASP8	*95%*	*95%*	*?Caspase 8 lymphadenopathy syndrome, 607271* *Hepatocellular carcinoma, somatic, 114550*
CASQ2	*100%*	*100%*	*Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938*
CASR	*100%*	*100%*	*Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198* *Hyperparathyroidism, neonatal, 239200* *Hypocalcemia, autosomal dominant, 601198* *Hypocalciuric hypercalcemia, type I, 145980*
CAST	*100%*	*100%*	*Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295*
CAT	*100%*	*100%*	*Acatalasemia, 614097*
CATSPER1	*100%*	*100%*	*Spermatogenic failure 7, 612997*
CAV1	*100%*	*100%*	*?Lipodystrophy, congenital generalized, type 3, 612526* *Pulmonary hypertension, primary, 3, 615343* *Lipodystrophy, familial partial, type 7, 606721*

CAVIN1	*100%*	*100%*	*Lipodystrophy, congenital generalized, type 4, 613327*
CBLIF	*100%*	*100%*	*Intrinsic factor deficiency, 261000*
CBS	*100%*	*100%*	*Thrombosis, hyperhomocysteinemic, 236200* *Homocystinuria, B6-responsive and nonresponsive types, 236200*
CC2D1A	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 3, 608443*
CC2D2A	*98%*	*98%*	*COACH syndrome 2, 619111* *Retinitis pigmentosa 93, 619845* *Meckel syndrome 6, 612284* *Joubert syndrome 9, 612285*
CCBE1	*100%*	*100%*	*Hennekam lymphangiectasia-lymphedema syndrome 1, 235510*
CCDC103	*100%*	*100%*	*Ciliary dyskinesia, primary, 17, 614679*
CCDC115	*100%*	*100%*	*Congenital disorder of glycosylation, type IIo, 616828*
CCDC174	*100%*	*100%*	*Hypotonia, infantile, with psychomotor retardation, 616816*
CCDC39	*100%*	*100%*	*Ciliary dyskinesia, primary, 14, 613807*
CCDC40	*100%*	*100%*	*Ciliary dyskinesia, primary, 15, 613808*
CCDC47	*100%*	*100%*	*Trichohepatoneurodevelopmental syndrome, 618268*
CCDC65	*100%*	*100%*	*Ciliary dyskinesia, primary, 27, 615504*
CCDC8	*100%*	*100%*	*3-M syndrome 3, 614205*
CCDC88A	*97%*	*97%*	*?PEHO syndrome-like, 617507*
CCDC88C	*100%*	*100%*	*?Spinocerebellar ataxia 40, 616053* *Hydrocephalus, congenital, 1, 236600*
CCN6	*100%*	*100%*	*Progressive pseudorheumatoid dysplasia, 208230*
CCNO	*100%*	*100%*	*Ciliary dyskinesia, primary, 29, 615872*
CCT5	*100%*	*100%*	*Neuropathy, hereditary sensory, with spastic paraplegia, 256840*
CD151	*100%*	*100%*	*Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057*
CD19	*100%*	*100%*	*Immunodeficiency, common variable, 3, 613493*
CD247	*100%*	*100%*	*?Immunodeficiency 25, 610163*
CD27	*100%*	*100%*	*Lymphoproliferative syndrome 2, 615122*
CD2AP	*100%*	*100%*	*Glomerulosclerosis, focal segmental, 3, 607832*
CD320	*100%*	*100%*	*Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646*
CD3D	*100%*	*100%*	*Immunodeficiency 19, severe combined, 615617*
CD3E	*100%*	*100%*	*Immunodeficiency 18, 615615* *Immunodeficiency 18, SCID variant, 615615*
CD3G	*100%*	*100%*	*Immunodeficiency 17, CD3 gamma deficient, 615607*

CD40	*100%*	*100%*	*Immunodeficiency with hyper-IgM, type 3, 606843*
CD55	*96%*	*93%*	*Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300*
CD59	*100%*	*100%*	*Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300*
CD79A	*100%*	*100%*	*Agammaglobulinemia 3, 613501*
CD79B	*100%*	*100%*	*Agammaglobulinemia 6, 612692*
CD81	*100%*	*100%*	*Immunodeficiency, common variable, 6, 613496*
CD8A	*100%*	*100%*	*CD8 deficiency, familial, 608957*
CDAN1	*100%*	*100%*	*Dyserythropoietic anemia, congenital, type Ia, 224120*
CDC14A	*100%*	*100%*	*Deafness, autosomal recessive 32, with or without immotile sperm, 608653*
CDC45	*100%*	*100%*	*Meier-Gorlin syndrome 7, 617063*
CDC6	*100%*	*100%*	*?Meier-Gorlin syndrome 5, 613805*
CDCA7	*100%*	*100%*	*Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910*
CDH11	*100%*	*100%*	*Teebi hypertelorism syndrome 2, 619736* *Elsahy-Waters syndrome, 211380*
CDH23	*100%*	*100%*	*Usher syndrome, type 1D, 601067* *Usher syndrome, type 1D/F digenic, 601067* *Deafness, autosomal recessive 12, 601386*
CDH3	*100%*	*100%*	*Hypotrichosis, congenital, with juvenile macular dystrophy, 601553* *Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280*
CDHR1	*100%*	*100%*	*Cone-rod dystrophy 15, 613660* *Retinitis pigmentosa 65, 613660*
C15orf41	*100%*	*100%*	*Dyserythropoietic anemia, congenital, type Ib, 615631*
CDK10	*100%*	*100%*	*Al Kaissi syndrome, 617694*
CDK5	*100%*	*100%*	*?Lissencephaly 7 with cerebellar hypoplasia, 616342*
CDK5RAP2	*100%*	*100%*	*Microcephaly 3, primary, autosomal recessive, 604804*
CDK6	*100%*	*100%*	*?Microcephaly 12, primary, autosomal recessive, 616080*
CDSN	*100%*	*100%*	*Hypotrichosis 2, 146520* *Peeling skin syndrome 1, 270300*
CDT1	*100%*	*100%*	*Meier-Gorlin syndrome 4, 613804*
CEACAM16	*100%*	*100%*	*Deafness, autosomal dominant 4B, 614614* *Deafness, autosomal recessive 113, 618410*
CEBPE	*100%*	*100%*	*?Immunodeficiency 108 with autoinflammation, 260570* *Specific granule deficiency, 245480*
CENPE	*100%*	*100%*	*?Microcephaly 13, primary, autosomal recessive, 616051*

CENPF	*100%*	*100%*	*Stromme syndrome, 243605*
CENPJ	*100%*	*100%*	*Microcephaly 6, primary, autosomal recessive, 608393* *?Seckel syndrome 4, 613676*
CEP104	*100%*	*100%*	*Joubert syndrome 25, 616781* *Intellectual developmental disorder, autosomal recessive 77, 619988*
CEP120	*100%*	*100%*	*Short-rib thoracic dysplasia 13 with or without polydactyly, 616300* *Joubert syndrome 31, 617761*
CEP135	*100%*	*100%*	*Microcephaly 8, primary, autosomal recessive, 614673*
CEP152	*100%*	*100%*	*Microcephaly 9, primary, autosomal recessive, 614852* *Seckel syndrome 5, 613823*
CEP164	*100%*	*100%*	*Nephronophthisis 15, 614845*
CEP19	*100%*	*100%*	*Morbid obesity and spermatogenic failure, 615703*
CEP290	*100%*	*100%*	*Leber congenital amaurosis 10, 611755* *Joubert syndrome 5, 610188* *Senior-Loken syndrome 6, 610189* *?Bardet-Biedl syndrome 14, 615991* *Meckel syndrome 4, 611134*
CEP41	*100%*	*100%*	*Joubert syndrome 15, 614464*
CEP55	*100%*	*100%*	*Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500*
CEP57	*100%*	*100%*	*Mosaic variegated aneuploidy syndrome 2, 614114*
CEP63	*100%*	*100%*	*?Seckel syndrome 6, 614728*
CEP78	*100%*	*100%*	*Cone-rod dystrophy and hearing loss, 617236*
CEP83	*100%*	*100%*	*Nephronophthisis 18, 615862*
CERKL	*99%*	*98%*	*Retinitis pigmentosa 26, 608380*
CERS1	*100%*	*100%*	*Epilepsy, progressive myoclonic, 8, 616230*
CERS3	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 9, 615023*
CFAP298	*100%*	*100%*	*Ciliary dyskinesia, primary, 26, 615500*
CFAP410	*100%*	*100%*	*Retinal dystrophy with macular staphyloma, 617547* *Spondylometaphyseal dysplasia, axial, 602271*
C8orf37	*100%*	*100%*	*Retinitis pigmentosa 64, 614500* *Cone-rod dystrophy 16, 614500* *Bardet-Biedl syndrome 21, 617406*
CFAP53	*100%*	*100%*	*Heterotaxy, visceral, 6, autosomal recessive, 614779*
CFD	*100%*	*100%*	*Complement factor D deficiency, 613912*

CFH	*100%*	*100%*	*Basal laminar drusen, 126700* *Complement factor H deficiency, 609814*
CFI	*100%*	*100%*	*Complement factor I deficiency, 610984*
CFL2	*100%*	*100%*	*Nemaline myopathy 7, autosomal recessive, 610687*
CFTR	*100%*	*100%*	*Cystic fibrosis, 219700* *Congenital bilateral absence of vas deferens, 277180* *Sweat chloride elevation without CF,*
CHAT	*100%*	*100%*	*Myasthenic syndrome, congenital, 6, presynaptic, 254210*
CHKB	*100%*	*100%*	*Muscular dystrophy, congenital, megaconial type, 602541*
CHMP1A	*100%*	*100%*	*Pontocerebellar hypoplasia, type 8, 614961*
CHP1	*100%*	*100%*	*?Spastic ataxia 9, autosomal recessive, 618438*
CHRM3	*100%*	*100%*	*Prune belly syndrome, 100100*
CHRNA1	*100%*	*100%*	*Myasthenic syndrome, congenital, 1B, fast-channel, 608930* *Myasthenic syndrome, congenital, 1A, slow-channel, 601462* *Multiple pterygium syndrome, lethal type, 253290*
CHRNB1	*100%*	*100%*	*?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314* *Myasthenic syndrome, congenital, 2A, slow-channel, 616313*
CHRND	*100%*	*100%*	*?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323* *Multiple pterygium syndrome, lethal type, 253290* *Myasthenic syndrome, congenital, 3B, fast-channel, 616322* *?Myasthenic syndrome, congenital, 3A, slow-channel, 616321*
CHRNE	*100%*	*100%*	*Myasthenic syndrome, congenital, 4A, slow-channel, 605809* *Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931* *Myasthenic syndrome, congenital, 4B, fast-channel, 616324*
CHRNG	*100%*	*100%*	*Multiple pterygium syndrome, lethal type, 253290* *Escobar syndrome, 265000*
CHST11	*100%*	*100%*	*?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167*
CHST14	*100%*	*100%*	*Ehlers-Danlos syndrome, musculocontractural type 1, 601776*
CHST3	*100%*	*100%*	*Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095*
CHST6	*100%*	*100%*	*Macular corneal dystrophy, 217800*
CHST8	*100%*	*100%*	*?Peeling skin syndrome 3, 616265*
CHSY1	*100%*	*100%*	*Temtamy preaxial brachydactyly syndrome, 605282*
CIB2	*100%*	*100%*	*Deafness, autosomal recessive 48, 609439* *Usher syndrome, type IJ, 614869*
CIDEC	*100%*	*100%*	*?Lipodystrophy, familial partial, type 5, 615238*

CIITA	*100%*	*100%*	*Bare lymphocyte syndrome, type II, complementation group A, 209920*
CILK1	*100%*	*100%*	*Endocrine-cerebroosteodysplasia, 612651*
CISD2	*100%*	*100%*	*Wolfram syndrome 2, 604928*
CIT	*100%*	*100%*	*Microcephaly 17, primary, autosomal recessive, 617090*
CKAP2L	*100%*	*100%*	*Filippi syndrome, 272440*
CLCF1	*100%*	*100%*	*Cold-induced sweating syndrome 2, 610313*
CLCN1	*100%*	*100%*	*Myotonia congenita, recessive, 255700* *Myotonia congenita, dominant, 160800* *Myotonia levior, recessive,*
CLCN2	*100%*	*100%*	*Leukoencephalopathy with ataxia, 615651* *Hyperaldosteronism, familial, type II, 605635*
CLCN7	*100%*	*100%*	*Hypopigmentation, organomegaly, and delayed myelination and development, 618541* *Osteopetrosis, autosomal recessive 4, 611490* *Osteopetrosis, autosomal dominant 2, 166600*
CLCNKB	*100%*	*100%*	*Bartter syndrome, type 3, 607364* *Bartter syndrome, type 4b, digenic, 613090*
CLDN1	*100%*	*100%*	*Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626*
CLDN10	*100%*	*100%*	*HELIX syndrome, 617671*
CLDN14	*100%*	*100%*	*Deafness, autosomal recessive 29, 614035*
CLDN16	*100%*	*100%*	*Hypomagnesemia 3, renal, 248250*
CLDN19	*100%*	*100%*	*Hypomagnesemia 5, renal, with ocular involvement, 248190*
CLIP1	*100%*	*100%*	*No OMIM disease ID*
CLMP	*100%*	*100%*	*Congenital short bowel syndrome, 615237*
CLN3	*93%*	*93%*	*Ceroid lipofuscinosis, neuronal, 3, 204200*
CLN5	*83%*	*83%*	*Ceroid lipofuscinosis, neuronal, 5, 256731*
CLN6	*100%*	*100%*	*Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300* *Ceroid lipofuscinosis, neuronal, 6A, 601780*
CLN8	*100%*	*100%*	*Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003* *Ceroid lipofuscinosis, neuronal, 8, 600143*
CLP1	*100%*	*100%*	*Pontocerebellar hypoplasia, type 10, 615803*
CLPB	*100%*	*100%*	*Neutropenia, severe congenital, 9, autosomal dominant, 619813* *3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271* *3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835*
CLPP	*100%*	*100%*	*Perrault syndrome 3, 614129*

CLRN1	*100%*	*100%*	*Usher syndrome, type 3A, 276902* *Retinitis pigmentosa 61, 614180*
CNGA1	*91%*	*91%*	*Retinitis pigmentosa 49, 613756*
CNGA3	*100%*	*100%*	*Achromatopsia 2, 216900*
CNGB1	*100%*	*100%*	*Retinitis pigmentosa 45, 613767*
CNGB3	*100%*	*100%*	*Achromatopsia 3, 262300*
CNNM2	*100%*	*100%*	*Hypomagnesemia 6, renal, 613882* *Hypomagnesemia, seizures, and impaired intellectual development 1, 616418*
CNNM4	*100%*	*100%*	*Jalili syndrome, 217080*
CNPY3	*100%*	*100%*	*Developmental and epileptic encephalopathy 60, 617929*
CNTN1	*100%*	*100%*	*?Myopathy, congenital, Compton-North, 612540*
CNTN2	*100%*	*100%*	*?Epilepsy, myoclonic, familial adult, 5, 615400*
CNTNAP1	*100%*	*100%*	*Lethal congenital contracture syndrome 7, 616286* *Hypomyelinating neuropathy, congenital, 3, 618186*
CNTNAP2	*100%*	*100%*	*Pitt-Hopkins like syndrome 1, 610042*
COA5	*82%*	*82%*	*?Mitochondrial complex IV, deficiency, nuclear type 9, 616500*
COA6	*100%*	*100%*	*Mitochondrial complex IV deficiency, nuclear type 13, 616501*
COA7	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387*
COA8	*100%*	*100%*	*Mitochondrial complex IV deficiency, nuclear type 17, 619061*
COASY	*100%*	*100%*	*Pontocerebellar hypoplasia, type 12, 618266* *Neurodegeneration with brain iron accumulation 6, 615643*
COCH	*100%*	*100%*	*Deafness, autosomal dominant 9, 601369* *?Deafness, autosomal recessive 110, 618094*
COG1	*100%*	*100%*	*Congenital disorder of glycosylation, type IIg, 611209*
COG4	*100%*	*100%*	*Congenital disorder of glycosylation, type IIj, 613489* *Saul-Wilson syndrome, 618150*
COG5	*100%*	*100%*	*Congenital disorder of glycosylation, type IIi, 613612*
COG6	*100%*	*100%*	*Shaheen syndrome, 615328* *Congenital disorder of glycosylation, type III, 614576*
COG7	*100%*	*100%*	*Congenital disorder of glycosylation, type IIe, 608779*
COG8	*100%*	*100%*	*Congenital disorder of glycosylation, type IIh, 611182*
COL11A1	*100%*	*100%*	*Fibrochondrogenesis 1, 228520* *Stickler syndrome, type II, 604841* *Marshall syndrome, 154780* *Deafness, autosomal dominant 37, 618533*

COL11A2	*100%*	*100%*	*Deafness, autosomal dominant 13, 601868* *Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150* *Fibrochondrogenesis 2, 614524* *Deafness, autosomal recessive 53, 609706* *Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840*
COL12A1	*100%*	*100%*	*Bethlem myopathy 2, 616471* *?Ullrich congenital muscular dystrophy 2, 616470*
COL13A1	*100%*	*100%*	*Myasthenic syndrome, congenital, 19, 616720*
COL17A1	*100%*	*100%*	*Epithelial recurrent erosion dystrophy, 122400* *Epidermolysis bullosa, junctional 4, intermediate, 619787*
COL18A1	*100%*	*100%*	*Knobloch syndrome, type 1, 267750* *Glaucoma, primary closed-angle, 618880*
COL1A2	*100%*	*100%*	*Osteogenesis imperfecta, type III, 259420* *Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821* *Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120* *Ehlers-Danlos syndrome, cardiac valvular type, 225320* *Osteogenesis imperfecta, type IV, 166220* *Osteogenesis imperfecta, type II, 166210*
COL25A1	*99%*	*99%*	*Fibrosis of extraocular muscles, congenital, 5, 616219*
COL27A1	*100%*	*100%*	*Steel syndrome, 615155*
COL3A1	*100%*	*100%*	*Ehlers-Danlos syndrome, vascular type, 130050* *Polymicrogyria with or without vascular-type EDS, 618343*
COL4A3	*100%*	*100%*	*Hematuria, benign familial, 141200* *Alport syndrome 3, autosomal dominant, 104200* *Alport syndrome 2, autosomal recessive, 203780*
COL4A4	*100%*	*100%*	*Hematuria, familial benign, 141200* *Alport syndrome 2, autosomal recessive, 203780*
COL6A1	*100%*	*100%*	*Bethlem myopathy 1, 158810* *Ullrich congenital muscular dystrophy 1, 254090*
COL6A2	*100%*	*100%*	*Bethlem myopathy 1, 158810* *?Myosclerosis, congenital, 255600* *Ullrich congenital muscular dystrophy 1, 254090*
COL6A3	*100%*	*100%*	*Ullrich congenital muscular dystrophy 1, 254090* *Dystonia 27, 616411* *Bethlem myopathy 1, 158810*

COL7A1	*100%*	*100%*	*Epidermolysis bullosa, pretibial, 131850* *Transient bullous of the newborn, 131705* *EBD, Bart type, 132000* *Epidermolysis bullosa dystrophica, AD, 131750* *Epidermolysis bullosa pruriginosa, 604129* *EBD inversa, 226600* *Epidermolysis bullosa dystrophica, AR, 226600* *Toenail dystrophy, isolated, 607523* *EBD, localisata variant,*
COL9A1	*100%*	*100%*	*Stickler syndrome, type IV, 614134* *?Epiphyseal dysplasia, multiple, 6, 614135*
COL9A2	*100%*	*100%*	*Epiphyseal dysplasia, multiple, 2, 600204* *?Stickler syndrome, type V, 614284*
COLEC10	*100%*	*100%*	*3MC syndrome 3, 248340*
COLEC11	*100%*	*100%*	*3MC syndrome 2, 265050*
COLGALT1	*100%*	*100%*	*Brain small vessel disease 3, 618360*
COLQ	*100%*	*100%*	*Myasthenic syndrome, congenital, 5, 603034*
COPB2	*100%*	*100%*	*Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884* *?Microcephaly 19, primary, autosomal recessive, 617800*
COQ2	*96%*	*96%*	*Coenzyme Q10 deficiency, primary, 1, 607426*
COQ4	*100%*	*100%*	*Coenzyme Q10 deficiency, primary, 7, 616276*
COQ6	*100%*	*100%*	*Coenzyme Q10 deficiency, primary, 6, 614650*
COQ8A	*100%*	*100%*	*Coenzyme Q10 deficiency, primary, 4, 612016*
COQ8B	*100%*	*100%*	*Nephrotic syndrome, type 9, 615573*
COQ9	*100%*	*100%*	*Coenzyme Q10 deficiency, primary, 5, 614654*
CORO1A	*100%*	*100%*	*Immunodeficiency 8, 615401*
COX10	*100%*	*100%*	*Mitochondrial complex IV deficiency, nuclear type 3, 619046*
COX14	*100%*	*100%*	*?Mitochondrial complex IV deficiency, nuclear type 10, 619053*
COX15	*100%*	*100%*	*Mitochondrial complex IV deficiency, nuclear type 6, 615119*
COX20	*100%*	*100%*	*Mitochondrial complex IV deficiency, nuclear type 11, 619054*
COX4I2	*100%*	*100%*	*Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714*
COX6A1	*100%*	*100%*	*Charcot-Marie-Tooth disease, recessive intermediate D, 616039*
COX6A2	*100%*	*100%*	*Mitochondrial complex IV deficiency, nuclear type 18, 619062*
COX6B1	*100%*	*100%*	*Mitochondrial complex IV deficiency, nuclear type 7, 619051*
COX8A	*100%*	*100%*	*?Mitochondrial complex IV deficiency, nuclear type 15, 619059*

CP	*100%*	*100%*	*Cerebellar ataxia, 604290* *Hemosiderosis, systemic, due to aceruloplasminemia, 604290*
CPA6	*100%*	*100%*	*Febrile seizures, familial, 11, 614418* *Epilepsy, familial temporal lobe, 5, 614417*
CPAMD8	*100%*	*100%*	*Anterior segment dysgenesis 8, 617319*
CPLANE1	*100%*	*100%*	*Orofaciodigital syndrome VI, 277170* *Joubert syndrome 17, 614615*
CPLX1	*100%*	*100%*	*Developmental and epileptic encephalopathy 63, 617976*
CPN1	*100%*	*100%*	*Carboxypeptidase N deficiency, 212070*
CPOX	*100%*	*100%*	*Coproporphyria, 121300* *Harderoporphyria, 618892*
CPS1	*100%*	*100%*	*Carbamoylphosphate synthetase I deficiency, 237300*
CPT1A	*100%*	*100%*	*CPT deficiency, hepatic, type IA, 255120*
CPT2	*100%*	*100%*	*CPT II deficiency, infantile, 600649* *CPT II deficiency, lethal neonatal, 608836* *CPT II deficiency, myopathic, stress-induced, 255110*
CR2	*100%*	*100%*	*Immunodeficiency, common variable, 7, 614699*
CRADD	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499*
CRAT	*100%*	*100%*	*?Neurodegeneration with brain iron accumulation 8, 617917*
CRB1	*100%*	*100%*	*Leber congenital amaurosis 8, 613835* *Retinitis pigmentosa-12, 600105* *Pigmented paravenous chorioretinal atrophy, 172870*
CRB2	*100%*	*100%*	*Focal segmental glomerulosclerosis 9, 616220* *Ventriculomegaly with cystic kidney disease, 219730*
CRBN	*100%*	*99%*	*Intellectual developmental disorder, autosomal recessive 2, 607417*
CREB3L1	*100%*	*100%*	*Osteogenesis imperfecta, type XVI, 616229*
CRIPT	*100%*	*100%*	*Short stature with microcephaly and distinctive facies, 615789*
CRLF1	*100%*	*99%*	*Cold-induced sweating syndrome 1, 272430*
CRPPA	*100%*	*100%*	*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643*
CRTAP	*100%*	*100%*	*Osteogenesis imperfecta, type VII, 610682*
CRYAA	*100%*	*100%*	*Cataract 9, multiple types, 604219*

CRYAB	*100%*	*100%*	*Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869* *Myopathy, myofibrillar, 2, 608810* *Cataract 16, multiple types, 613763* *Cardiomyopathy, dilated, 1II, 615184*
CRYBB1	*100%*	*100%*	*Cataract 17, multiple types, 611544*
CRYBB3	*100%*	*100%*	*Cataract 22, 609741*
CSF1R	*100%*	*100%*	*Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476* *Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820*
CSF2RB	*100%*	*100%*	*Surfactant metabolism dysfunction, pulmonary, 5, 614370*
CSPP1	*100%*	*100%*	*Joubert syndrome 21, 615636*
CSTA	*100%*	*100%*	*Peeling skin syndrome 4, 607936*
CSTB	*100%*	*100%*	*Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800*
CTC1	*100%*	*100%*	*Cerebroretinal microangiopathy with calcifications and cysts, 612199*
CTDP1	*100%*	*100%*	*Congenital cataracts, facial dysmorphism, and neuropathy, 604168*
CTNNA2	*100%*	*99%*	*Cortical dysplasia, complex, with other brain malformations 9, 618174*
CTNS	*100%*	*100%*	*Cystinosis, nephropathic, 219800* *Cystinosis, ocular nonnephropathic, 219750* *Cystinosis, late-onset juvenile or adolescent nephropathic, 219900* *Cystinosis, atypical nephropathic, 219800*
CTPS1	*100%*	*100%*	*Immunodeficiency 24, 615897*
CTSA	*100%*	*100%*	*Galactosialidosis, 256540*
CTSC	*100%*	*100%*	*Periodontitis 1, juvenile, 170650* *Haim-Munk syndrome, 245010* *Papillon-Lefevre syndrome, 245000*
CTSD	*100%*	*100%*	*Ceroid lipofuscinosis, neuronal, 10, 610127*
CTSF	*100%*	*100%*	*Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362*
CTSK	*100%*	*100%*	*Pycnodysostosis, 265800*
CTU2	*100%*	*100%*	*Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142*
CUBN	*100%*	*100%*	*Imerslund-Grasbeck syndrome 1, 261100*
CUL7	*100%*	*100%*	*3-M syndrome 1, 273750*
CWC27	*100%*	*100%*	*Retinitis pigmentosa with or without skeletal anomalies, 250410*
CWF19L1	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive 17, 616127*
CYB5A	*100%*	*100%*	*Methemoglobinemia and ambiguous genitalia, 250790*
CYB5R3	*100%*	*100%*	*Methemoglobinemia, type I, 250800* *Methemoglobinemia, type II, 250800*

CYBA	*100%*	*100%*	*Chronic granulomatous disease 4, autosomal recessive, 233690*
CYC1	*100%*	*100%*	*Mitochondrial complex III deficiency, nuclear type 6, 615453*
CYP11A1	*100%*	*100%*	*Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743*
CYP11B1	*100%*	*100%*	*Aldosteronism, glucocorticoid-remediable, 103900* *Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010*
CYP11B2	*100%*	*100%*	*Hypoaldosteronism, congenital, due to CMO I deficiency, 203400* *Hypoaldosteronism, congenital, due to CMO II deficiency, 610600* *Aldosterone to renin ratio raised,*
CYP17A1	*100%*	*100%*	*17,20-lyase deficiency, isolated, 202110* *17-alpha-hydroxylase/17,20-lyase deficiency, 202110*
CYP19A1	*100%*	*100%*	*Aromatase deficiency, 613546* *Aromatase excess syndrome, 139300*
CYP1B1	*100%*	*100%*	*Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300* *Anterior segment dysgenesis 6, multiple subtypes, 617315*
CYP21A2	*100%*	*100%*	*Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910* *Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910*
CYP24A1	*100%*	*100%*	*Hypercalcemia, infantile, 1, 143880*
CYP26B1	*100%*	*100%*	*Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416*
CYP26C1	*100%*	*100%*	*Focal facial dermal dysplasia 4, 614974*
CYP27A1	*100%*	*100%*	*Cerebrotendinous xanthomatosis, 213700*
CYP27B1	*100%*	*100%*	*Vitamin D-dependent rickets, type I, 264700*
CYP2C8	*100%*	*100%*	*No OMIM disease ID*
CYP2R1	*100%*	*100%*	*Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081*
CYP2U1	*100%*	*100%*	*Spastic paraplegia 56, autosomal recessive, 615030*
CYP4F22	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 5, 604777*
CYP4V2	*100%*	*100%*	*Bietti crystalline corneoretinal dystrophy, 210370*
CYP7B1	*100%*	*100%*	*Spastic paraplegia 5A, autosomal recessive, 270800* *Bile acid synthesis defect, congenital, 3, 613812*
D2HGDH	*100%*	*100%*	*D-2-hydroxyglutaric aciduria, 600721*
DAG1	*100%*	*100%*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538* *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818*
DARS1	*100%*	*100%*	*Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281*
DARS2	*100%*	*100%*	*Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105*
DBH	*100%*	*100%*	*Orthostatic hypotension 1, due to DBH deficiency, 223360*

DBT	*100%*	*100%*	*Maple syrup urine disease, type II, 248600*
DCAF17	*100%*	*100%*	*Woodhouse-Sakati syndrome, 241080*
DCC	*100%*	*100%*	*Mirror movements 1 and/or agenesis of the corpus callosum, 157600* *Esophageal carcinoma, somatic, 133239* *Colorectal cancer, somatic, 114500* *Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542*
DCDC2	*100%*	*100%*	*Nephronophthisis 19, 616217* *?Deafness, autosomal recessive 66, 610212* *Sclerosing cholangitis, neonatal, 617394*
DCHS1	*100%*	*100%*	*Mitral valve prolapse 2, 607829* *Van Maldergem syndrome 1, 601390*
DCLRE1C	*100%*	*100%*	*Severe combined immunodeficiency, Athabascan type, 602450* *Omenn syndrome, 603554*
DCPS	*100%*	*100%*	*Al-Raqad syndrome, 616459*
DDB2	*100%*	*100%*	*Xeroderma pigmentosum, group E, DDB-negative subtype, 278740*
DDC	*100%*	*100%*	*Aromatic L-amino acid decarboxylase deficiency, 608643*
DDHD1	*100%*	*100%*	*Spastic paraplegia 28, autosomal recessive, 609340*
DDHD2	*100%*	*100%*	*Spastic paraplegia 54, autosomal recessive, 615033*
DDOST	*100%*	*100%*	*Congenital disorder of glycosylation, type Ir, 614507*
DDR2	*100%*	*100%*	*Warburg-Cinotti syndrome, 618175* *Spondylometaepiphyseal dysplasia, short limb-hand type, 271665*
DDRGK1	*100%*	*100%*	*Spondyloepimetaphyseal dysplasia, Shohat type, 602557*
DDX11	*100%*	*100%*	*Warsaw breakage syndrome, 613398*
DDX59	*100%*	*100%*	*Orofaciodigital syndrome V, 174300*
DEAF1	*100%*	*100%*	*Vulto-van Silfout-de Vries syndrome, 615828* *Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171*
DEGS1	*100%*	*100%*	*Leukodystrophy, hypomyelinating, 18, 618404*
DENND5A	*100%*	*100%*	*Developmental and epileptic encephalopathy 49, 617281*
DES	*100%*	*100%*	*Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400* *Cardiomyopathy, dilated, 1I, 604765* *Myopathy, myofibrillar, 1, 601419*
DGKE	*100%*	*100%*	*Nephrotic syndrome, type 7, 615008*

DGUOK	*100%*	*100%*	*Portal hypertension, noncirrhotic, 1, 617068* *Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070* *Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880*
DHCR24	*100%*	*100%*	*Desmosterolosis, 602398*
DHCR7	*100%*	*100%*	*Smith-Lemli-Opitz syndrome, 270400*
DHDDS	*94%*	*94%*	*Developmental delay and seizures with or without movement abnormalities, 617836* *?Congenital disorder of glycosylation, type 1bb, 613861* *Retinitis pigmentosa 59, 613861*
DHFR	*100%*	*100%*	*Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839*
DHH	*100%*	*100%*	*46XY gonadal dysgenesis with minifascicular neuropathy, 607080* *46XY sex reversal 7, 233420*
DHODH	*100%*	*100%*	*Miller syndrome, 263750*
DHPS	*97%*	*93%*	*Neurodevelopmental disorder with seizures and speech and walking impairment, 618480*
DHTKD1	*100%*	*100%*	*?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025* *Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750*
DHX38	*100%*	*100%*	*Retinitis pigmentosa 84, 618220*
DIAPH1	*100%*	*100%*	*Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900* *Seizures, cortical blindness, microcephaly syndrome, 616632*
DIS3L2	*100%*	*100%*	*Perlman syndrome, 267000*
DLAT	*100%*	*100%*	*Pyruvate dehydrogenase E2 deficiency, 245348*
DLD	*100%*	*100%*	*Dihydrolipoamide dehydrogenase deficiency, 246900*
DLL3	*100%*	*100%*	*Spondylocostal dysostosis 1, autosomal recessive, 277300*
DLX5	*100%*	*100%*	*Split-hand/foot malformation 1, 183600* *?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600*
DMGDH	*100%*	*100%*	*Dimethylglycine dehydrogenase deficiency, 605850*
DMP1	*100%*	*100%*	*Hypophosphatemic rickets, AR, 241520*
DMXL2	*100%*	*100%*	*Developmental and epileptic encephalopathy 81, 618663* *?Deafness, autosomal dominant 71, 617605* *?Polyendocrine-polyneuropathy syndrome, 616113*
DNA2	*100%*	*100%*	*?Seckel syndrome 8, 615807* *Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156*
DNAAF1	*100%*	*100%*	*Ciliary dyskinesia, primary, 13, 613193*
LRRC6	*100%*	*100%*	*Ciliary dyskinesia, primary, 19, 614935*

DNAAF2	*100%*	*100%*	*Ciliary dyskinesia, primary, 10, 612518*
DNAAF3	*100%*	*100%*	*Ciliary dyskinesia, primary, 2, 606763*
DNAAF4	*100%*	*100%*	*Ciliary dyskinesia, primary, 25, 615482*
DNAAF5	*100%*	*100%*	*Ciliary dyskinesia, primary, 18, 614874*
DNAH1	*100%*	*100%*	*Spermatogenic failure 18, 617576* *?Ciliary dyskinesia, primary, 37, 617577*
DNAH11	*100%*	*100%*	*Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884*
DNAH5	*100%*	*100%*	*Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644*
DNAH9	*100%*	*100%*	*Ciliary dyskinesia, primary, 40, 618300*
DNAI1	*100%*	*100%*	*Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400*
DNAI2	*100%*	*100%*	*Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444*
DNAJB2	*100%*	*100%*	*Spinal muscular atrophy, distal, autosomal recessive, 5, 614881*
DNAJC12	*100%*	*100%*	*Hyperphenylalaninemia, mild, non-BH4-deficient, 617384*
DNAJC19	*100%*	*100%*	*3-methylglutaconic aciduria, type V, 610198*
DNAJC21	*100%*	*100%*	*Bone marrow failure syndrome 3, 617052*
DNAJC3	*100%*	*100%*	*Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192*
DNAJC6	*100%*	*100%*	*Parkinson disease 19a, juvenile-onset, 615528* *Parkinson disease 19b, early-onset, 615528*
DNAL1	*100%*	*100%*	*Ciliary dyskinesia, primary, 16, 614017*
DNASE1L3	*100%*	*100%*	*Systemic lupus erythematosus 16, 614420*
DNM1L	*100%*	*100%*	*Optic atrophy 5, 610708* *Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388*
DNM2	*100%*	*100%*	*Centronuclear myopathy 1, 160150* *Charcot-Marie-Tooth disease, axonal type 2M, 606482* *Charcot-Marie-Tooth disease, dominant intermediate B, 606482* *Lethal congenital contracture syndrome 5, 615368*
DNMBP	*100%*	*100%*	*Cataract 48, 618415*
DNMT3B	*100%*	*100%*	*Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860* *Facioscapulohumeral muscular dystrophy 4, digenic, 619478*
DOCK2	*100%*	*100%*	*Immunodeficiency 40, 616433*
DOCK3	*100%*	*100%*	*Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292*
DOCK6	*100%*	*100%*	*Adams-Oliver syndrome 2, 614219*
DOCK7	*100%*	*100%*	*Developmental and epileptic encephalopathy 23, 615859*
DOCK8	*100%*	*100%*	*Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700*

DOK7	*100%*	*100%*	*Fetal akinesia deformation sequence 3, 618389* *Myasthenic syndrome, congenital, 10, 254300*
DOLK	*100%*	*100%*	*Congenital disorder of glycosylation, type Im, 610768*
DONSON	*100%*	*100%*	*Microcephaly, short stature, and limb abnormalities, 617604* *Microcephaly-micromelia syndrome, 251230*
DPAGT1	*100%*	*100%*	*Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750* *Congenital disorder of glycosylation, type Ij, 608093*
DPH1	*100%*	*100%*	*Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901*
DPM1	*99%*	*97%*	*Congenital disorder of glycosylation, type Ie, 608799*
DPM2	*100%*	*100%*	*Congenital disorder of glycosylation, type Iu, 615042*
DPM3	*100%*	*100%*	*?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992* *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937*
DPY19L2	*100%*	*100%*	*Spermatogenic failure 9, 613958*
DPYD	*100%*	*100%*	*Dihydropyrimidine dehydrogenase deficiency, 274270* *5-fluorouracil toxicity, 274270*
DPYS	*100%*	*100%*	*Dihydropyrimidinuria, 222748*
DRAM2	*100%*	*100%*	*Cone-rod dystrophy 21, 616502*
DRC1	*100%*	*100%*	*Ciliary dyskinesia, primary, 21, 615294*
DSC2	*100%*	*100%*	*Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476* *Arrhythmogenic right ventricular dysplasia 11, 610476*
DSC3	*100%*	*100%*	*Hypotrichosis and recurrent skin vesicles, 613102*
DSE	*100%*	*100%*	*Ehlers-Danlos syndrome, musculocontractural type 2, 615539*
DSG4	*100%*	*100%*	*Hypotrichosis 6, 607903*
DSP	*100%*	*100%*	*Arrhythmogenic right ventricular dysplasia 8, 607450* *Skin fragility-woolly hair syndrome, 607655* *Epidermolysis bullosa, lethal acantholytic, 609638* *Keratosis palmoplantaris striata II, 612908* *Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821* *Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676*
DST	*100%*	*100%*	*Neuropathy, hereditary sensory and autonomic, type VI, 614653* *Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425*

DSTYK	*100%*	*100%*	*Congenital anomalies of kidney and urinary tract 1, 610805* *Spastic paraplegia 23, 270750*
DTNBP1	*100%*	*100%*	*Hermansky-Pudlak syndrome 7, 614076*
DTYMK	*100%*	*100%*	*Neurodegeneration, childhood-onset, with progressive microcephaly, 619847*
DUOX2	*100%*	*100%*	*Thyroid dyshormonogenesis 6, 607200*
DUOXA2	*100%*	*100%*	*Thyroid dyshormonogenesis 5, 274900*
DYM	*100%*	*100%*	*Smith-McCort dysplasia, 607326* *Dyggve-Melchior-Clausen disease, 223800*
DYNC1I2	*100%*	*100%*	*Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492*
DYNC2H1	*100%*	*99%*	*Short-rib thoracic dysplasia 3 with or without polydactyly, 613091*
WDR60	*100%*	*100%*	*Short-rib thoracic dysplasia 8 with or without polydactyly, 615503*
WDR34	*100%*	*100%*	*Short-rib thoracic dysplasia 11 with or without polydactyly, 615633*
DYNC2LI1	*100%*	*100%*	*Short-rib thoracic dysplasia 15 with polydactyly, 617088*
TCTEX1D2	*100%*	*100%*	*Short-rib thoracic dysplasia 17 with or without polydactyly, 617405*
DYSF	*100%*	*100%*	*Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601* *Miyoshi muscular dystrophy 1, 254130* *Myopathy, distal, with anterior tibial onset, 606768*
DZIP1L	*100%*	*100%*	*Polycystic kidney disease 5, 617610*
EARS2	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 12, 614924*
ECEL1	*100%*	*100%*	*Arthrogryposis, distal, type 5D, 615065*
ECHS1	*100%*	*100%*	*Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277*
ECM1	*100%*	*100%*	*Urbach-Wiethe disease, 247100*
EDAR	*100%*	*100%*	*Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490* *Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900*
EDARADD	*100%*	*100%*	*Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941* *Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940*
EDC3	*100%*	*100%*	*?Intellectual developmental disorder, autosomal recessive 50, 616460*
EDEM3	*100%*	*100%*	*Congenital disorder of glycosylation, type IIv, 619493*
EDN1	*100%*	*100%*	*Question mark ears, isolated, 612798* *Auriculocondylar syndrome 3, 615706*
EDN3	*100%*	*100%*	*Waardenburg syndrome, type 4B, 613265*
EDNRB	*100%*	*100%*	*?ABCD syndrome, 600501* *Waardenburg syndrome, type 4A, 277580*
EFEMP2	*100%*	*100%*	*Cutis laxa, autosomal recessive, type IB, 614437*

EFL1	*100%*	*100%*	*Shwachman-Diamond syndrome 2, 617941*
EGF	*100%*	*100%*	*?Hypomagnesemia 4, renal, 611718*
EGFR	*100%*	*100%*	*?Inflammatory skin and bowel disease, neonatal, 2, 616069* *Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980* *Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980*
EGR2	*100%*	*100%*	*Dejerine-Sottas disease, 145900* *Charcot-Marie-Tooth disease, type 1D, 607678* *Hypomyelinating neuropathy, congenital, 1, 605253*
EIF2AK3	*100%*	*100%*	*Wolcott-Rallison syndrome, 226980*
EIF2AK4	*100%*	*100%*	*Pulmonary venoocclusive disease 2, 234810*
EIF2B1	*100%*	*100%*	*Leukoencephalopathy with vanishing white matter, 603896*
EIF2B2	*100%*	*100%*	*Leukoencephalopathy with vanishing white matter, 603896* *Ovarioleukodystrophy, 603896*
EIF2B3	*100%*	*100%*	*Leukoencephalopathy with vanishing white matter, 603896*
EIF2B4	*100%*	*100%*	*Ovarioleukodystrophy, 603896* *Leukoencephalopathy with vanishing white matter, 603896*
EIF2B5	*100%*	*100%*	*Ovarioleukodystrophy, 603896* *Leukoencephalopathy with vanishing white matter, 603896*
EIF3F	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 67, 618295*
EIF4A3	*100%*	*100%*	*Robin sequence with cleft mandible and limb anomalies, 268305*
ELAC2	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 17, 615440*
ELMO2	*100%*	*100%*	*Vascular malformation, primary intraosseous, 606893*
ELOVL4	*100%*	*100%*	*Spinocerebellar ataxia 34, 133190* *Stargardt disease 3, 600110* *Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457*
ELP1	*100%*	*100%*	*Dysautonomia, familial, 223900*
ELP2	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 58, 617270*
EMC1	*100%*	*100%*	*Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875*
EMG1	*100%*	*100%*	*Bowen-Conradi syndrome, 211180*
EML1	*100%*	*100%*	*Band heterotopia, 600348*
EMP2	*100%*	*100%*	*Nephrotic syndrome, type 10, 615861*
ENAM	*100%*	*100%*	*Amelogenesis imperfecta, type IC, 204650* *Amelogenesis imperfecta, type IB, 104500*
ENO3	*100%*	*100%*	*Glycogen storage disease XIII, 612932*

ENPP1	*100%*	*100%*	*Hypophosphatemic rickets, autosomal recessive, 2, 613312* *Arterial calcification, generalized, of infancy, 1, 208000* *Cole disease, 615522*
ENTPD1	*100%*	*100%*	*Spastic paraplegia 64, autosomal recessive, 615683*
EOGT	*98%*	*94%*	*Adams-Oliver syndrome 4, 615297*
EPB41	*100%*	*100%*	*Elliptocytosis-1, 611804*
EPB42	*100%*	*100%*	*Spherocytosis, type 5, 612690*
EPCAM	*100%*	*100%*	*Diarrhea 5, with tufting enteropathy, congenital, 613217* *Lynch syndrome 8, 613244*
EPG5	*100%*	*100%*	*Vici syndrome, 242840*
EPHX1	*100%*	*100%*	*No OMIM disease ID*
EPM2A	*100%*	*100%*	*Epilepsy, progressive myoclonic 2A (Lafora), 254780*
EPO	*100%*	*100%*	*Erythrocytosis, familial, 5, 617907* *?Diamond-Blackfan anemia-like, 617911*
EPRS1	*100%*	*100%*	*Leukodystrophy, hypomyelinating, 15, 617951*
EPS8L2	*100%*	*100%*	*Deafness autosomal recessive 106, 617637*
ERAL1	*100%*	*100%*	*Perrault syndrome 6, 617565*
ERBB3	*100%*	*100%*	*?Lethal congenital contractural syndrome 2, 607598* *Visceral neuropathy, familial, 1, autosomal recessive, 243180*
ERCC1	*100%*	*100%*	*Cerebrooculofacioskeletal syndrome 4, 610758*
ERCC2	*100%*	*100%*	*Xeroderma pigmentosum, group D, 278730* *Trichothiodystrophy 1, photosensitive, 601675* *?Cerebrooculofacioskeletal syndrome 2, 610756*
ERCC3	*100%*	*100%*	*Trichothiodystrophy 2, photosensitive, 616390* *Xeroderma pigmentosum, group B, 610651*
ERCC4	*100%*	*100%*	*Xeroderma pigmentosum, type F/Cockayne syndrome, 278760* *XFE progeroid syndrome, 610965* *Xeroderma pigmentosum, group F, 278760* *Fanconi anemia, complementation group Q, 615272*
ERCC5	*100%*	*100%*	*Xeroderma pigmentosum, group G, 278780* *Cerebrooculofacioskeletal syndrome 3, 616570* *Xeroderma pigmentosum, group G/Cockayne syndrome, 278780*

ERCC6	*100%*	*100%*	*UV-sensitive syndrome 1, 600630* *Cerebrooculofacioskeletal syndrome 1, 214150* *?De Sanctis-Cacchione syndrome, 278800* *Cockayne syndrome, type B, 133540* *Premature ovarian failure 11, 616946*
ERCC6L2	*100%*	*100%*	*Bone marrow failure syndrome 2, 615715*
ERCC8	*100%*	*100%*	*UV-sensitive syndrome 2, 614621* *Cockayne syndrome, type A, 216400*
ERLIN1	*100%*	*100%*	*Spastic paraplegia 62, 615681*
ERLIN2	*100%*	*100%*	*Spastic paraplegia 18, autosomal recessive, 611225*
ESCO2	*100%*	*100%*	*Juberg-Hayward syndrome, 216100* *Roberts-SC phocomelia syndrome, 268300*
ESPN	*100%*	*100%*	*Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006* *Deafness, autosomal recessive 36, 609006* *?Usher syndrome, type 1M, 618632*
ESR1	*100%*	*100%*	*Breast cancer, somatic, 114480* *Estrogen resistance, 615363*
ESRRB	*100%*	*100%*	*Deafness, autosomal recessive 35, 608565*
ETFA	*100%*	*100%*	*Glutaric acidemia IIA, 231680*
ETFB	*100%*	*100%*	*Glutaric acidemia IIB, 231680*
ETFDH	*100%*	*100%*	*Glutaric acidemia IIC, 231680*
ETHE1	*100%*	*100%*	*Ethylmalonic encephalopathy, 602473*
EVC	*100%*	*100%*	*Ellis-van Creveld syndrome, 225500* *?Weyers acrofacial dysostosis, 193530*
EVC2	*100%*	*100%*	*Ellis-van Creveld syndrome, 225500* *Weyers acrofacial dysostosis, 193530*
EXOC6B	*100%*	*100%*	*Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395*
EXOSC2	*100%*	*100%*	*Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763*
EXOSC3	*100%*	*100%*	*Pontocerebellar hypoplasia, type 1B, 614678*
EXOSC8	*100%*	*100%*	*Pontocerebellar hypoplasia, type 1C, 616081*
EXOSC9	*100%*	*100%*	*Pontocerebellar hypoplasia, type 1D, 618065*
EXPH5	*100%*	*100%*	*Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028*
EXT2	*100%*	*100%*	*Seizures, scoliosis, and macrocephaly syndrome, 616682* *Exostoses, multiple, type 2, 133701*
EXTL3	*100%*	*100%*	*Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425*

EYS	*100%*	*100%*	*Retinitis pigmentosa 25, 602772*
F10	*100%*	*100%*	*Factor X deficiency, 227600*
F11	*100%*	*100%*	*Factor XI deficiency, autosomal dominant, 612416* *Factor XI deficiency, autosomal recessive, 612416*
F12	*100%*	*100%*	*Angioedema, hereditary, 3, 610618* *Factor XII deficiency, 234000*
F13A1	*100%*	*100%*	*Factor XIIIA deficiency, 613225*
F13B	*100%*	*99%*	*Factor XIIIB deficiency, 613235*
F2	*100%*	*100%*	*Hypoprothrombinemia, 613679* *Dysprothrombinemia, 613679* *Thrombophilia 1 due to thrombin defect, 188050*
F5	*100%*	*100%*	*Thrombophilia 2 due to activated protein C resistance, 188055* *Factor V deficiency, 227400*
F7	*100%*	*100%*	*Factor VII deficiency, 227500*
FA2H	*100%*	*100%*	*Spastic paraplegia 35, autosomal recessive, 612319*
FADD	*100%*	*100%*	*Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759*
FAH	*100%*	*100%*	*Tyrosinemia, type I, 276700*
FAM161A	*100%*	*100%*	*Retinitis pigmentosa 28, 606068*
FAM20A	*100%*	*100%*	*Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690*
FAM20C	*100%*	*100%*	*Raine syndrome, 259775*
FAN1	*100%*	*100%*	*Interstitial nephritis, karyomegalic, 614817*
FANCA	*100%*	*100%*	*Fanconi anemia, complementation group A, 227650*
FANCB	*100%*	*100%*	*Fanconi anemia, complementation group B, 300514*
FANCC	*100%*	*100%*	*Fanconi anemia, complementation group C, 227645*
FANCD2	*100%*	*100%*	*Fanconi anemia, complementation group D2, 227646*
FANCE	*100%*	*100%*	*Fanconi anemia, complementation group E, 600901*
FANCF	*100%*	*100%*	*Fanconi anemia, complementation group F, 603467*
FANCG	*100%*	*100%*	*Fanconi anemia, complementation group G, 614082*
FANCI	*100%*	*100%*	*Fanconi anemia, complementation group I, 609053*
FANCL	*100%*	*100%*	*Fanconi anemia, complementation group L, 614083*
FAR1	*100%*	*100%*	*Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154* *Cataracts, spastic paraparesis, and speech delay, 619338*
FARS2	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 14, 614946* *Spastic paraplegia 77, autosomal recessive, 617046*

FARSB	*100%*	*100%*	*Rajab interstitial lung disease with brain calcifications 1, 613658*
FASTKD2	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 44, 618855*
FAT4	*100%*	*100%*	*Van Maldergem syndrome 2, 615546* *Hennekam lymphangiectasia-lymphedema syndrome 2, 616006*
FBLN5	*92%*	*92%*	*Cutis laxa, autosomal recessive, type IA, 219100* *Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764* *Macular degeneration, age-related, 3, 608895* *Neuropathy, hereditary, with or without age-related macular degeneration, 608895* *?Cutis laxa, autosomal dominant 2, 614434*
FBP1	*100%*	*100%*	*Fructose-1,6-bisphosphatase deficiency, 229700*
FBXL3	*100%*	*100%*	*Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220*
FBXL4	*100%*	*100%*	*Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471*
FBXO31	*100%*	*100%*	*?Intellectual developmental disorder, autosomal recessive 45, 615979*
FBXO7	*100%*	*100%*	*Parkinson disease 15, autosomal recessive, 260300*
FCGR3A	*100%*	*100%*	*Immunodeficiency 20, 615707*
FCN3	*100%*	*100%*	*Immunodeficiency due to ficolin 3 deficiency, 613860*
FCSK	*100%*	*100%*	*Congenital disorder of glycosylation with defective fucosylation 2, 618324*
FDFT1	*100%*	*100%*	*Squalene synthase deficiency, 618156*
FDX2	*100%*	*100%*	*Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900*
FDXR	*100%*	*100%*	*Auditory neuropathy and optic atrophy, 617717*
FECH	*100%*	*100%*	*Protoporphyria, erythropoietic, 1, 177000*
FERMT1	*100%*	*100%*	*Kindler syndrome, 173650*
FERMT3	*100%*	*100%*	*Leukocyte adhesion deficiency, type III, 612840*
FEZF1	*100%*	*100%*	*Hypogonadotropic hypogonadism 22, with or without anosmia, 616030*
FGA	*100%*	*100%*	*Hypodysfibrinogenemia, congenital, 616004* *Dysfibrinogenemia, congenital, 616004* *Amyloidosis, familial visceral, 105200* *Afibrinogenemia, congenital, 202400*
FGB	*100%*	*100%*	*Hypofibrinogenemia, congenital, 202400* *Dysfibrinogenemia, congenital, 616004* *Afibrinogenemia, congenital, 202400*
FGD4	*100%*	*100%*	*Charcot-Marie-Tooth disease, type 4H, 609311*
FGF20	*100%*	*100%*	*?Renal hypodysplasia/aplasia 2, 615721*
FGF23	*100%*	*100%*	*Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993* *Hypophosphatemic rickets, autosomal dominant, 193100*

FGF3	*100%*	*100%*	*Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706*
FGG	*100%*	*100%*	*Dysfibrinogenemia, congenital, 616004* *Hypodysfibrinogenemia, 616004* *Hypofibrinogenemia, congenital, 202400* *Afibrinogenemia, congenital, 202400*
FH	*100%*	*100%*	*Leiomyomatosis and renal cell cancer, 150800* *Fumarase deficiency, 606812*
FIBP	*100%*	*100%*	*Thauvin-Robinet-Faivre syndrome, 617107*
FIG4	*100%*	*100%*	*Yunis-Varon syndrome, 216340* *?Polymicrogyria, bilateral temporooccipital, 612691* *Amyotrophic lateral sclerosis 11, 612577* *Charcot-Marie-Tooth disease, type 4J, 611228*
FITM2	*100%*	*100%*	*Siddiqi syndrome, 618635*
FKBP10	*100%*	*100%*	*Osteogenesis imperfecta, type XI, 610968* *Bruck syndrome 1, 259450*
FKBP14	*100%*	*100%*	*Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557*
FKRP	*100%*	*100%*	*Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612* *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153*
FKTN	*100%*	*100%*	*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800* *Cardiomyopathy, dilated, 1X, 611615* *Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152*
FLAD1	*100%*	*100%*	*Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100*
FLNB	*100%*	*100%*	*Larsen syndrome, 150250* *Atelosteogenesis, type I, 108720* *Atelosteogenesis, type III, 108721* *Spondylocarpotarsal synostosis syndrome, 272460* *Boomerang dysplasia, 112310*
FLVCR1	*100%*	*100%*	*Ataxia, posterior column, with retinitis pigmentosa, 609033*
FLVCR2	*100%*	*100%*	*Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790*

FMN2	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 47, 616193*
FMO3	*100%*	*100%*	*Trimethylaminuria, 602079*
FOLR1	*100%*	*100%*	*Neurodegeneration due to cerebral folate transport deficiency, 613068*
FOXE1	*100%*	*100%*	*Bamforth-Lazarus syndrome, 241850*
FOXE3	*100%*	*99%*	*Anterior segment dysgenesis 2, multiple subtypes, 610256* *Cataract 34, multiple types, 612968*
FOXI1	*100%*	*100%*	*Enlarged vestibular aqueduct, 600791*
FOXN1	*100%*	*100%*	*T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806* *T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705*
FOXRED1	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 19, 618241*
FRAS1	*100%*	*100%*	*Fraser syndrome 1, 219000*
FREM1	*100%*	*100%*	*Manitoba oculotrichoanal syndrome, 248450* *Bifid nose with or without anorectal and renal anomalies, 608980* *Trigonocephaly 2, 614485*
FREM2	*100%*	*100%*	*Fraser syndrome 2, 617666* *Cryptophthalmos, unilateral or bilateral, isolated, 123570*
FRRS1L	*100%*	*100%*	*Developmental and epileptic encephalopathy 37, 616981*
FSHB	*99%*	*98%*	*Hypogonadotropic hypogonadism 24 without anosmia, 229070*
FSHR	*100%*	*100%*	*Ovarian response to FSH stimulation, 276400* *Ovarian hyperstimulation syndrome, 608115* *Ovarian dysgenesis 1, 233300*
FTCD	*100%*	*100%*	*Glutamate formiminotransferase deficiency, 229100*
FTO	*95%*	*95%*	*Growth retardation, developmental delay, facial dysmorphism, 612938*
FUCA1	*100%*	*100%*	*Fucosidosis, 230000*
FUT8	*100%*	*100%*	*Congenital disorder of glycosylation with defective fucosylation 1, 618005*
FXN	*100%*	*100%*	*Friedreich ataxia with retained reflexes, 229300* *Friedreich ataxia, 229300*
FYCO1	*100%*	*100%*	*Cataract 18, autosomal recessive, 610019*
FZD6	*100%*	*100%*	*Nail disorder, nonsyndromic congenital, 1, 161050*
G6PC	*100%*	*100%*	*Glycogen storage disease Ia, 232200*
G6PC3	*100%*	*100%*	*Dursun syndrome, 612541* *Neutropenia, severe congenital 4, autosomal recessive, 612541*
GAA	*100%*	*100%*	*Glycogen storage disease II, 232300*
GAD1	*100%*	*100%*	*Developmental and epileptic encephalopathy 89, 619124*
GALC	*100%*	*100%*	*Krabbe disease, 245200*

GALE	*100%*	*100%*	*Galactose epimerase deficiency, 230350*
GALK1	*100%*	*100%*	*Galactokinase deficiency with cataracts, 230200*
GALNS	*100%*	*100%*	*Mucopolysaccharidosis IVA, 253000*
GALNT2	*100%*	*100%*	*Congenital disorder of glycosylation, type IIt, 618885*
GALNT3	*100%*	*100%*	*Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900*
GALT	*100%*	*100%*	*Galactosemia, 230400*
GAMT	*100%*	*100%*	*Cerebral creatine deficiency syndrome 2, 612736*
GAN	*100%*	*100%*	*Giant axonal neuropathy-1, 256850*
GAS8	*100%*	*100%*	*Ciliary dyskinesia, primary, 33, 616726*
GATM	*100%*	*100%*	*Cerebral creatine deficiency syndrome 3, 612718* *Fanconi renotubular syndrome 1, 134600*
GBA	*100%*	*100%*	*Gaucher disease, type II, 230900* *Gaucher disease, type IIIC, 231005* *Gaucher disease, type III, 231000* *Gaucher disease, type I, 230800* *Gaucher disease, perinatal lethal, 608013*
GBA2	*100%*	*100%*	*Spastic paraplegia 46, autosomal recessive, 614409*
GBE1	*100%*	*100%*	*Glycogen storage disease IV, 232500* *Polyglucosan body disease, adult form, 263570*
GCDH	*100%*	*100%*	*Glutaricaciduria, type I, 231670*
GCH1	*100%*	*100%*	*Dystonia, DOPA-responsive, 128230* *Hyperphenylalaninemia, BH4-deficient, B, 233910*
GCK	*100%*	*100%*	*MODY, type II, 125851* *Diabetes mellitus, permanent neonatal 1, 606176* *Hyperinsulinemic hypoglycemia, familial, 3, 602485* *Diabetes mellitus, noninsulin-dependent, late onset, 125853*
GCLC	*100%*	*100%*	*Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450*
GCNT2	*100%*	*100%*	*Adult i phenotype without cataract, 110800* *Cataract 13 with adult i phenotype, 116700*
GCSH	*100%*	*100%*	*?Glycine encephalopathy, 605899*
GDAP1	*100%*	*100%*	*Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706* *Charcot-Marie-Tooth disease, recessive intermediate, A, 608340* *Charcot-Marie-Tooth disease, axonal, type 2K, 607831* *Charcot-Marie-Tooth disease, type 4A, 214400*
GDAP2	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive 27, 618369*

GDF1	*100%*	*100%*	*Congenital heart defects, multiple types, 6, 613854* *Right atrial isomerism (Ivemark), 208530*
GDF5	*100%*	*100%*	*Acromesomelic dysplasia 2A, 200700* *Acromesomelic dysplasia 2B, 228900* *Multiple synostoses syndrome 2, 610017* *Symphalangism, proximal, 1B, 615298* *Brachydactyly, type A2, 112600* *?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250* *Brachydactyly, type C, 113100* *Brachydactyly, type A1, C, 615072*
GEMIN4	*100%*	*100%*	*Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913*
GFER	*100%*	*100%*	*Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076*
GFM1	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 1, 609060*
GFM2	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 39, 618397*
GFPT1	*100%*	*100%*	*Myasthenia, congenital, 12, with tubular aggregates, 610542*
GGCX	*100%*	*100%*	*Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450* *Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842*
GGT1	*100%*	*100%*	*?Glutathioninuria, 231950*
GH1	*100%*	*100%*	*Kowarski syndrome, 262650* *Growth hormone deficiency, isolated, type II, 173100* *Growth hormone deficiency, isolated, type IB, 612781* *Growth hormone deficiency, isolated, type IA, 262400*
GHR	*100%*	*100%*	*Laron dwarfism, 262500* *Increased responsiveness to growth hormone, 604271* *Growth hormone insensitivity, partial, 604271*
GHRHR	*100%*	*100%*	*Growth hormone deficiency, isolated, type IV, 618157*
GHSR	*100%*	*100%*	*Growth hormone deficiency, isolated partial, 615925*
GINS1	*100%*	*100%*	*Immunodeficiency 55, 617827*
GIPC3	*100%*	*100%*	*Deafness, autosomal recessive 15, 601869*

GJA1	*100%*	*100%*	*Erythrokeratodermia variabilis et progressiva 3, 617525* *Craniometaphyseal dysplasia, autosomal recessive, 218400* *Oculodentodigital dysplasia, 164200* *Hypoplastic left heart syndrome 1, 241550* *Palmoplantar keratoderma with congenital alopecia, 104100* *Syndactyly, type III, 186100* *Oculodentodigital dysplasia, autosomal recessive, 257850* *Atrioventricular septal defect 3, 600309*
GJB2	*100%*	*100%*	*Keratoderma, palmoplantar, with deafness, 148350* *Deafness, autosomal recessive 1A, 220290* *Deafness, autosomal dominant 3A, 601544* *Hystrix-like ichthyosis with deafness, 602540* *Bart-Pumphrey syndrome, 149200* *Keratitis-ichthyosis-deafness syndrome, 148210* *Vohwinkel syndrome, 124500*
GJB6	*100%*	*100%*	*Ectodermal dysplasia 2, Clouston type, 129500* *Deafness, autosomal dominant 3B, 612643* *Deafness, autosomal recessive 1B, 612645* *Deafness, digenic GJB2/GJB6, 220290*
GJC2	*100%*	*99%*	*Lymphatic malformation 3, 613480* *?Spastic paraplegia 44, autosomal recessive, 613206* *Leukodystrophy, hypomyelinating, 2, 608804*
GLB1	*100%*	*100%*	*GM1-gangliosidosis, type I, 230500* *GM1-gangliosidosis, type III, 230650* *Mucopolysaccharidosis type IVB (Morquio), 253010* *GM1-gangliosidosis, type II, 230600*
GLDC	*100%*	*100%*	*Glycine encephalopathy, 605899*
GLDN	*100%*	*100%*	*Lethal congenital contracture syndrome 11, 617194*
GLE1	*100%*	*100%*	*Lethal congenital contracture syndrome 1, 253310* *Congenital arthrogryposis with anterior horn cell disease, 611890*
GLIS2	*100%*	*100%*	*Nephronophthisis 7, 611498*
GLIS3	*100%*	*100%*	*Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199*
GLRA1	*100%*	*100%*	*Hyperekplexia 1, 149400*
GLRB	*100%*	*100%*	*Hyperekplexia 2, 614619*

GLRX5	*100%*	*100%*	*Anemia, sideroblastic, 3, pyridoxine-refractory, 616860* *Spasticity, childhood-onset, with hyperglycinemia, 616859*
GLS	*100%*	*100%*	*Global developmental delay, progressive ataxia, and elevated glutamine, 618412* *?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339* *Developmental and epileptic encephalopathy 71, 618328*
GLUL	*100%*	*100%*	*Glutamine deficiency, congenital, 610015*
GLYCTK	*100%*	*100%*	*D-glyceric aciduria, 220120*
GM2A	*100%*	*100%*	*GM2-gangliosidosis, AB variant, 272750*
GMPPA	*100%*	*100%*	*Alacrima, achalasia, and impaired intellectual development syndrome, 615510*
GMPPB	*100%*	*100%*	*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352* *Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350*
GNAT2	*100%*	*100%*	*Achromatopsia 4, 613856*
GNB3	*100%*	*100%*	*Night blindness, congenital stationary, type 1H, 617024*
GNB5	*100%*	*100%*	*Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182* *Intellectual developmental disorder with cardiac arrhythmia, 617173*
GNE	*100%*	*100%*	*Sialuria, 269921* *Nonaka myopathy, 605820*
GNMT	*100%*	*100%*	*Glycine N-methyltransferase deficiency, 606664*
GNPAT	*100%*	*100%*	*Rhizomelic chondrodysplasia punctata, type 2, 222765*
GNPTAB	*100%*	*100%*	*Mucolipidosis III alpha/beta, 252600* *Mucolipidosis II alpha/beta, 252500*
GNPTG	*100%*	*100%*	*Mucolipidosis III gamma, 252605*
GNRHR	*100%*	*100%*	*Hypogonadotropic hypogonadism 7 without anosmia, 146110*
GNS	*100%*	*100%*	*Mucopolysaccharidosis type IIID, 252940*
GORAB	*100%*	*100%*	*Geroderma osteodysplasticum, 231070*
GOSR2	*100%*	*100%*	*Epilepsy, progressive myoclonic 6, 614018*
GP1BA	*100%*	*100%*	*Bernard-Soulier syndrome, type A1 (recessive), 231200* *Bernard-Soulier syndrome, type A2 (dominant), 153670* *von Willebrand disease, platelet-type, 177820*
GP1BB	*100%*	*100%*	*Giant platelet disorder, isolated, 231200* *Bernard-Soulier syndrome, type B, 231200*

GP6	*99%*	*96%*	*Bleeding disorder, platelet-type, 11, 614201*
GP9	*100%*	*100%*	*Bernard-Soulier syndrome, type C, 231200*
GPAA1	*100%*	*100%*	*Glycosylphosphatidylinositol biosynthesis defect 15, 617810*
GPC6	*100%*	*100%*	*Omodysplasia 1, 258315*
GPD1	*100%*	*100%*	*Hypertriglyceridemia, transient infantile, 614480*
GPHN	*100%*	*100%*	*Molybdenum cofactor deficiency C, 615501*
GPI	*100%*	*100%*	*Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470*
GPIHBP1	*100%*	*100%*	*Hyperlipoproteinemia, type 1D, 615947*
GPNMB	*95%*	*95%*	*Amyloidosis, primary localized cutaneous, 3, 617920*
GPR179	*100%*	*100%*	*Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565*
GPSM2	*100%*	*100%*	*Chudley-McCullough syndrome, 604213*
GPT2	*100%*	*100%*	*Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281*
GPX4	*100%*	*100%*	*Spondylometaphyseal dysplasia, Sedaghatian type, 250220*
GRAP	*100%*	*100%*	*Deafness, autosomal recessive 114, 618456*
GRHPR	*100%*	*100%*	*Hyperoxaluria, primary, type II, 260000*
GRID2	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive 18, 616204*
GRIK2	*96%*	*96%*	*Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580* *Intellectual developmental disorder, autosomal recessive 6, 611092*
GRIN1	*100%*	*100%*	*Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820* *Developmental and epileptic encephalopathy 101, 619814* *Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254*
GRIP1	*100%*	*100%*	*Fraser syndrome 3, 617667*
GRK1	*100%*	*100%*	*Oguchi disease-2, 613411*
GRM1	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive 13, 614831* *Spinocerebellar ataxia 44, 617691*
GRM6	*100%*	*100%*	*Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270*
GRN	*100%*	*100%*	*Aphasia, primary progressive, 607485* *Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485* *Ceroid lipofuscinosis, neuronal, 11, 614706*
GRXCR1	*100%*	*99%*	*Deafness, autosomal recessive 25, 613285*
GSC	*100%*	*100%*	*Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471*

GSS	*100%*	*100%*	*Hemolytic anemia due to glutathione synthetase deficiency, 231900* *Glutathione synthetase deficiency, 266130*
GSX2	*100%*	*100%*	*Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646*
GTF2E2	*100%*	*100%*	*Trichothiodystrophy 6, nonphotosensitive, 616943*
GTF2H5	*70%*	*70%*	*Trichothiodystrophy 3, photosensitive, 616395*
GTPBP2	*100%*	*100%*	*Jaberi-Elahi syndrome, 617988*
GTPBP3	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 23, 616198*
GUCY1A1	*100%*	*100%*	*Moyamoya 6 with achalasia, 615750*
GUCY2C	*100%*	*100%*	*Diarrhea 6, 614616* *Meconium ileus, 614665*
GUCY2D	*100%*	*100%*	*Cone-rod dystrophy 6, 601777* *?Choroidal dystrophy, central areolar 1, 215500* *Leber congenital amaurosis 1, 204000* *Night blindness, congenital stationary, type 1I, 618555*
GUSB	*100%*	*100%*	*Mucopolysaccharidosis VII, 253220*
GYG1	*100%*	*100%*	*?Glycogen storage disease XV, 613507* *Polyglucosan body myopathy 2, 616199*
GYS1	*100%*	*100%*	*Glycogen storage disease 0, muscle, 611556*
GYS2	*100%*	*100%*	*Glycogen storage disease 0, liver, 240600*
GZF1	*100%*	*100%*	*Joint laxity, short stature, and myopia, 617662*
H6PD	*100%*	*100%*	*Cortisone reductase deficiency 1, 604931*
HAAO	*100%*	*100%*	*Vertebral, cardiac, renal, and limb defects syndrome 1, 617660*
HACE1	*100%*	*100%*	*Spastic paraplegia and psychomotor retardation with or without seizures, 616756*
HADH	*100%*	*100%*	*Hyperinsulinemic hypoglycemia, familial, 4, 609975* *3-hydroxyacyl-CoA dehydrogenase deficiency, 231530*
HADHA	*100%*	*100%*	*HELLP syndrome, maternal, of pregnancy, 609016* *Mitochondrial trifunctional protein deficiency, 609015* *LCHAD deficiency, 609016* *Fatty liver, acute, of pregnancy, 609016*
HADHB	*100%*	*100%*	*Trifunctional protein deficiency, 609015*
HAMP	*100%*	*100%*	*Hemochromatosis, type 2B, 613313*
HARS1	*100%*	*100%*	*Charcot-Marie-Tooth disease, axonal, type 2W, 616625* *Usher syndrome type 3B, 614504*
HARS2	*100%*	*100%*	*Perrault syndrome 2, 614926*
HAVCR2	*100%*	*100%*	*T-cell lymphoma, subcutaneous panniculitis-like, 618398*

HAX1	*100%*	*100%*	*Neutropenia, severe congenital 3, autosomal recessive, 610738*
HBB	*100%*	*100%*	*Methemoglobinemia, beta type, 617971* *Thalassemia-beta, dominant inclusion-body, 603902* *Sickle cell anemia, 603903* *Thalassemia, beta, 613985* *Delta-beta thalassemia, 141749* *Hereditary persistence of fetal hemoglobin, 141749* *Heinz body anemia, 140700* *Erythrocytosis 6, 617980*
HELLS	*100%*	*100%*	*Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911*
HEPACAM	*100%*	*100%*	*Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925* *Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926*
HERC1	*100%*	*100%*	*Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011*
HERC2	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 38, 615516*
HES7	*100%*	*100%*	*Spondylocostal dysostosis 4, autosomal recessive, 613686*
HESX1	*100%*	*100%*	*Pituitary hormone deficiency, combined, 5, 182230* *Septooptic dysplasia, 182230* *Growth hormone deficiency with pituitary anomalies, 182230*
HEXA	*100%*	*100%*	*GM2-gangliosidosis, several forms, 272800* *Tay-Sachs disease, 272800*
HEXB	*100%*	*100%*	*Sandhoff disease, infantile, juvenile, and adult forms, 268800*
HFM1	*100%*	*100%*	*Premature ovarian failure 9, 615724*
HGD	*100%*	*100%*	*Alkaptonuria, 203500*
HGF	*100%*	*100%*	*Deafness, autosomal recessive 39, 608265*
HGSNAT	*92%*	*92%*	*Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930* *Retinitis pigmentosa 73, 616544*
HIBCH	*100%*	*100%*	*3-hydroxyisobutryl-CoA hydrolase deficiency, 250620*
HIKESHI	*100%*	*100%*	*Leukodystrophy, hypomyelinating, 13, 616881*
HINT1	*100%*	*100%*	*Neuromyotonia and axonal neuropathy, autosomal recessive, 137200*
HJV	*100%*	*100%*	*Hemochromatosis, type 2A, 602390*
HK1	*100%*	*100%*	*Retinitis pigmentosa 79, 617460* *Neuropathy, hereditary motor and sensory, Russe type, 605285* *Neurodevelopmental disorder with visual defects and brain anomalies, 618547* *Hemolytic anemia due to hexokinase deficiency, 235700*

HLCS	*100%*	*100%*	*Holocarboxylase synthetase deficiency, 253270*
HMGCL	*100%*	*100%*	*HMG-CoA lyase deficiency, 246450*
HMGCS2	*100%*	*100%*	*HMG-CoA synthase-2 deficiency, 605911*
HMOX1	*100%*	*100%*	*Heme oxygenase-1 deficiency, 614034*
HMX1	*100%*	*100%*	*Oculoauricular syndrome, 612109*
HNMT	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 51, 616739*
HOGA1	*100%*	*100%*	*Hyperoxaluria, primary, type III, 613616*
HOXA1	*100%*	*100%*	*Bosley-Salih-Alorainy syndrome, 601536* *Athabaskan brainstem dysgenesis syndrome, 601536*
HOXA2	*100%*	*100%*	*Microtia with or without hearing impairment (AD), 612290* *?Microtia, hearing impairment, and cleft palate (AR), 612290*
HOXB1	*100%*	*100%*	*Facial paresis, hereditary congenital, 3, 614744*
HOXC13	*100%*	*100%*	*Ectodermal dysplasia 9, hair/nail type, 614931*
HPCA	*100%*	*100%*	*Dystonia 2, torsion, autosomal recessive, 224500*
HPD	*100%*	*100%*	*Hawkinsinuria, 140350* *Tyrosinemia, type III, 276710*
HPDL	*100%*	*100%*	*Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026* *Spastic paraplegia 83, autosomal recessive, 619027*
HPGD	*100%*	*100%*	*?Digital clubbing, isolated congenital, 119900* *Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100* *Cranioosteoarthropathy, 259100*
HPS1	*100%*	*100%*	*Hermansky-Pudlak syndrome 1, 203300*
HPS3	*100%*	*100%*	*Hermansky-Pudlak syndrome 3, 614072*
HPS4	*100%*	*100%*	*Hermansky-Pudlak syndrome 4, 614073*
HPS5	*100%*	*100%*	*Hermansky-Pudlak syndrome 5, 614074*
HPS6	*100%*	*100%*	*Hermansky-Pudlak syndrome 6, 614075*
HPSE2	*100%*	*100%*	*Urofacial syndrome 1, 236730*
HR	*100%*	*100%*	*Atrichia with papular lesions, 209500* *Alopecia universalis, 203655*
HSD11B2	*100%*	*100%*	*Apparent mineralocorticoid excess, 218030*
HSD17B3	*100%*	*100%*	*Pseudohermaphroditism, male, with gynecomastia, 264300*
HSD17B4	*97%*	*97%*	*D-bifunctional protein deficiency, 261515* *Perrault syndrome 1, 233400*
HSD3B2	*100%*	*99%*	*Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810*

HSD3B7	*100%*	*100%*	*Bile acid synthesis defect, congenital, 1, 607765*
HSPA9	*100%*	*100%*	*Even-plus syndrome, 616854* *Anemia, sideroblastic, 4, 182170*
HSPD1	*100%*	*100%*	*Spastic paraplegia 13, autosomal dominant, 605280* *Leukodystrophy, hypomyelinating, 4, 612233*
HSPG2	*100%*	*100%*	*Dyssegmental dysplasia, Silverman-Handmaker type, 224410* *Schwartz-Jampel syndrome, type 1, 255800*
HTRA1	*100%*	*100%*	*CARASIL syndrome, 600142* *Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779*
HTRA2	*100%*	*100%*	*3-methylglutaconic aciduria, type VIII, 617248*
HYAL1	*100%*	*100%*	*Mucopolysaccharidosis type IX, 601492*
FAM126A	*100%*	*100%*	*Leukodystrophy, hypomyelinating, 5, 610532*
HYDIN	*100%*	*100%*	*Ciliary dyskinesia, primary, 5, 608647*
HYLS1	*100%*	*100%*	*Hydrolethalus syndrome, 236680*
IARS1	*100%*	*100%*	*Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093*
IARS2	*100%*	*100%*	*Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007*
IBA57	*100%*	*100%*	*Multiple mitochondrial dysfunctions syndrome 3, 615330* *?Spastic paraplegia 74, autosomal recessive, 616451*
ICOS	*100%*	*100%*	*Immunodeficiency, common variable, 1, 607594*
IDH3B	*100%*	*100%*	*Retinitis pigmentosa 46, 612572*
IDUA	*100%*	*100%*	*Mucopolysaccharidosis Is, 607016* *Mucopolysaccharidosis Ih/s, 607015* *Mucopolysaccharidosis Ih, 607014*
IER3IP1	*100%*	*100%*	*Microcephaly, epilepsy, and diabetes syndrome, 614231*
IFNAR2	*100%*	*100%*	*Immunodeficiency 45, 616669*
IFNGR1	*100%*	*100%*	*Immunodeficiency 27A, mycobacteriosis, AR, 209950* *Immunodeficiency 27B, mycobacteriosis, AD, 615978*
IFNGR2	*100%*	*100%*	*Immunodeficiency 28, mycobacteriosis, 614889*
IFT122	*100%*	*100%*	*Cranioectodermal dysplasia 1, 218330*
IFT140	*100%*	*100%*	*Short-rib thoracic dysplasia 9 with or without polydactyly, 266920* *Retinitis pigmentosa 80, 617781*
IFT172	*100%*	*100%*	*Retinitis pigmentosa 71, 616394* *Bardet-Biedl syndrome 20, 619471* *Short-rib thoracic dysplasia 10 with or without polydactyly, 615630*

IFT27	*100%*	*100%*	*Bardet-Biedl syndrome 19, 615996*
IFT43	*100%*	*100%*	*?Cranioectodermal dysplasia 3, 614099* *?Retinitis pigmentosa 81, 617871* *Short-rib thoracic dysplasia 18 with polydactyly, 617866*
IFT52	*100%*	*100%*	*Short-rib thoracic dysplasia 16 with or without polydactyly, 617102*
IFT57	*100%*	*100%*	*?Orofaciodigital syndrome XVIII, 617927*
IFT74	*100%*	*100%*	*Bardet-Biedl syndrome 22, 617119* *Spermatogenic failure 58, 619585* *Joubert syndrome 40, 619582*
IFT80	*100%*	*100%*	*Short-rib thoracic dysplasia 2 with or without polydactyly, 611263*
IFT81	*95%*	*95%*	*Short-rib thoracic dysplasia 19 with or without polydactyly, 617895*
IGF1	*100%*	*100%*	*Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747*
IGF1R	*100%*	*100%*	*Insulin-like growth factor I, resistance to, 270450*
IGFALS	*100%*	*100%*	*Acid-labile subunit, deficiency of, 615961*
IGFBP7	*100%*	*100%*	*Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224*
IGHM	*100%*	*100%*	*Agammaglobulinemia 1, 601495*
IGHMBP2	*100%*	*100%*	*Neuronopathy, distal hereditary motor, type VI, 604320* *Charcot-Marie-Tooth disease, axonal, type 2S, 616155*
IGKC	*100%*	*100%*	*Kappa light chain deficiency, 614102*
IGLL1	*100%*	*100%*	*Agammaglobulinemia 2, 613500*
IHH	*100%*	*100%*	*Acrocapitofemoral dysplasia, 607778* *Brachydactyly, type A1, 112500*
IKBKB	*100%*	*100%*	*Immunodeficiency 15B, 615592* *Immunodeficiency 15A, 618204*
IL10RA	*100%*	*100%*	*Inflammatory bowel disease 28, early onset, autosomal recessive, 613148*
IL10RB	*100%*	*100%*	*Inflammatory bowel disease 25, early onset, autosomal recessive, 612567*
IL11RA	*100%*	*100%*	*Craniosynostosis and dental anomalies, 614188*
IL12B	*100%*	*100%*	*Immunodeficiency 29, mycobacteriosis, 614890*
IL12RB1	*94%*	*94%*	*Immunodeficiency 30, 614891*
IL17RA	*100%*	*100%*	*Immunodeficiency 51, 613953*
IL17RC	*100%*	*100%*	*Candidiasis, familial, 9, 616445*
IL1RN	*100%*	*100%*	*Interleukin 1 receptor antagonist deficiency, 612852*
IL21R	*100%*	*100%*	*Immunodeficiency 56, 615207*
IL2RA	*100%*	*100%*	*Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367*

IL2RB	*100%*	*100%*	*Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495*
IL36RN	*100%*	*100%*	*Psoriasis 14, pustular, 614204*
IL6ST	*100%*	*100%*	*Stuve-Wiedemann syndrome 2, 619751* *Hyper-IgE recurrent infection syndrome 4A, autosomal dominant, 619752* *?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750* *Hyper-IgE recurrent infection syndrome 4B, autosomal recessive, 618523*
IL7R	*100%*	*100%*	*Immunodeficiency 104, severe combined, 608971*
ILDR1	*100%*	*100%*	*Deafness, autosomal recessive 42, 609646*
IMPA1	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 59, 617323*
IMPG2	*100%*	*100%*	*Retinitis pigmentosa 56, 613581* *Macular dystrophy, vitelliform, 5, 616152*
INPP5E	*100%*	*100%*	*Joubert syndrome 1, 213300* *Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156*
INPP5K	*100%*	*100%*	*Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404*
INPPL1	*100%*	*100%*	*Opsismodysplasia, 258480*
INSR	*100%*	*100%*	*Rabson-Mendenhall syndrome, 262190* *Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549* *Donohue syndrome, 246200* *Hyperinsulinemic hypoglycemia, familial, 5, 609968*
INTS1	*100%*	*100%*	*Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571*
INTS8	*100%*	*100%*	*?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572*
INTU	*100%*	*100%*	*?Orofaciodigital syndrome XVII, 617926* *?Short-rib thoracic dysplasia 20 with polydactyly, 617925*
INVS	*100%*	*100%*	*Nephronophthisis 2, infantile, 602088*
IPO8	*100%*	*100%*	*VISS syndrome, 619472*
IQCB1	*100%*	*100%*	*Senior-Loken syndrome 5, 609254*
IQSEC1	*100%*	*100%*	*Intellectual developmental disorder with short stature and behavioral abnormalities, 618687*
IRAK4	*100%*	*100%*	*Immunodeficiency 67, 607676*
IREB2	*100%*	*100%*	*Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451*
IRF7	*100%*	*100%*	*?Immunodeficiency 39, 616345*
IRF8	*100%*	*100%*	*Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893* *Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990*
IRF9	*100%*	*100%*	*Immunodeficiency 65, susceptibility to viral infections, 618648*
IRX5	*100%*	*100%*	*Hamamy syndrome, 611174*
ISCA1	*92%*	*92%*	*Multiple mitochondrial dysfunctions syndrome 5, 617613*

ISCA2	*100%*	*100%*	*Multiple mitochondrial dysfunctions syndrome 4, 616370*
ISCU	*100%*	*100%*	*Myopathy with lactic acidosis, hereditary, 255125*
ISG15	*100%*	*100%*	*Immunodeficiency 38, 616126*
ITCH	*96%*	*96%*	*Autoimmune disease, multisystem, with facial dysmorphism, 613385*
ITGA2B	*100%*	*100%*	*Glanzmann thrombasthenia 1, 273800* *Bleeding disorder, platelet-type, 16, autosomal dominant, 187800* *Thrombocytopenia, neonatal alloimmune, BAK antigen related,*
ITGA3	*100%*	*100%*	*Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748*
ITGA6	*100%*	*100%*	*Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817*
ITGA7	*100%*	*100%*	*Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204*
ITGA8	*100%*	*100%*	*Renal hypodysplasia/aplasia 1, 191830*
ITGB2	*100%*	*100%*	*Leukocyte adhesion deficiency, 116920*
ITGB3	*100%*	*100%*	*Bleeding disorder, platelet-type, 24, autosomal dominant, 619271* *Glanzmann thrombasthenia 2, 619267* *Thrombocytopenia, neonatal alloimmune,* *Purpura, posttransfusion,*
ITGB4	*100%*	*100%*	*Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730* *Epidermolysis bullosa, junctional 5A, intermediate, 619816*
ITGB6	*100%*	*100%*	*Amelogenesis imperfecta, type IH, 616221*
ITK	*100%*	*100%*	*Lymphoproliferative syndrome 1, 613011*
ITPA	*100%*	*100%*	*Developmental and epileptic encephalopathy 35, 616647*
ITPR1	*100%*	*100%*	*Gillespie syndrome, 206700* *Spinocerebellar ataxia 29, congenital nonprogressive, 117360* *Spinocerebellar ataxia 15, 606658*
IVD	*100%*	*100%*	*Isovaleric acidemia, 243500*
IYD	*100%*	*100%*	*Thyroid dyshormonogenesis 4, 274800*
JAGN1	*100%*	*100%*	*Neutropenia, severe congenital, 6, autosomal recessive, 616022*
JAK3	*100%*	*100%*	*SCID, autosomal recessive, T-negative/B-positive type, 600802*
JAM2	*92%*	*92%*	*Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824*
JAM3	*100%*	*100%*	*Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730*
JUP	*100%*	*100%*	*Naxos disease, 601214* *?Arrhythmogenic right ventricular dysplasia 12, 611528*
KALRN	*100%*	*100%*	*No OMIM disease ID*
KANK2	*100%*	*100%*	*Nephrotic syndrome, type 16, 617783* *Palmoplantar keratoderma and woolly hair, 616099*

KARS1	*100%*	*100%*	*Deafness, autosomal recessive 89, 613916* *Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147* *?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641* *Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196*
KATNB1	*100%*	*100%*	*Lissencephaly 6, with microcephaly, 616212*
KIAA0556	*100%*	*100%*	*Joubert syndrome 26, 616784*
KCNE1	*100%*	*100%*	*Jervell and Lange-Nielsen syndrome 2, 612347* *Long QT syndrome 5, 613695*
KCNJ1	*100%*	*100%*	*Bartter syndrome, type 2, 241200*
KCNJ10	*100%*	*100%*	*Enlarged vestibular aqueduct, digenic, 600791* *SESAME syndrome, 612780*
KCNJ11	*100%*	*100%*	*Diabetes, permanent neonatal 2, with or without neurologic features, 618856* *Maturity-onset diabetes of the young, type 13, 616329* *Diabetes mellitus, transient neonatal 3, 610582* *Hyperinsulinemic hypoglycemia, familial, 2, 601820*
KCNJ13	*100%*	*100%*	*Snowflake vitreoretinal degeneration, 193230* *Leber congenital amaurosis 16, 614186*
KCNMA1	*100%*	*100%*	*Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446* *Cerebellar atrophy, developmental delay, and seizures, 617643* *Liang-Wang syndrome, 618729*
KCNQ1	*100%*	*100%*	*Short QT syndrome 2, 609621* *Atrial fibrillation, familial, 3, 607554* *Long QT syndrome 1, 192500* *Jervell and Lange-Nielsen syndrome, 220400*
KCNV2	*100%*	*100%*	*Retinal cone dystrophy 3B, 610356*
KCTD7	*100%*	*100%*	*Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726*
KDM5B	*98%*	*96%*	*Intellectual developmental disorder, autosomal recessive 65, 618109*
KERA	*100%*	*100%*	*Cornea plana 2, autosomal recessive, 217300*
KHDC3L	*100%*	*100%*	*Hydatidiform mole, recurrent, 2, 614293*
KIAA0586	*96%*	*96%*	*Short-rib thoracic dysplasia 14 with polydactyly, 616546* *Joubert syndrome 23, 616490*
KIAA0753	*100%*	*100%*	*?Orofaciodigital syndrome XV, 617127* *?Joubert syndrome 38, 619476* *Short-rib thoracic dysplasia 21 without polydactyly, 619479*
KIAA1549	*100%*	*100%*	*Retinitis pigmentosa 86, 618613*

KIF14	*100%*	*100%*	*Microcephaly 20, primary, autosomal recessive, 617914* *?Meckel syndrome 12, 616258*
KIF1A	*100%*	*100%*	*NESCAV syndrome, 614255* *Neuropathy, hereditary sensory, type IIC, 614213* *Spastic paraplegia 30, autosomal dominant, 610357* *Spastic paraplegia 30, autosomal recessive, 610357*
KIF1C	*100%*	*100%*	*Spastic ataxia 2, autosomal recessive, 611302*
KIF7	*100%*	*100%*	*Joubert syndrome 12, 200990* *Acrocallosal syndrome, 200990* *?Hydrolethalus syndrome 2, 614120* *?Al-Gazali-Bakalinova syndrome, 607131*
KIFBP	*96%*	*96%*	*Goldberg-Shprintzen megacolon syndrome, 609460*
KISS1R	*100%*	*100%*	*Hypogonadotropic hypogonadism 8 with or without anosmia, 614837* *?Precocious puberty, central, 1, 176400*
KIZ	*100%*	*100%*	*Retinitis pigmentosa 69, 615780*
KL	*100%*	*99%*	*?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994*
KLC2	*100%*	*100%*	*Spastic paraplegia, optic atrophy, and neuropathy, 609541*
KLHL3	*100%*	*100%*	*Pseudohypoaldosteronism, type IID, 614495*
KLHL40	*100%*	*100%*	*Nemaline myopathy 8, autosomal recessive, 615348*
KLHL41	*100%*	*100%*	*Nemaline myopathy 9, 615731*
KLHL7	*100%*	*100%*	*Retinitis pigmentosa 42, 612943* *PERCHING syndrome, 617055*
KLK4	*100%*	*100%*	*Amelogenesis imperfecta, type IIA1, 204700*
KLKB1	*100%*	*100%*	*Fletcher factor (prekallikrein) deficiency, 612423*
KMT2B	*100%*	*100%*	*Intellectual developmental disorder, autosomal dominant 68, 619934* *Dystonia 28, childhood-onset, 617284*
KNL1	*99%*	*99%*	*Microcephaly 4, primary, autosomal recessive, 604321*
KPTN	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 41, 615637*
KRT10	*100%*	*100%*	*Epidermolytic hyperkeratosis, 113800* *Ichthyosis with confetti, 609165* *Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602*

KRT14	*100%*	*100%*	*Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001* *Epidermolysis bullosa simplex 1C, localized, 131800* *Dermatopathia pigmentosa reticularis, 125595* *Epidermolysis bullosa simplex 1A, generalized severe, 131760* *Naegeli-Franceschetti-Jadassohn syndrome, 161000* *Epidermolysis bullosa simplex 1B, generalized intermediate, 131900*
KRT18	*100%*	*100%*	*Cirrhosis, cryptogenic, 215600*
KRT5	*100%*	*100%*	*Epidermolysis bullosa simplex 2A, generalized severe, 619555* *Dowling-Degos disease 1, 179850* *Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960* *Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599* *Epidermolysis bullosa simplex 2B, generalized intermediate, 619588* *Epidermolysis bullosa simplex 2C, localized, 619594* *Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352*
KRT8	*100%*	*100%*	*No OMIM disease ID*
KRT85	*100%*	*100%*	*Ectodermal dysplasia 4, hair/nail type, 602032*
KY	*100%*	*100%*	*Myopathy, myofibrillar, 7, 617114*
KYNU	*100%*	*100%*	*?Hydroxykynureninuria, 236800* *Vertebral, cardiac, renal, and limb defects syndrome 2, 617661*
L2HGDH	*100%*	*100%*	*L-2-hydroxyglutaric aciduria, 236792*
LAMA1	*100%*	*100%*	*Poretti-Boltshauser syndrome, 615960*
LAMA2	*100%*	*100%*	*Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138* *Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855*
LAMA3	*100%*	*100%*	*Epidermolysis bullosa, junctional 2A, intermediate, 619783* *Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660* *Epidermolysis bullosa, junctional 2B, severe, 619784*
LAMB1	*100%*	*100%*	*Lissencephaly 5, 615191*
LAMB2	*100%*	*100%*	*Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199* *Pierson syndrome, 609049*
LAMB3	*100%*	*100%*	*Epidermolysis bullosa, junctional 1B, severe, 226700* *Epidermolysis bullosa, junctional 1A, intermediate, 226650* *Amelogenesis imperfecta, type IA, 104530*

LAMC2	*100%*	*100%*	*Epidermolysis bullosa, junctional 3B, severe, 619786* *Epidermolysis bullosa, junctional 3A, intermediate, 619785*
LAMC3	*100%*	*100%*	*Cortical malformations, occipital, 614115*
LAMTOR2	*100%*	*100%*	*Immunodeficiency due to defect in MAPBP-interacting protein, 610798*
LARGE1	*100%*	*100%*	*Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154*
LARP7	*100%*	*100%*	*Alazami syndrome, 615071*
LARS1	*100%*	*100%*	*?Infantile liver failure syndrome 1, 615438*
LARS2	*100%*	*100%*	*Perrault syndrome 4, 615300* *Hydrops, lactic acidosis, and sideroblastic anemia, 617021*
LAT	*100%*	*100%*	*Immunodeficiency 52, 617514*
LBR	*100%*	*100%*	*Pelger-Huet anomaly, 169400* *?Reynolds syndrome, 613471* *Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019* *Greenberg skeletal dysplasia, 215140*
LCA5	*100%*	*100%*	*Leber congenital amaurosis 5, 604537*
LCAT	*100%*	*100%*	*Fish-eye disease, 136120* *Norum disease, 245900*
LCK	*100%*	*100%*	*?Immunodeficiency 22, 615758*
LCT	*100%*	*100%*	*Lactase deficiency, congenital, 223000*
LDHA	*100%*	*100%*	*Glycogen storage disease XI, 612933*
LDHD	*100%*	*100%*	*D-lactic aciduria with susceptibility to gout, 245450*
LEMD2	*100%*	*100%*	*Marbach-Rustad progeroid syndrome, 619322* *Cataract 46, juvenile-onset, 212500*
LEP	*100%*	*100%*	*Obesity, morbid, due to leptin deficiency, 614962*
LEPR	*95%*	*95%*	*Obesity, morbid, due to leptin receptor deficiency, 614963*
LFNG	*99%*	*97%*	*Spondylocostal dysostosis 3, autosomal recessive, 609813*
LGI4	*100%*	*100%*	*Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468*
LHB	*100%*	*100%*	*Hypogonadotropic hypogonadism 23 with or without anosmia, 228300*

LHCGR	*100%*	*100%*	*Leydig cell adenoma, somatic, with precocious puberty, 176410* *Leydig cell hypoplasia with pseudohermaphroditism, 238320* *Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320* *Luteinizing hormone resistance, female, 238320* *Precocious puberty, male, 176410*
LHFPL5	*100%*	*100%*	*Deafness, autosomal recessive 67, 610265*
LHX3	*100%*	*100%*	*Pituitary hormone deficiency, combined, 3, 221750*
LIAS	*100%*	*100%*	*Hyperglycinemia, lactic acidosis, and seizures, 614462*
LIFR	*100%*	*100%*	*Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559*
LIG4	*100%*	*100%*	*LIG4 syndrome, 606593*
LIM2	*100%*	*100%*	*Cataract 19, multiple types, 615277*
LIMS2	*100%*	*100%*	*?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827*
LINGO1	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 64, 618103*
LINS1	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 27, 614340*
LIPA	*97%*	*95%*	*Wolman disease, 278000* *Cholesteryl ester storage disease, 278000*
LIPE	*100%*	*100%*	*Lipodystrophy, familial partial, type 6, 615980*
LIPH	*100%*	*100%*	*Hypotrichosis 7, 604379* *Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379*
LIPN	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 8, 613943*
LIPT1	*100%*	*100%*	*Lipoyltransferase 1 deficiency, 616299*
LIPT2	*100%*	*100%*	*Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668*
LMAN1	*100%*	*100%*	*Combined factor V and VIII deficiency, 227300*
LMAN2L	*100%*	*100%*	*?Intellectual developmental disorder, autosomal dominant 69, 617863* *?Intellectual developmental disorder, autosomal recessive 52, 616887*
LMBR1	*100%*	*99%*	*Triphalangeal thumb, type I, 174500* *Syndactyly, type IV, 186200* *Laurin-Sandrow syndrome, 135750* *Hypoplastic or aplastic tibia with polydactyly, 188740* *Polydactyly, preaxial type II, 174500* *Acheiropody, 200500* *Triphalangeal thumb-polysyndactyly syndrome, 190605*
LMBRD1	*100%*	*100%*	*Methylmalonic aciduria and homocystinuria, cblF type, 277380*
LMF1	*100%*	*100%*	*Lipase deficiency, combined, 246650*

LMNA	*100%*	*100%*	*Mandibuloacral dysplasia, 248370* *Heart-hand syndrome, Slovenian type, 610140* *Cardiomyopathy, dilated, 1A, 115200* *Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516* *Restrictive dermopathy 2, 619793* *Charcot-Marie-Tooth disease, type 2B1, 605588* *Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350* *Hutchinson-Gilford progeria, 176670* *Lipodystrophy, familial partial, type 2, 151660* *Muscular dystrophy, congenital, 613205* *Malouf syndrome, 212112*
LMNB2	*100%*	*100%*	*Microcephaly 27, primary, autosomal dominant, 619180* *?Epilepsy, progressive myoclonic, 9, 616540*
LMOD3	*100%*	*100%*	*Nemaline myopathy 10, 616165*
LNPK	*93%*	*93%*	*Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090*
LONP1	*100%*	*100%*	*CODAS syndrome, 600373*
LOXHD1	*100%*	*100%*	*Deafness, autosomal recessive 77, 613079*
LPAR6	*100%*	*100%*	*Hypotrichosis 8, 278150* *Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150*
LPIN1	*100%*	*100%*	*Myoglobinuria, acute recurrent, autosomal recessive, 268200*
LPIN2	*100%*	*100%*	*Majeed syndrome, 609628*
LPL	*100%*	*100%*	*Lipoprotein lipase deficiency, 238600* *Combined hyperlipidemia, familial, 144250*
LRAT	*100%*	*100%*	*Leber congenital amaurosis 14, 613341* *Retinal dystrophy, early-onset severe, 613341* *Retinitis pigmentosa, juvenile, 613341*
LRBA	*100%*	*100%*	*Immunodeficiency, common variable, 8, with autoimmunity, 614700*
LRIG2	*100%*	*100%*	*Urofacial syndrome 2, 615112*
LRIT3	*100%*	*100%*	*Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058*
LRMDA	*98%*	*98%*	*Albinism, oculocutaneous, type VII, 615179*
LRP1	*100%*	*100%*	*?Keratosis pilaris atrophicans, 604093*
LRP2	*100%*	*100%*	*Donnai-Barrow syndrome, 222448*
LRP4	*100%*	*100%*	*?Myasthenic syndrome, congenital, 17, 616304* *Sclerosteosis 2, 614305* *Cenani-Lenz syndactyly syndrome, 212780*

LRP5	*100%*	*100%*	*Osteopetrosis, autosomal dominant 1, 607634* *Hyperostosis, endosteal, 144750* *Osteosclerosis, 144750* *Polycystic liver disease 4 with or without kidney cysts, 617875* *Osteoporosis-pseudoglioma syndrome, 259770* *Exudative vitreoretinopathy 4, 601813* *van Buchem disease, type 2, 607636*
LRPAP1	*100%*	*100%*	*Myopia 23, autosomal recessive, 615431*
LRPPRC	*100%*	*100%*	*Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111*
LRRC56	*100%*	*100%*	*Ciliary dyskinesia, primary, 39, 618254*
LRSAM1	*100%*	*100%*	*Charcot-Marie-Tooth disease, axonal, type 2P, 614436*
LRTOMT	*100%*	*100%*	*Deafness, autosomal recessive 63, 611451*
LSS	*100%*	*100%*	*Hypotrichosis 14, 618275* *Cataract 44, 616509* *Alopecia-intellectual disability syndrome 4, 618840*
LTBP1	*100%*	*100%*	*Cutis laxa, autosomal recessive, type IIE, 619451*
LTBP2	*100%*	*100%*	*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*
LTBP3	*100%*	*100%*	*Dental anomalies and short stature, 601216* *Geleophysic dysplasia 3, 617809*
LTBP4	*100%*	*100%*	*Cutis laxa, autosomal recessive, type IC, 613177*
LTC4S	*100%*	*100%*	*No OMIM disease ID*
LYRM7	*100%*	*100%*	*Mitochondrial complex III deficiency, nuclear type 8, 615838*
LYST	*100%*	*100%*	*Chediak-Higashi syndrome, 214500*
LZTFL1	*100%*	*100%*	*Bardet-Biedl syndrome 17, 615994*
LZTR1	*100%*	*100%*	*Noonan syndrome 2, 605275* *Noonan syndrome 10, 616564*
MAB21L1	*100%*	*100%*	*Cerebellar, ocular, craniofacial, and genital syndrome, 618479*
MAB21L2	*100%*	*100%*	*Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877*
MADD	*100%*	*100%*	*Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005* *DEEAH syndrome, 619004*
MAG	*100%*	*100%*	*Spastic paraplegia 75, autosomal recessive, 616680*
MAGI2	*99%*	*97%*	*Nephrotic syndrome, type 15, 617609*

MAK	*100%*	*100%*	*Retinitis pigmentosa 62, 614181*
MALT1	*100%*	*100%*	*Immunodeficiency 12, 615468*
MAN1B1	*100%*	*100%*	*Rafiq syndrome, 614202*
MAN2B1	*100%*	*100%*	*Mannosidosis, alpha-, types I and II, 248500*
MANBA	*100%*	*100%*	*Mannosidosis, beta, 248510*
MAP3K20	*100%*	*100%*	*Centronuclear myopathy 6 with fiber-type disproportion, 617760* *Split-foot malformation with mesoaxial polydactyly, 616890*
MAPKBP1	*100%*	*100%*	*Nephronophthisis 20, 617271*
MAPT	*100%*	*100%*	*Supranuclear palsy, progressive, 601104* *Supranuclear palsy, progressive atypical, 260540* *Dementia, frontotemporal, with or without parkinsonism, 600274* *Pick disease, 172700*
MARS1	*100%*	*100%*	*Interstitial lung and liver disease, 615486* *?Trichothiodystrophy 9, nonphotosensitive, 619692* *Charcot-Marie-Tooth disease, axonal, type 2U, 616280*
MARS2	*100%*	*100%*	*?Combined oxidative phosphorylation deficiency 25, 616430* *Spastic ataxia 3, autosomal recessive, 611390*
MARVELD2	*100%*	*100%*	*Deafness, autosomal recessive 49, 610153*
MASP1	*100%*	*100%*	*3MC syndrome 1, 257920*
MAT1A	*100%*	*100%*	*Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850* *Methionine adenosyltransferase deficiency, autosomal recessive, 250850*
MATN3	*100%*	*100%*	*Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728* *Epiphyseal dysplasia, multiple, 5, 607078*
MBOAT7	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 57, 617188*
MBTPS1	*100%*	*100%*	*?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392*
MC2R	*100%*	*100%*	*Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200*
MCCC1	*100%*	*100%*	*3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200*
MCCC2	*100%*	*100%*	*3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210*
MCEE	*100%*	*100%*	*Methylmalonyl-CoA epimerase deficiency, 251120*
MCFD2	*100%*	*100%*	*Factor V and factor VIII, combined deficiency of, 613625*
MCM3AP	*100%*	*100%*	*Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124*
MCM4	*95%*	*95%*	*Immunodeficiency 54, 609981*
MCM5	*100%*	*100%*	*?Meier-Gorlin syndrome 8, 617564*
MCM9	*100%*	*100%*	*Ovarian dysgenesis 4, 616185*

MCOLN1	*100%*	*100%*	*Mucolipidosis IV, 252650*
MCPH1	*100%*	*100%*	*Microcephaly 1, primary, autosomal recessive, 251200*
MDH2	*100%*	*100%*	*Developmental and epileptic encephalopathy 51, 617339*
MECR	*100%*	*100%*	*Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282*
MED17	*100%*	*100%*	*Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668*
MED23	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249*
MED25	*100%*	*100%*	*Basel-Vanagait-Smirin-Yosef syndrome, 616449*
MEFV	*96%*	*96%*	*Neutrophilic dermatosis, acute febrile, 608068* *Familial Mediterranean fever, AR, 249100* *Familial Mediterranean fever, AD, 134610*
MEGF10	*100%*	*100%*	*Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399* *Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399*
MEGF8	*100%*	*100%*	*Carpenter syndrome 2, 614976*
MEOX1	*100%*	*100%*	*Klippel-Feil syndrome 2, 214300*
MERTK	*99%*	*99%*	*Retinitis pigmentosa 38, 613862*
MESD	*100%*	*100%*	*Osteogenesis imperfecta, type XX, 618644*
MESP2	*100%*	*100%*	*Spondylocostal dysostosis 2, autosomal recessive, 608681*
MET	*100%*	*100%*	*Renal cell carcinoma, papillary, 1, familial and somatic, 605074* *?Arthrogryposis, distal, type 11, 620019* *Hepatocellular carcinoma, childhood type, somatic, 114550* *?Deafness, autosomal recessive 97, 616705*
METTL23	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 44, 615942*
MFF	*100%*	*100%*	*Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086*
MFN2	*100%*	*100%*	*Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260* *Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087* *Hereditary motor and sensory neuropathy VIA, 601152*
MFRP	*100%*	*100%*	*Microphthalmia, isolated 5, 611040* *Nanophthalmos 2, 609549*
MFSD2A	*100%*	*100%*	*Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486*
MFSD8	*100%*	*100%*	*Macular dystrophy with central cone involvement, 616170* *Ceroid lipofuscinosis, neuronal, 7, 610951*
MGAT2	*100%*	*100%*	*Congenital disorder of glycosylation, type IIa, 212066*
MGME1	*100%*	*100%*	*Mitochondrial DNA depletion syndrome 11, 615084*
MGP	*100%*	*100%*	*Keutel syndrome, 245150*
MICOS13	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 37, 618329*

MICU1	*100%*	*100%*	*Myopathy with extrapyramidal signs, 615673*
MIPEP	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 31, 617228*
MITF	*100%*	*100%*	*Waardenburg syndrome, type 2A, 193510* *Tietz albinism-deafness syndrome, 103500* *Waardenburg syndrome/ocular albinism, digenic, 103470* *COMMAD syndrome, 617306*
MKKS	*100%*	*100%*	*McKusick-Kaufman syndrome, 236700* *Bardet-Biedl syndrome 6, 605231*
MKS1	*100%*	*100%*	*Bardet-Biedl syndrome 13, 615990* *Meckel syndrome 1, 249000* *Joubert syndrome 28, 617121*
MLC1	*100%*	*100%*	*Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004*
MLH1	*100%*	*100%*	*Lynch syndrome 2, 609310* *Muir-Torre syndrome, 158320* *Mismatch repair cancer syndrome 1, 276300*
MLPH	*100%*	*100%*	*Griscelli syndrome, type 3, 609227*
MLYCD	*100%*	*100%*	*Malonyl-CoA decarboxylase deficiency, 248360*
MMAA	*100%*	*100%*	*Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100*
MMAB	*100%*	*100%*	*Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110*
MMACHC	*100%*	*100%*	*Methylmalonic aciduria and homocystinuria, cblC type, 277400*
MMADHC	*89%*	*89%*	*Methylmalonic aciduria, cblD type, variant 2, 277410* *Methylmalonic aciduria and homocystinuria, cblD type, 277410* *Homocystinuria, cblD type, variant 1, 277410*
MME	*98%*	*97%*	*?Spinocerebellar ataxia 43, 617018* *Charcot-Marie-Tooth disease, axonal, type 2T, 617017*
MMP13	*92%*	*92%*	*?Spondyloepimetaphyseal dysplasia, Missouri type, 602111* *Metaphyseal anadysplasia 1, 602111* *Metaphyseal dysplasia, Spahr type, 250400*
MMP14	*100%*	*100%*	*?Winchester syndrome, 277950*
MMP2	*100%*	*100%*	*Multicentric osteolysis, nodulosis, and arthropathy, 259600*
MMP20	*100%*	*100%*	*Amelogenesis imperfecta, type IIA2, 612529*
MMP21	*100%*	*100%*	*Heterotaxy, visceral, 7, autosomal, 616749*
MMP9	*100%*	*100%*	*Metaphyseal anadysplasia 2, 613073*
MMUT	*100%*	*100%*	*Methylmalonic aciduria, mut(0) type, 251000*
MOCOS	*100%*	*100%*	*Xanthinuria, type II, 603592*

MOCS1	*100%*	*100%*	*Molybdenum cofactor deficiency A, 252150*
MOCS2	*100%*	*100%*	*Molybdenum cofactor deficiency B, 252160*
MOGS	*100%*	*100%*	*Congenital disorder of glycosylation, type IIb, 606056*
MPC1	*100%*	*100%*	*Mitochondrial pyruvate carrier deficiency, 614741*
MPDU1	*100%*	*100%*	*Congenital disorder of glycosylation, type If, 609180*
MPDZ	*100%*	*99%*	*Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219*
MPI	*100%*	*100%*	*Congenital disorder of glycosylation, type Ib, 602579*
MPL	*100%*	*100%*	*Myelofibrosis with myeloid metaplasia, somatic, 254450* *Thrombocythemia 2, 601977* *Thrombocytopenia, congenital amegakaryocytic, 604498*
MPLKIP	*100%*	*100%*	*Trichothiodystrophy 4, nonphotosensitive, 234050*
MPO	*100%*	*100%*	*Myeloperoxidase deficiency, 254600*
MPV17	*100%*	*100%*	*Charcot-Marie-Tooth disease, axonal, type 2EE, 618400* *Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810*
MPZ	*100%*	*100%*	*Charcot-Marie-Tooth disease, type 2I, 607677* *Dejerine-Sottas disease, 145900* *Charcot-Marie-Tooth disease, type 1B, 118200* *Roussy-Levy syndrome, 180800* *Charcot-Marie-Tooth disease, dominant intermediate D, 607791* *Hypomyelinating neuropathy, congenital, 2, 618184* *Charcot-Marie-Tooth disease, type 2J, 607736*
MPZL2	*100%*	*100%*	*Deafness, autosomal recessive 111, 618145*
MRAP	*100%*	*100%*	*Glucocorticoid deficiency 2, 607398*
MRE11	*100%*	*100%*	*Ataxia-telangiectasia-like disorder 1, 604391*
MRM2	*97%*	*97%*	*?Mitochondrial DNA depletion syndrome 17, 618567*
MRPL3	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 9, 614582*
MRPL44	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 16, 615395*
MRPS16	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 2, 610498*
MRPS2	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 36, 617950*
MRPS22	*100%*	*100%*	*Ovarian dysgenesis 7, 618117* *Combined oxidative phosphorylation deficiency 5, 611719*
MRPS34	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 32, 617664*
MS4A1	*100%*	*100%*	*?Immunodeficiency, common variable, 5, 613495*

MSH2	*100%*	*100%*	*Lynch syndrome 1, 120435* *Muir-Torre syndrome, 158320* *Mismatch repair cancer syndrome 2, 619096*
MSH3	*100%*	*100%*	*Familial adenomatous polyposis 4, 617100* *Endometrial carcinoma, somatic, 608089*
MSH6	*100%*	*100%*	*Lynch syndrome 5, 614350* *Mismatch repair cancer syndrome 3, 619097*
MSMO1	*100%*	*100%*	*Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834*
MSRB3	*100%*	*100%*	*Deafness, autosomal recessive 74, 613718*
MSTO1	*100%*	*100%*	*Myopathy, mitochondrial, and ataxia, 617675*
MTFMT	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 15, 614947* *Mitochondrial complex I deficiency, nuclear type 27, 618248*
MTHFD1	*100%*	*100%*	*Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780*
MTHFR	*100%*	*100%*	*Homocystinuria due to MTHFR deficiency, 236250*
MTHFS	*100%*	*100%*	*Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367*
MTMR2	*100%*	*100%*	*Charcot-Marie-Tooth disease, type 4B1, 601382*
MTO1	*94%*	*91%*	*Combined oxidative phosphorylation deficiency 10, 614702*
MTPAP	*100%*	*100%*	*?Spastic ataxia 4, autosomal recessive, 613672*
MTR	*100%*	*100%*	*Homocystinuria-megaloblastic anemia, cblG complementation type, 250940*
C12orf65	*100%*	*100%*	*Spastic paraplegia 55, autosomal recessive, 615035* *Combined oxidative phosphorylation deficiency 7, 613559*
MTRR	*100%*	*100%*	*Homocystinuria-megaloblastic anemia, cbl E type, 236270*
MTTP	*100%*	*100%*	*Abetalipoproteinemia, 200100*
MUSK	*100%*	*100%*	*Fetal akinesia deformation sequence 1, 208150* *Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325*
MUTYH	*100%*	*100%*	*Adenomas, multiple colorectal, 608456* *Gastric cancer, somatic, 613659*
MVK	*90%*	*90%*	*Hyper-IgD syndrome, 260920* *Porokeratosis 3, multiple types, 175900* *Mevalonic aciduria, 610377*
MYBPC1	*100%*	*100%*	*Myopathy, congenital, with tremor, 618524* *Lethal congenital contracture syndrome 4, 614915* *Arthrogryposis, distal, type 1B, 614335*
MYD88	*100%*	*100%*	*Macroglobulinemia, Waldenstrom, somatic, 153600* *Immunodeficiency 68, 612260*

MYF5	*100%*	*100%*	*Ophthalmoplegia, external, with rib and vertebral anomalies, 618155*
MYH2	*100%*	*100%*	*Proximal myopathy and ophthalmoplegia, 605637*
MYH3	*100%*	*100%*	*Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110* *Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469* *Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436* *Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700*
MYL1	*100%*	*100%*	*Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414*
MYL2	*100%*	*100%*	*Cardiomyopathy, hypertrophic, 10, 608758* *Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424*
MYL3	*100%*	*100%*	*Cardiomyopathy, hypertrophic, 8, 608751*
MYLK	*100%*	*100%*	*Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210* *Aortic aneurysm, familial thoracic 7, 613780*
MYMK	*100%*	*100%*	*Carey-Fineman-Ziter syndrome, 254940*
MYO15A	*100%*	*100%*	*Deafness, autosomal recessive 3, 600316*
MYO18B	*100%*	*100%*	*Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549*
MYO1E	*100%*	*100%*	*Glomerulosclerosis, focal segmental, 6, 614131*
MYO3A	*100%*	*100%*	*Deafness, autosomal recessive 30, 607101*
MYO5A	*100%*	*100%*	*Griscelli syndrome, type 1, 214450*
MYO5B	*100%*	*100%*	*Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850* *Cholestasis, progressive familial intrahepatic, 10, 619868*
MYO6	*100%*	*100%*	*Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346* *Deafness, autosomal dominant 22, 606346* *Deafness, autosomal recessive 37, 607821*
MYO7A	*100%*	*100%*	*Deafness, autosomal recessive 2, 600060* *Usher syndrome, type 1B, 276900* *Deafness, autosomal dominant 11, 601317*
MYO9A	*100%*	*100%*	*Myasthenic syndrome, congenital, 24, presynaptic, 618198*
MYORG	*100%*	*100%*	*Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317*
MYPN	*100%*	*100%*	*Cardiomyopathy, hypertrophic, 22, 615248* *Cardiomyopathy, familial restrictive, 4, 615248* *Cardiomyopathy, dilated, 1KK, 615248* *Nemaline myopathy 11, autosomal recessive, 617336*
MYSM1	*100%*	*100%*	*Bone marrow failure syndrome 4, 618116*
NADK2	*100%*	*100%*	*2,4-dienoyl-CoA reductase deficiency, 616034*
NADSYN1	*100%*	*100%*	*Vertebral, cardiac, renal, and limb defects syndrome 3, 618845*

NAGA	*100%*	*100%*	*Schindler disease, type I, 609241* *Kanzaki disease, 609242* *Schindler disease, type III, 609241*
NAGLU	*100%*	*100%*	*?Charcot-Marie-Tooth disease, axonal, type 2V, 616491* *Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920*
NAGS	*100%*	*100%*	*N-acetylglutamate synthase deficiency, 237310*
NALCN	*100%*	*100%*	*Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266* *Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419*
NANS	*100%*	*100%*	*Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442*
NARS2	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 24, 616239* *?Deafness, autosomal recessive 94, 618434*
NAT8L	*99%*	*93%*	*?N-acetylaspartate deficiency, 614063*
NAXD	*100%*	*100%*	*Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321*
NAXE	*100%*	*100%*	*Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186*
NBAS	*100%*	*100%*	*Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800* *Infantile liver failure syndrome 2, 616483*
NBEAL2	*100%*	*100%*	*Gray platelet syndrome, 139090*
NBN	*100%*	*100%*	*Leukemia, acute lymphoblastic, 613065* *Aplastic anemia, 609135* *Nijmegen breakage syndrome, 251260*
NCAPD2	*100%*	*100%*	*?Microcephaly 21, primary, autosomal recessive, 617983*
NCAPD3	*100%*	*100%*	*Microcephaly 22, primary, autosomal recessive, 617984*
NCAPG2	*100%*	*100%*	*Khan-Khan-Katsanis syndrome, 618460*
NCAPH	*100%*	*100%*	*?Microcephaly 23, primary, autosomal recessive, 617985*
NCF1	*100%*	*100%*	*Chronic granulomatous disease 1, autosomal recessive, 233700*
NCF2	*100%*	*100%*	*Chronic granulomatous disease 2, autosomal recessive, 233710*
NCF4	*100%*	*100%*	*Chronic granulomatous disease 3, autosomal recessive, 613960*
NDE1	*100%*	*100%*	*Microhydranencephaly, 605013* *Lissencephaly 4 (with microcephaly), 614019*
NDRG1	*100%*	*100%*	*Charcot-Marie-Tooth disease, type 4D, 601455*
NDST1	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 46, 616116*
NDUFA10	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 22, 618243*
NDUFA11	*100%*	*99%*	*Mitochondrial complex I deficiency, nuclear type 14, 618236*
NDUFA12	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 23, 618244*
NDUFA13	*100%*	*100%*	*?Mitochondrial complex I deficiency, nuclear type 28, 618249*

NDUFA2	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 13, 618235*
NDUFA6	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 33, 618253*
NDUFA9	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 26, 618247*
NDUFAF1	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 11, 618234*
NDUFAF2	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 10, 618233*
NDUFAF3	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 18, 618240*
NDUFAF4	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 15, 618237*
NDUFAF5	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 16, 618238*
NDUFAF6	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 17, 618239* *Fanconi renotubular syndrome 5, 618913*
NDUFB11	*100%*	*98%*	*Linear skin defects with multiple congenital anomalies 3, 300952* *?Mitochondrial complex I deficiency, nuclear type 30, 301021*
NDUFB3	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 25, 618246*
NDUFB8	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 32, 618252*
NDUFB9	*100%*	*100%*	*?Mitochondrial complex I deficiency, nuclear type 24, 618245*
NDUFS1	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 5, 618226*
NDUFS2	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 6, 618228*
NDUFS3	*97%*	*91%*	*Mitochondrial complex I deficiency, nuclear type 8, 618230*
NDUFS4	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 1, 252010*
NDUFS6	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 9, 618232*
NDUFS7	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 3, 618224*
NDUFS8	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 2, 618222*
NDUFV1	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 4, 618225*
NDUFV2	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 7, 618229*
NEB	*100%*	*99%*	*Nemaline myopathy 2, autosomal recessive, 256030* *Arthrogryposis multiplex congenita 6, 619334*
NECAP1	*100%*	*100%*	*Developmental and epileptic encephalopathy 21, 615833*
NECTIN1	*100%*	*100%*	*Cleft lip/palate-ectodermal dysplasia syndrome, 225060* *Orofacial cleft 7, 225060*
NECTIN4	*100%*	*100%*	*Ectodermal dysplasia-syndactyly syndrome 1, 613573*
NEK1	*100%*	*100%*	*Short-rib thoracic dysplasia 6 with or without polydactyly, 263520*
NEK2	*96%*	*96%*	*?Retinitis pigmentosa 67, 615565*
NEK8	*100%*	*100%*	*Renal-hepatic-pancreatic dysplasia 2, 615415* *?Nephronophthisis 9, 613824*

NEK9	*100%*	*100%*	*?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262* *Nevus comedonicus, somatic, 617025* *Lethal congenital contracture syndrome 10, 617022*
NEPRO	*100%*	*100%*	*Anauxetic dysplasia 3, 618853*
NEU1	*100%*	*100%*	*Sialidosis, type II, 256550* *Sialidosis, type I, 256550*
NEUROG3	*100%*	*100%*	*Diarrhea 4, malabsorptive, congenital, 610370*
NFASC	*100%*	*100%*	*Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356*
NFU1	*100%*	*100%*	*Multiple mitochondrial dysfunctions syndrome 1, 605711*
NGF	*100%*	*100%*	*Neuropathy, hereditary sensory and autonomic, type V, 608654*
NGLY1	*100%*	*100%*	*Congenital disorder of deglycosylation 1, 615273*
NHEJ1	*100%*	*100%*	*Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291*
NHLRC1	*100%*	*100%*	*Epilepsy, progressive myoclonic 2B (Lafora), 254780*
NHLRC2	*100%*	*100%*	*FINCA syndrome, 618278*
NHP2	*100%*	*100%*	*Dyskeratosis congenita, autosomal recessive 2, 613987*
NIN	*100%*	*100%*	*?Seckel syndrome 7, 614851*
NIPAL4	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 6, 612281*
NKX2-6	*100%*	*100%*	*Persistent truncus arteriosus, 217095* *Conotruncal heart malformations, 217095*
NKX3-2	*100%*	*100%*	*Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330*
NKX6-2	*100%*	*100%*	*Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560*
NLRP1	*100%*	*100%*	*?Respiratory papillomatosis, juvenile recurrent, congenital, 618803* *Autoinflammation with arthritis and dyskeratosis, 617388* *Palmoplantar carcinoma, multiple self-healing, 615225*
NLRP7	*100%*	*100%*	*Hydatidiform mole, recurrent, 1, 231090*
NME8	*100%*	*100%*	*Ciliary dyskinesia, primary, 6, 610852*
NMNAT1	*100%*	*98%*	*Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260* *Leber congenital amaurosis 9, 608553*
NNT	*96%*	*96%*	*Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736*
NOP10	*100%*	*100%*	*Dyskeratosis congenita, autosomal recessive 1, 224230*
NPC1	*100%*	*100%*	*Niemann-Pick disease, type C1, 257220* *Niemann-Pick disease, type D, 257220*
NPC2	*100%*	*100%*	*Niemann-pick disease, type C2, 607625*

NPHP1	*100%*	*100%*	*Joubert syndrome 4, 609583* *Nephronophthisis 1, juvenile, 256100* *Senior-Loken syndrome-1, 266900*
NPHP3	*100%*	*100%*	*Nephronophthisis 3, 604387* *Renal-hepatic-pancreatic dysplasia 1, 208540* *Meckel syndrome 7, 267010*
NPHP4	*100%*	*100%*	*Senior-Loken syndrome 4, 606996* *Nephronophthisis 4, 606966*
NPHS1	*100%*	*100%*	*Nephrotic syndrome, type 1, 256300*
NPHS2	*100%*	*100%*	*Nephrotic syndrome, type 2, 600995*
NPPA	*100%*	*100%*	*Atrial standstill 2, 615745* *Atrial fibrillation, familial, 6, 612201*
NPR2	*100%*	*100%*	*Epiphyseal chondrodysplasia, Miura type, 615923* *Short stature with nonspecific skeletal abnormalities, 616255* *Acromesomelic dysplasia 1, Maroteaux type, 602875*
NR0B2	*100%*	*100%*	*Obesity, mild, early-onset, 601665*
NR1H4	*100%*	*100%*	*Cholestasis, progressive familial intrahepatic, 5, 617049*
NR2E3	*100%*	*100%*	*Retinitis pigmentosa 37, 611131* *Enhanced S-cone syndrome, 268100*
NRCAM	*100%*	*100%*	*Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833*
NRROS	*100%*	*100%*	*Seizures, early-onset, with neurodegeneration and brain calcification, 618875*
NRXN1	*100%*	*100%*	*Pitt-Hopkins-like syndrome 2, 614325*
NSMCE2	*100%*	*100%*	*Seckel syndrome 10, 617253*
NSMCE3	*100%*	*100%*	*Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241*
NSUN2	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 5, 611091*
NT5C2	*100%*	*100%*	*Spastic paraplegia 45, autosomal recessive, 613162*
NT5C3A	*100%*	*100%*	*Anemia, hemolytic, due to UMPH1 deficiency, 266120*
NT5E	*100%*	*100%*	*Calcification of joints and arteries, 211800*
NTHL1	*100%*	*100%*	*Familial adenomatous polyposis 3, 616415*
NTNG2	*100%*	*100%*	*Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718*
NTRK1	*100%*	*100%*	*Insensitivity to pain, congenital, with anhidrosis, 256800*
NUBPL	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 21, 618242*
NUP107	*100%*	*100%*	*?Ovarian dysgenesis 6, 618078* *Galloway-Mowat syndrome 7, 618348* *Nephrotic syndrome, type 11, 616730*

NUP133	*100%*	*100%*	*?Galloway-Mowat syndrome 8, 618349* *Nephrotic syndrome, type 18, 618177*
NUP160	*100%*	*100%*	*?Nephrotic syndrome, type 19, 618178*
NUP188	*100%*	*100%*	*Sandestig-Stefanova syndrome, 618804*
NUP205	*100%*	*100%*	*?Nephrotic syndrome, type 13, 616893*
NUP214	*100%*	*100%*	*Leukemia, T-cell acute lymphoblastic, somatic, 613065* *Leukemia, acute myeloid, somatic, 601626*
NUP37	*100%*	*100%*	*?Microcephaly 24, primary, autosomal recessive, 618179*
NUP62	*100%*	*100%*	*Striatonigral degeneration, infantile, 271930*
NUP85	*100%*	*100%*	*Nephrotic syndrome, type 17, 618176*
NUP88	*100%*	*100%*	*Fetal akinesia deformation sequence 4, 618393*
NUP93	*96%*	*96%*	*Nephrotic syndrome, type 12, 616892*
NUS1	*100%*	*100%*	*Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831* *?Congenital disorder of glycosylation, type 1aa, 617082*
NXN	*100%*	*100%*	*Robinow syndrome, autosomal recessive 2, 618529*
OAT	*100%*	*100%*	*Gyrate atrophy of choroid and retina with or without ornithinemia, 258870*
OBSL1	*100%*	*100%*	*3-M syndrome 2, 612921*
OCA2	*100%*	*100%*	*Albinism, brown oculocutaneous, 203200* *Albinism, oculocutaneous, type II, 203200*
OCLN	*100%*	*100%*	*Pseudo-TORCH syndrome 1, 251290*
CCDC114	*100%*	*100%*	*Ciliary dyskinesia, primary, 20, 615067*
ARMC4	*96%*	*96%*	*Ciliary dyskinesia, primary, 23, 615451*
CCDC151	*100%*	*100%*	*Ciliary dyskinesia, primary, 30, 616037*
TTC25	*100%*	*100%*	*Ciliary dyskinesia, primary, 35, 617092*
ODAPH	*100%*	*100%*	*Amelogenesis imperfecta, type IIA4, 614832*
OGDH	*100%*	*100%*	*?Oxoglutarate dehydrogenase deficiency, 203740*
OGDHL	*100%*	*100%*	*Yoon-Bellen neurodevelopmental syndrome, 619701*
OPA1	*100%*	*100%*	*Optic atrophy plus syndrome, 125250* *Optic atrophy 1, 165500* *Behr syndrome, 210000* *?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896*
OPA3	*100%*	*100%*	*3-methylglutaconic aciduria, type III, 258501* *Optic atrophy 3 with cataract, 165300*
OPLAH	*100%*	*100%*	*5-oxoprolinase deficiency, 260005*

ORAI1	*100%*	*100%*	*Immunodeficiency 9, 612782* *Myopathy, tubular aggregate, 2, 615883*
ORC1	*100%*	*100%*	*Meier-Gorlin syndrome 1, 224690*
ORC4	*99%*	*98%*	*Meier-Gorlin syndrome 2, 613800*
ORC6	*100%*	*100%*	*Meier-Gorlin syndrome 3, 613803*
OSGEP	*100%*	*100%*	*Galloway-Mowat syndrome 3, 617729*
OSTM1	*100%*	*100%*	*Osteopetrosis, autosomal recessive 5, 259720*
OTOA	*100%*	*100%*	*Deafness, autosomal recessive 22, 607039*
OTOF	*100%*	*100%*	*Auditory neuropathy, autosomal recessive, 1, 601071* *Deafness, autosomal recessive 9, 601071*
OTOG	*100%*	*100%*	*Deafness, autosomal recessive 18B, 614945*
OTOGL	*100%*	*100%*	*Deafness, autosomal recessive 84B, 614944*
OTUD6B	*100%*	*100%*	*Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452*
OTULIN	*100%*	*100%*	*Autoinflammation, panniculitis, and dermatosis syndrome, 617099*
OXCT1	*100%*	*100%*	*Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050*
P2RY12	*100%*	*100%*	*Bleeding disorder, platelet-type, 8, 609821*
P3H1	*100%*	*100%*	*Osteogenesis imperfecta, type VIII, 610915*
P3H2	*100%*	*100%*	*Myopia, high, with cataract and vitreoretinal degeneration, 614292*
P4HTM	*100%*	*100%*	*Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493*
PAH	*100%*	*100%*	*Phenylketonuria, 261600*
PAM16	*85%*	*85%*	*Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320*
PANK2	*100%*	*100%*	*HARP syndrome, 607236* *Neurodegeneration with brain iron accumulation 1, 234200*
PAPSS2	*100%*	*100%*	*Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847*
PARK7	*100%*	*100%*	*Parkinson disease 7, autosomal recessive early-onset, 606324*
PARN	*97%*	*96%*	*Dyskeratosis congenita, autosomal recessive 6, 616353* *Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371*
PARS2	*100%*	*100%*	*Developmental and epileptic encephalopathy 75, 618437*
PATL2	*100%*	*100%*	*Oocyte maturation defect 4, 617743*
PAX1	*100%*	*100%*	*Otofaciocervical syndrome 2, 615560*
PAX3	*100%*	*100%*	*Craniofacial-deafness-hand syndrome, 122880* *Waardenburg syndrome, type 3, 148820* *Waardenburg syndrome, type 1, 193500* *Rhabdomyosarcoma 2, alveolar, 268220*

PAX7	*100%*	*100%*	*Rhabdomyosarcoma 2, alveolar, 268220* *Myopathy, congenital, progressive, with scoliosis, 618578*
PC	*100%*	*100%*	*Pyruvate carboxylase deficiency, 266150*
PCARE	*100%*	*100%*	*Retinitis pigmentosa 54, 613428*
PCBD1	*100%*	*100%*	*Hyperphenylalaninemia, BH4-deficient, D, 264070*
PCCA	*100%*	*100%*	*Propionicacidemia, 606054*
PCCB	*100%*	*98%*	*Propionicacidemia, 606054*
PCDH12	*100%*	*100%*	*Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280*
PCDH15	*100%*	*100%*	*Usher syndrome, type 1D/F digenic, 601067* *Deafness, autosomal recessive 23, 609533* *Usher syndrome, type 1F, 602083*
PCK1	*100%*	*100%*	*?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680*
PCK2	*100%*	*100%*	*No OMIM disease ID*
PCLO	*100%*	*100%*	*?Pontocerebellar hypoplasia, type 3, 608027*
PCNT	*100%*	*100%*	*Microcephalic osteodysplastic primordial dwarfism, type II, 210720*
PCSK1	*100%*	*100%*	*Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955*
PCYT1A	*100%*	*100%*	*Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940*
PDE10A	*100%*	*99%*	*Striatal degeneration, autosomal dominant, 616922* *Dyskinesia, limb and orofacial, infantile-onset, 616921*
PDE6A	*100%*	*100%*	*Retinitis pigmentosa 43, 613810*
PDE6B	*100%*	*100%*	*Retinitis pigmentosa-40, 613801* *Night blindness, congenital stationary, autosomal dominant 2, 163500*
PDE6C	*100%*	*100%*	*Cone dystrophy 4, 613093*
PDE6D	*100%*	*100%*	*Joubert syndrome 22, 615665*
PDE6G	*100%*	*100%*	*Retinitis pigmentosa 57, 613582*
PDE6H	*100%*	*100%*	*Retinal cone dystrophy 3, 610024* *Achromatopsia 6, 610024*
PDHB	*100%*	*100%*	*Pyruvate dehydrogenase E1-beta deficiency, 614111*
PDHX	*100%*	*100%*	*Lacticacidemia due to PDX1 deficiency, 245349*
PDP1	*100%*	*100%*	*Pyruvate dehydrogenase phosphatase deficiency, 608782*
PDSS1	*100%*	*100%*	*Coenzyme Q10 deficiency, primary, 2, 614651*
PDSS2	*100%*	*100%*	*Coenzyme Q10 deficiency, primary, 3, 614652*
PDX1	*100%*	*100%*	*Pancreatic agenesis 1, 260370* *MODY, type IV, 606392*

PDXK	*100%*	*97%*	*Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511*
PDZD7	*100%*	*99%*	*Deafness, autosomal recessive 57, 618003* *Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472*
PEPD	*100%*	*100%*	*Prolidase deficiency, 170100*
PET100	*100%*	*100%*	*Mitochondrial complex IV deficiency, nuclear type 12, 619055*
PEX1	*100%*	*100%*	*Heimler syndrome 1, 234580* *Peroxisome biogenesis disorder 1B (NALD/IRD), 601539* *Peroxisome biogenesis disorder 1A (Zellweger), 214100*
PEX10	*100%*	*100%*	*Peroxisome biogenesis disorder 6A (Zellweger), 614870* *Peroxisome biogenesis disorder 6B, 614871*
PEX11B	*100%*	*100%*	*Peroxisome biogenesis disorder 14B, 614920*
PEX12	*100%*	*100%*	*Peroxisome biogenesis disorder 3B, 266510* *Peroxisome biogenesis disorder 3A (Zellweger), 614859*
PEX13	*100%*	*100%*	*Peroxisome biogenesis disorder 11A (Zellweger), 614883* *Peroxisome biogenesis disorder 11B, 614885*
PEX14	*100%*	*100%*	*Peroxisome biogenesis disorder 13A (Zellweger), 614887*
PEX16	*100%*	*100%*	*Peroxisome biogenesis disorder 8B, 614877* *Peroxisome biogenesis disorder 8A (Zellweger), 614876*
PEX19	*100%*	*100%*	*Peroxisome biogenesis disorder 12A (Zellweger), 614886*
PEX2	*100%*	*100%*	*Peroxisome biogenesis disorder 5A (Zellweger), 614866* *Peroxisome biogenesis disorder 5B, 614867*
PEX26	*100%*	*100%*	*Peroxisome biogenesis disorder 7B, 614873* *Peroxisome biogenesis disorder 7A (Zellweger), 614872*
PEX3	*100%*	*100%*	*Peroxisome biogenesis disorder 10A (Zellweger), 614882* *?Peroxisome biogenesis disorder 10B, 617370*
PEX5	*100%*	*100%*	*Peroxisome biogenesis disorder 2B, 202370* *Peroxisome biogenesis disorder 2A (Zellweger), 214110* *Rhizomelic chondrodysplasia punctata, type 5, 616716*
PEX6	*100%*	*100%*	*Peroxisome biogenesis disorder 4B, 614863* *Peroxisome biogenesis disorder 4A (Zellweger), 614862* *Heimler syndrome 2, 616617*
PEX7	*91%*	*91%*	*Rhizomelic chondrodysplasia punctata, type 1, 215100* *Peroxisome biogenesis disorder 9B, 614879*
PFKM	*100%*	*100%*	*Glycogen storage disease VII, 232800*
PGAM2	*100%*	*100%*	*Glycogen storage disease X, 261670*

PGAP1	*100%*	*100%*	*Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802*
PGAP2	*100%*	*100%*	*Hyperphosphatasia with impaired intellectual development syndrome 3, 614207*
PGAP3	*100%*	*100%*	*Hyperphosphatasia with impaired intellectual development syndrome 4, 615716*
PGM1	*94%*	*94%*	*Congenital disorder of glycosylation, type It, 614921*
PGM3	*100%*	*100%*	*Immunodeficiency 23, 615816*
PHGDH	*100%*	*100%*	*Neu-Laxova syndrome 1, 256520* *Phosphoglycerate dehydrogenase deficiency, 601815*
PHKB	*100%*	*100%*	*Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750*
PHKG2	*100%*	*100%*	*Glycogen storage disease IXc, 613027*
PHOX2A	*100%*	*100%*	*Fibrosis of extraocular muscles, congenital, 2, 602078*
PHYH	*100%*	*100%*	*Refsum disease, 266500*
PI4KA	*100%*	*100%*	*Spastic paraplegia 84, autosomal recessive, 619621* *Gastrointestinal defects and immunodeficiency syndrome 2, 619708* *Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531*
PIBF1	*100%*	*100%*	*Joubert syndrome 33, 617767*
PIEZO1	*100%*	*100%*	*Lymphatic malformation 6, 616843* *Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380*
PIEZO2	*100%*	*100%*	*Arthrogryposis, distal, type 5, 108145* *Arthrogryposis, distal, with impaired proprioception and touch, 617146* *Arthrogryposis, distal, type 3, 114300* *?Marden-Walker syndrome, 248700*
PIGB	*100%*	*100%*	*Developmental and epileptic encephalopathy 80, 618580*
PIGC	*100%*	*100%*	*Glycosylphosphatidylinositol biosynthesis defect 16, 617816*
PIGG	*100%*	*100%*	*Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917*
PIGH	*81%*	*75%*	*Glycosylphosphatidylinositol biosynthesis defect 17, 618010*
PIGK	*100%*	*100%*	*Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879*
PIGL	*100%*	*100%*	*CHIME syndrome, 280000*
PIGM	*100%*	*100%*	*Glycosylphosphatidylinositol deficiency, 610293*
PIGN	*100%*	*100%*	*Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080*
PIGO	*100%*	*100%*	*Hyperphosphatasia with impaired intellectual development syndrome 2, 614749*
PIGP	*100%*	*100%*	*Developmental and epileptic encephalopathy 55, 617599*
PIGQ	*100%*	*100%*	*Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548*
PIGS	*100%*	*100%*	*Developmental and epileptic encephalopathy 95, 618143*

PIGT	*100%*	*100%*	*?Paroxysmal nocturnal hemoglobinuria 2, 615399* *Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398*
PIGU	*100%*	*100%*	*Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590*
PIGV	*100%*	*100%*	*Hyperphosphatasia with impaired intellectual development syndrome 1, 239300*
PIGW	*100%*	*100%*	*Glycosylphosphatidylinositol biosynthesis defect 11, 616025*
PIGY	*100%*	*100%*	*Hyperphosphatasia with impaired intellectual development syndrome 6, 616809*
PIK3C2A	*100%*	*100%*	*Oculoskeletodental syndrome, 618440*
PIK3R1	*100%*	*100%*	*Immunodeficiency 36, 616005* *?Agammaglobulinemia 7, autosomal recessive, 615214* *SHORT syndrome, 269880*
PIK3R5	*100%*	*100%*	*Ataxia-oculomotor apraxia 3, 615217*
PINK1	*100%*	*100%*	*Parkinson disease 6, early onset, 605909*
PIP5K1C	*100%*	*100%*	*Lethal congenital contractural syndrome 3, 611369*
PJVK	*100%*	*100%*	*Deafness, autosomal recessive 59, 610220*
PKD1L1	*100%*	*100%*	*Heterotaxy, visceral, 8, autosomal, 617205*
PKHD1	*100%*	*100%*	*Polycystic kidney disease 4, with or without hepatic disease, 263200*
PKLR	*100%*	*100%*	*Adenosine triphosphate, elevated, of erythrocytes, 102900* *Pyruvate kinase deficiency, 266200*
PKP1	*100%*	*100%*	*Ectodermal dysplasia/skin fragility syndrome, 604536*
PLA2G6	*100%*	*100%*	*Parkinson disease 14, autosomal recessive, 612953* *Neurodegeneration with brain iron accumulation 2B, 610217* *Infantile neuroaxonal dystrophy 1, 256600*
PLA2G7	*100%*	*100%*	*Platelet-activating factor acetylhydrolase deficiency, 614278*
PLAA	*100%*	*100%*	*Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527*
PLCB1	*100%*	*100%*	*Developmental and epileptic encephalopathy 12, 613722*
PLCB4	*100%*	*100%*	*Auriculocondylar syndrome 2, 614669*
PLCD1	*100%*	*100%*	*Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600*
PLCE1	*100%*	*100%*	*Nephrotic syndrome, type 3, 610725*
PLD1	*100%*	*100%*	*Cardiac valvular dysplasia 1, 212093*
PLEC	*100%*	*100%*	*?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487* *Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670* *Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138* *Epidermolysis bullosa simplex 5A, Ogna type, 131950* *Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723*
PLEKHG2	*100%*	*100%*	*Leukodystrophy and acquired microcephaly with or without dystonia, 616763*

PLEKHG5	*100%*	*100%*	*Spinal muscular atrophy, distal, autosomal recessive, 4, 611067* *Charcot-Marie-Tooth disease, recessive intermediate C, 615376*
PLEKHM1	*100%*	*100%*	*?Osteopetrosis, autosomal recessive 6, 611497* *Osteopetrosis, autosomal dominant 3, 618107*
PLG	*100%*	*100%*	*Dysplasminogenemia, 217090* *Angioedema, hereditary, 4, 619360* *Plasminogen deficiency, type I, 217090*
PLK4	*100%*	*100%*	*Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171*
PLOD1	*100%*	*100%*	*Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400*
PLOD2	*100%*	*100%*	*Bruck syndrome 2, 609220*
PLOD3	*100%*	*100%*	*Lysyl hydroxylase 3 deficiency, 612394*
PLPBP	*100%*	*100%*	*Epilepsy, early-onset, vitamin B6-dependent, 617290*
PLVAP	*100%*	*100%*	*Diarrhea 10, protein-losing enteropathy type, 618183*
PMM2	*100%*	*100%*	*Congenital disorder of glycosylation, type Ia, 212065*
PMPCA	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive 2, 213200*
PMPCB	*100%*	*100%*	*Multiple mitochondrial dysfunctions syndrome 6, 617954*
PMS2	*100%*	*100%*	*Lynch syndrome 4, 614337* *Mismatch repair cancer syndrome 4, 619101*
PNKP	*100%*	*100%*	*?Charcot-Marie-Tooth disease, type 2B2, 605589* *Ataxia-oculomotor apraxia 4, 616267* *Microcephaly, seizures, and developmental delay, 613402*
PNLIP	*100%*	*100%*	*?Pancreatic lipase deficiency, 614338*
PNP	*100%*	*100%*	*Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179*
PNPLA1	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 10, 615024*
PNPLA2	*100%*	*100%*	*Neutral lipid storage disease with myopathy, 610717*
PNPLA6	*100%*	*100%*	*Spastic paraplegia 39, autosomal recessive, 612020* *Oliver-McFarlane syndrome, 275400* *?Laurence-Moon syndrome, 245800* *Boucher-Neuhauser syndrome, 215470*
PNPLA8	*100%*	*100%*	*?Mitochondrial myopathy with lactic acidosis, 251950*
PNPO	*100%*	*100%*	*Pyridoxamine 5'-phosphate oxidase deficiency, 610090*
PNPT1	*100%*	*100%*	*Spinocerebellar ataxia 25, 608703* *Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934* *Combined oxidative phosphorylation deficiency 13, 614932*
POC1A	*100%*	*100%*	*Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813*

POC1B	*100%*	*100%*	*Cone-rod dystrophy 20, 615973*
POGLUT1	*100%*	*100%*	*Dowling-Degos disease 4, 615696* *Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232*
POLE	*100%*	*100%*	*FILS syndrome, 615139* *IMAGE-I syndrome, 618336*
POLG	*100%*	*100%*	*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 dominant 1, 157640* *Progressive external ophthalmoplegia, autosomal recessive 1, 258450*
POLH	*100%*	*100%*	*Xeroderma pigmentosum, variant type, 278750*
POLR1C	*83%*	*83%*	*Leukodystrophy, hypomyelinating, 11, 616494* *Treacher Collins syndrome 3, 248390*
POLR1D	*100%*	*100%*	*Treacher Collins syndrome 2, 613717*
POLR3A	*100%*	*100%*	*Wiedemann-Rautenstrauch syndrome, 264090* *Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694*
POLR3B	*100%*	*100%*	*Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381* *Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742*
POMC	*100%*	*100%*	*Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734*
POMGNT1	*100%*	*100%*	*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157* *Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151* *Retinitis pigmentosa 76, 617123* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280*
POMGNT2	*100%*	*100%*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830* *Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135*
POMK	*100%*	*100%*	*?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094* *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249*
POMP	*100%*	*100%*	*Proteasome-associated autoinflammatory syndrome 2, 618048* *Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952*

POMT1	*100%*	*100%*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670* *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308* *Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155*
POMT2	*100%*	*100%*	*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 impaired intellectual development), type B, 2, 613156*
POP1	*100%*	*100%*	*Anauxetic dysplasia 2, 617396*
POR	*100%*	*100%*	*Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750* *Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571*
POU1F1	*100%*	*100%*	*Pituitary hormone deficiency, combined or isolated, 1, 613038*
PPA2	*100%*	*100%*	*?Sudden cardiac failure, alcohol-induced, 617223* *Sudden cardiac failure, infantile, 617222*
PPCS	*100%*	*100%*	*Cardiomyopathy, dilated, 2C, 618189*
PPIB	*100%*	*100%*	*Osteogenesis imperfecta, type IX, 259440*
PPIL1	*100%*	*100%*	*Pontocerebellar hypoplasia, type 14, 619301*
PPIP5K2	*100%*	*100%*	*Deafness, autosomal recessive 100, 618422*
PPM1K	*100%*	*100%*	*?Maple syrup urine disease, mild variant, 615135*
PPP1R15B	*100%*	*100%*	*Microcephaly, short stature, and impaired glucose metabolism 2, 616817*
PPP1R21	*100%*	*100%*	*Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383*
PPP2R3C	*100%*	*100%*	*Spermatogenic failure 36, 618420* *Myoectodermal gonadal dysgenesis syndrome, 618419*
PPT1	*90%*	*90%*	*Ceroid lipofuscinosis, neuronal, 1, 256730*
PRCD	*100%*	*100%*	*Retinitis pigmentosa 36, 610599*
PRDM12	*96%*	*92%*	*Neuropathy, hereditary sensory and autonomic, type VIII, 616488*
PRDM13	*100%*	*100%*	*Pontocerebellar hypoplasia, type 17, 619909* *Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761*
PRDM5	*100%*	*100%*	*Brittle cornea syndrome 2, 614170*
PRDX1	*100%*	*100%*	*Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400*
PREPL	*100%*	*100%*	*Myasthenic syndrome, congenital, 22, 616224*

PRF1	*100%*	*100%*	*Hemophagocytic lymphohistiocytosis, familial, 2, 603553* *Aplastic anemia, 609135* *Lymphoma, non-Hodgkin, 605027*
PRG4	*100%*	*100%*	*Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250*
PRICKLE1	*100%*	*100%*	*Epilepsy, progressive myoclonic 1B, 612437*
PRKCD	*100%*	*100%*	*Autoimmune lymphoproliferative syndrome, type III, 615559*
PRKDC	*100%*	*100%*	*Immunodeficiency 26, with or without neurologic abnormalities, 615966*
PRKN	*92%*	*91%*	*Adenocarcinoma of lung, somatic, 211980* *Parkinson disease, juvenile, type 2, 600116* *Ovarian cancer, somatic, 167000*
PRKRA	*100%*	*100%*	*Dystonia 16, 612067*
PRMT7	*100%*	*100%*	*Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157*
PROC	*100%*	*100%*	*Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860* *Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304*
PRODH	*100%*	*100%*	*Hyperprolinemia, type I, 239500*
PROM1	*100%*	*100%*	*Macular dystrophy, retinal, 2, 608051* *Retinitis pigmentosa 41, 612095* *Stargardt disease 4, 603786* *Cone-rod dystrophy 12, 612657*
PROP1	*100%*	*100%*	*Pituitary hormone deficiency, combined, 2, 262600*
PRORP	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 54, 619737*
PROS1	*100%*	*100%*	*Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514* *Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336*
PRSS12	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 1, 249500*
PRSS56	*100%*	*100%*	*Microphthalmia, isolated 6, 613517*
PRUNE1	*93%*	*93%*	*Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481*
PRX	*100%*	*100%*	*Charcot-Marie-Tooth disease, type 4F, 614895* *Dejerine-Sottas disease, 145900*
PSAP	*100%*	*100%*	*Combined SAP deficiency, 611721* *Krabbe disease, atypical, 611722* *Metachromatic leukodystrophy due to SAP-b deficiency, 249900* *Gaucher disease, atypical, 610539*
PSAT1	*100%*	*100%*	*Neu-Laxova syndrome 2, 616038* *?Phosphoserine aminotransferase deficiency, 610992*
PSMB4	*100%*	*100%*	*?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591*

PSMB8	*100%*	*100%*	*Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040*
PSMB9	*100%*	*100%*	*?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591*
PSMC3IP	*100%*	*100%*	*Ovarian dysgenesis 3, 614324*
PSPH	*100%*	*100%*	*Phosphoserine phosphatase deficiency, 614023*
PTF1A	*100%*	*100%*	*Pancreatic and cerebellar agenesis, 609069* *Pancreatic agenesis 2, 615935*
PTH1R	*100%*	*100%*	*Metaphyseal chondrodysplasia, Murk Jansen type, 156400* *Eiken syndrome, 600002* *Failure of tooth eruption, primary, 125350* *Chondrodysplasia, Blomstrand type, 215045*
PTPN14	*100%*	*100%*	*Choanal atresia and lymphedema, 613611*
PTPN23	*100%*	*100%*	*Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890*
PTPRC	*100%*	*100%*	*Immunodeficiency 105, severe combined, 619924*
PTPRO	*100%*	*99%*	*Nephrotic syndrome, type 6, 614196*
PTPRQ	*92%*	*92%*	*Deafness, autosomal dominant 73, 617663* *Deafness, autosomal recessive 84A, 613391*
PTRH2	*100%*	*100%*	*Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263*
PTS	*100%*	*100%*	*Hyperphenylalaninemia, BH4-deficient, A, 261640*
PUS1	*100%*	*100%*	*Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462*
PUS3	*100%*	*100%*	*Neurodevelopmental disorder with microcephaly and gray sclerae, 617051*
PUS7	*100%*	*100%*	*Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342*
PXDN	*100%*	*100%*	*Anterior segment dysgenesis 7, with sclerocornea, 269400*
PYCR1	*100%*	*100%*	*Cutis laxa, autosomal recessive, type IIIB, 614438* *Cutis laxa, autosomal recessive, type IIB, 612940*
PYCR2	*100%*	*100%*	*Leukodystrophy, hypomyelinating, 10, 616420*
PYGL	*100%*	*100%*	*Glycogen storage disease VI, 232700*
PYGM	*100%*	*100%*	*McArdle disease, 232600*
PYROXD1	*100%*	*100%*	*Myopathy, myofibrillar, 8, 617258*
QARS1	*100%*	*100%*	*Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760*
QDPR	*100%*	*100%*	*Hyperphenylalaninemia, BH4-deficient, C, 261630*
RAB18	*100%*	*100%*	*Warburg micro syndrome 3, 614222*
RAB23	*100%*	*100%*	*Carpenter syndrome, 201000*
RAB27A	*100%*	*100%*	*Griscelli syndrome, type 2, 607624*
RAB28	*100%*	*100%*	*Cone-rod dystrophy 18, 615374*

RAB33B	*100%*	*100%*	*Smith-McCort dysplasia 2, 615222*
RAB3GAP1	*99%*	*99%*	*Martsolf syndrome 2, 619420* *Warburg micro syndrome 1, 600118*
RAB3GAP2	*100%*	*100%*	*Martsolf syndrome 1, 212720* *Warburg micro syndrome 2, 614225*
RAD50	*100%*	*100%*	*Nijmegen breakage syndrome-like disorder, 613078*
RAD51C	*100%*	*100%*	*Fanconi anemia, complementation group O, 613390*
RAG1	*100%*	*100%*	*Omenn syndrome, 603554* *Severe combined immunodeficiency, B cell-negative, 601457* *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*
RAG2	*100%*	*100%*	*Severe combined immunodeficiency, B cell-negative, 601457* *Combined cellular and humoral immune defects with granulomas, 233650* *Omenn syndrome, 603554*
RALGAPA1	*100%*	*100%*	*Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797*
RAPSN	*100%*	*100%*	*Fetal akinesia deformation sequence 2, 618388* *Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326*
RARB	*100%*	*100%*	*Microphthalmia, syndromic 12, 615524*
RARS1	*94%*	*94%*	*Leukodystrophy, hypomyelinating, 9, 616140*
RARS2	*100%*	*100%*	*Pontocerebellar hypoplasia, type 6, 611523*
RASGRP1	*100%*	*100%*	*Immunodeficiency 64, 618534*
RAX	*100%*	*100%*	*Microphthalmia, syndromic 16, 611038*
RBBP8	*100%*	*100%*	*Seckel syndrome 2, 606744* *Jawad syndrome, 251255* *Pancreatic carcinoma, somatic,*
RBCK1	*100%*	*100%*	*Polyglucosan body myopathy 1 with or without immunodeficiency, 615895*
RBM8A	*100%*	*100%*	*Thrombocytopenia-absent radius syndrome, 274000*
RBP3	*100%*	*100%*	*?Retinitis pigmentosa 66, 615233*
RBP4	*100%*	*100%*	*Microphthalmia, isolated, with coloboma 10, 616428* *Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147*
RCBTB1	*100%*	*100%*	*Retinal dystrophy with or without extraocular anomalies, 617175*
RD3	*100%*	*100%*	*Leber congenital amaurosis 12, 610612*
RDH11	*100%*	*100%*	*?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108*
RDH12	*100%*	*100%*	*Leber congenital amaurosis 13, 612712*

RDH5	*100%*	*100%*	*Fundus albipunctatus, 136880*
RDX	*100%*	*100%*	*Deafness, autosomal recessive 24, 611022*
RECQL4	*100%*	*100%*	*Baller-Gerold syndrome, 218600* *Rothmund-Thomson syndrome, type 2, 268400* *RAPADILINO syndrome, 266280*
REEP2	*100%*	*100%*	*?Spastic paraplegia 72, autosomal dominant, 615625* *?Spastic paraplegia 72, autosomal recessive, 615625*
REEP6	*100%*	*100%*	*Retinitis pigmentosa 77, 617304*
RELB	*100%*	*100%*	*?Immunodeficiency 53, 617585*
RELN	*100%*	*100%*	*Lissencephaly 2 (Norman-Roberts type), 257320*
REN	*100%*	*100%*	*Renal tubular dysgenesis, 267430* *Tubulointerstitial kidney disease, autosomal dominant, 4, 613092*
REPS1	*100%*	*100%*	*?Neurodegeneration with brain iron accumulation 7, 617916*
RETREG1	*100%*	*100%*	*Neuropathy, hereditary sensory and autonomic, type IIB, 613115*
RFC1	*100%*	*100%*	*Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575*
RFT1	*100%*	*100%*	*Congenital disorder of glycosylation, type In, 612015*
RFWD3	*100%*	*100%*	*?Fanconi anemia, complementation group W, 617784*
RFX5	*100%*	*100%*	*Bare lymphocyte syndrome, type II, complementation group C, 209920* *Bare lymphocyte syndrome, type II, complementation group E, 209920*
RFX6	*100%*	*100%*	*Mitchell-Riley syndrome, 615710*
RFXANK	*100%*	*100%*	*Bare lymphocyte syndrome, type II, complementation group B, 209920*
RFXAP	*100%*	*100%*	*Bare lymphocyte syndrome, type II, complementation group D, 209920*
RGR	*100%*	*100%*	*Retinitis pigmentosa 44, 613769*
RHO	*100%*	*100%*	*Night blindness, congenital stationary, autosomal dominant 1, 610445* *Retinitis pigmentosa 4, autosomal dominant or recessive, 613731* *Retinitis punctata albescens, 136880*
RIMS2	*100%*	*100%*	*Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970*
RIN2	*100%*	*100%*	*Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075*
RINT1	*100%*	*100%*	*Infantile liver failure syndrome 3, 618641*
RIPK1	*100%*	*100%*	*Immunodeficiency 57 with autoinflammation, 618108* *Autoinflammation with episodic fever and lymphadenopathy, 618852*
RIPK4	*100%*	*100%*	*CHAND syndrome, 214350* *Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650*
RIPOR2	*100%*	*100%*	*Deafness, autosomal dominant 21, 607017* *?Deafness, autosomal recessive 104, 616515*

RIPPLY2	*100%*	*100%*	*?Spondylocostal dysostosis 6, 616566*
RLBP1	*100%*	*100%*	*Bothnia retinal dystrophy, 607475* *Newfoundland rod-cone dystrophy, 607476* *Retinitis punctata albescens, 136880* *Fundus albipunctatus, 136880*
RMND1	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 11, 614922*
RMRP	*NC*	*NC*	*Anauxetic dysplasia 1, 607095* *Metaphyseal dysplasia without hypotrichosis, 250460* *Cartilage-hair hypoplasia, 250250*
RNASEH1	*100%*	*100%*	*Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479*
RNASEH2A	*100%*	*100%*	*Aicardi-Goutieres syndrome 4, 610333*
RNASEH2B	*91%*	*91%*	*Aicardi-Goutieres syndrome 2, 610181*
RNASEH2C	*100%*	*100%*	*Aicardi-Goutieres syndrome 3, 610329*
RNASET2	*100%*	*100%*	*Leukoencephalopathy, cystic, without megalencephaly, 612951*
RNF168	*100%*	*100%*	*RIDDLE syndrome, 611943*
RNF216	*100%*	*100%*	*Cerebellar ataxia and hypogonadotropic hypogonadism, 212840*
RNPC3	*100%*	*100%*	*Pituitary hormone deficiency, combined or isolated, 7, 618160*
ROBO3	*100%*	*100%*	*Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313*
ROGDI	*100%*	*100%*	*Kohlschutter-Tonz syndrome, 226750*
ROR2	*100%*	*100%*	*Brachydactyly, type B1, 113000* *Robinow syndrome, autosomal recessive, 268310*
RORC	*100%*	*100%*	*Immunodeficiency 42, 616622*
RP1	*100%*	*100%*	*Retinitis pigmentosa 1, 180100*
RP1L1	*100%*	*100%*	*Occult macular dystrophy, 613587* *Retinitis pigmentosa 88, 618826*
RPE65	*100%*	*100%*	*Retinitis pigmentosa 20, 613794* *Retinitis pigmentosa 87 with choroidal involvement, 618697* *Leber congenital amaurosis 2, 204100*
RPGRIP1	*100%*	*100%*	*Cone-rod dystrophy 13, 608194* *Leber congenital amaurosis 6, 613826*
RPGRIP1L	*100%*	*100%*	*Joubert syndrome 7, 611560* *Meckel syndrome 5, 611561* *?COACH syndrome 3, 619113*
RPIA	*100%*	*100%*	*Ribose 5-phosphate isomerase deficiency, 608611*

RRM2B	*100%*	*100%*	*Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075* *Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075* *Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315* *Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077*
RSPH1	*100%*	*100%*	*Ciliary dyskinesia, primary, 24, 615481*
RSPH3	*100%*	*100%*	*Ciliary dyskinesia, primary, 32, 616481*
RSPH4A	*100%*	*100%*	*Ciliary dyskinesia, primary, 11, 612649*
RSPH9	*100%*	*100%*	*Ciliary dyskinesia, primary, 12, 612650*
RSPO1	*100%*	*100%*	*Palmoplantar hyperkeratosis and true hermaphroditism, 610644* *Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644*
RSPO2	*100%*	*100%*	*?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022* *Tetraamelia syndrome 2, 618021*
RSPO4	*100%*	*100%*	*Anonychia congenita, 206800*
RSPRY1	*100%*	*100%*	*Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723*
RSRC1	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 70, 618402*
RTEL1	*100%*	*100%*	*Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373* *Dyskeratosis congenita, autosomal dominant 4, 615190* *Dyskeratosis congenita, autosomal recessive 5, 615190*
RTN4IP1	*100%*	*100%*	*Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732*
RTTN	*100%*	*100%*	*Microcephaly, short stature, and polymicrogyria with seizures, 614833*
RUBCN	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive 15, 615705*
RUSC2	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 61, 617773*
RXYLT1	*100%*	*100%*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041*
RYR1	*100%*	*100%*	*Neuromuscular disease, congenital, with uniform type 1 fiber, 117000* *Central core disease, 117000* *King-Denborough syndrome, 619542* *Minicore myopathy with external ophthalmoplegia, 255320*
S1PR2	*100%*	*100%*	*Deafness, autosomal recessive 68, 610419*
SACS	*100%*	*100%*	*Spastic ataxia, Charlevoix-Saguenay type, 270550*
SAG	*100%*	*100%*	*Retinitis pigmentosa 47, 613758* *Oguchi disease-1, 258100*

SAMD9	*100%*	*100%*	*Tumoral calcinosis, familial, normophosphatemic, 610455* *Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041* *MIRAGE syndrome, 617053*
SAMHD1	*100%*	*100%*	*?Chilblain lupus 2, 614415* *Aicardi-Goutieres syndrome 5, 612952*
SAR1B	*100%*	*100%*	*Chylomicron retention disease, 246700*
SARS1	*100%*	*100%*	*Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709*
SARS2	*100%*	*100%*	*Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845*
SASH1	*100%*	*100%*	*Dyschromatosis universalis hereditaria 1, 127500* *?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373*
SASS6	*100%*	*100%*	*Microcephaly 14, primary, autosomal recessive, 616402*
SBDS	*100%*	*100%*	*Shwachman-Diamond syndrome 1, 260400*
SBF1	*100%*	*100%*	*Charcot-Marie-Tooth disease, type 4B3, 615284*
SBF2	*100%*	*100%*	*Charcot-Marie-Tooth disease, type 4B2, 604563*
SC5D	*100%*	*100%*	*Lathosterolosis, 607330*
SCAPER	*100%*	*100%*	*Intellectual developmental disorder and retinitis pigmentosa, 618195*
SCARB2	*100%*	*100%*	*Epilepsy, progressive myoclonic 4, with or without renal failure, 254900*
SCARF2	*100%*	*100%*	*Van den Ende-Gupta syndrome, 600920*
SCN1B	*100%*	*100%*	*Generalized epilepsy with febrile seizures plus, type 1, 604233* *Developmental and epileptic encephalopathy 52, 617350* *Cardiac conduction defect, nonspecific, 612838* *Atrial fibrillation, familial, 13, 615377* *Brugada syndrome 5, 612838*
SCN4A	*100%*	*100%*	*Paramyotonia congenita, 168300* *Hypokalemic periodic paralysis, type 2, 613345* *Myotonia congenita, atypical, acetazolamide-responsive, 608390* *Myasthenic syndrome, congenital, 16, 614198* *Hyperkalemic periodic paralysis, type 2, 170500*
SCN9A	*100%*	*100%*	*Erythermalgia, primary, 133020* *Insensitivity to pain, congenital, 243000* *Small fiber neuropathy, 133020* *Paroxysmal extreme pain disorder, 167400* *Neuropathy, hereditary sensory and autonomic, type IID, 243000*

SCNN1A	*100%*	*100%*	*Pseudohypoaldosteronism, type I, 264350* *?Liddle syndrome 3, 618126* *Bronchiectasis with or without elevated sweat chloride 2, 613021*
SCNN1B	*100%*	*100%*	*Bronchiectasis with or without elevated sweat chloride 1, 211400* *Pseudohypoaldosteronism, type I, 264350* *Liddle syndrome 1, 177200*
SCNN1G	*100%*	*100%*	*Bronchiectasis with or without elevated sweat chloride 3, 613071* *Pseudohypoaldosteronism, type I, 264350* *Liddle syndrome 2, 618114*
SCO1	*100%*	*100%*	*Mitochondrial complex IV deficiency, nuclear type 4, 619048*
SCO2	*100%*	*100%*	*Myopia 6, 608908* *Mitochondrial complex IV deficiency, nuclear type 2, 604377*
SCP2	*100%*	*100%*	*?Leukoencephalopathy with dystonia and motor neuropathy, 613724*
SCYL1	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive 21, 616719*
SDCCAG8	*100%*	*100%*	*Senior-Loken syndrome 7, 613615* *Bardet-Biedl syndrome 16, 615993*
SDHA	*100%*	*100%*	*Cardiomyopathy, dilated, 1GG, 613642* *Mitochondrial complex II deficiency, nuclear type 1, 252011* *Neurodegeneration with ataxia and late-onset optic atrophy, 619259* *Paragangliomas 5, 614165*
SDHAF1	*100%*	*100%*	*Mitochondrial complex II deficiency, nuclear type 2, 619166*
SDHB	*100%*	*100%*	*Paragangliomas 4, 115310* *Mitochondrial complex II deficiency, nuclear type 4, 619224* *Gastrointestinal stromal tumor, 606764* *Pheochromocytoma, 171300* *Paraganglioma and gastric stromal sarcoma, 606864*
SDHD	*79%*	*79%*	*Paragangliomas 1, with or without deafness, 168000* *Paraganglioma and gastric stromal sarcoma, 606864* *Mitochondrial complex II deficiency, nuclear type 3, 619167* *Pheochromocytoma, 171300*
SDR9C7	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 13, 617574*
SEC23A	*100%*	*100%*	*Craniolenticulosutural dysplasia, 607812*
SEC23B	*100%*	*100%*	*?Cowden syndrome 7, 616858* *Dyserythropoietic anemia, congenital, type II, 224100*
SEC24D	*100%*	*100%*	*Cole-Carpenter syndrome 2, 616294*

SEC31A	*100%*	*100%*	*?Halperin-Birk syndrome, 618651*
SECISBP2	*100%*	*100%*	*Thyroid hormone metabolism, abnormal, 1, 609698* *Thyroid hormone metabolism, abnormal, 609698*
SELENON	*93%*	*91%*	*Myopathy, congenital, with fiber-type disproportion, 255310* *Muscular dystrophy, rigid spine, 1, 602771*
SEMA4A	*100%*	*100%*	*Retinitis pigmentosa 35, 610282* *Cone-rod dystrophy 10, 610283*
SEPSECS	*100%*	*100%*	*Pontocerebellar hypoplasia type 2D, 613811*
SERAC1	*100%*	*100%*	*3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739*
SERPINA1	*100%*	*100%*	*Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490* *Emphysema due to AAT deficiency, 613490* *Emphysema-cirrhosis, due to AAT deficiency, 613490*
SERPINA6	*100%*	*100%*	*Corticosteroid-binding globulin deficiency, 611489*
SERPINB7	*100%*	*100%*	*Palmoplantar keratoderma, Nagashima type, 615598*
SERPINB8	*100%*	*100%*	*Peeling skin syndrome 5, 617115*
SERPINC1	*100%*	*100%*	*Thrombophilia 7 due to antithrombin III deficiency, 613118*
SERPINE1	*100%*	*100%*	*Plasminogen activator inhibitor-1 deficiency, 613329*
SERPINF1	*100%*	*100%*	*Osteogenesis imperfecta, type VI, 613982*
SERPINF2	*100%*	*100%*	*Alpha-2-plasmin inhibitor deficiency, 262850*
SERPING1	*100%*	*100%*	*Angioedema, hereditary, 1 and 2, 106100* *Complement component 4, partial deficiency of, 120790*
SERPINH1	*100%*	*100%*	*Osteogenesis imperfecta, type X, 613848*
SETX	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002* *Amyotrophic lateral sclerosis 4, juvenile, 602433*
SFRP4	*100%*	*100%*	*Pyle disease, 265900*
SFTPB	*100%*	*100%*	*Surfactant metabolism dysfunction, pulmonary, 1, 265120*
SFXN4	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 18, 615578*
SGCA	*100%*	*100%*	*Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099*
SGCB	*100%*	*100%*	*Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286*
SGCD	*100%*	*100%*	*Cardiomyopathy, dilated, 1L, 606685* *Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287*
SGCG	*100%*	*100%*	*Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700*
SGO1	*100%*	*100%*	*Chronic atrial and intestinal dysrhythmia, 616201*
SGPL1	*100%*	*100%*	*Nephrotic syndrome, type 14, 617575*

SGSH	*100%*	*100%*	*Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900*
SH3PXD2B	*100%*	*100%*	*Frank-ter Haar syndrome, 249420*
SH3TC2	*100%*	*100%*	*Charcot-Marie-Tooth disease, type 4C, 601596* *Mononeuropathy of the median nerve, mild, 613353*
SI	*99%*	*98%*	*Sucrase-isomaltase deficiency, congenital, 222900*
SIGMAR1	*100%*	*100%*	*?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726* *?Amyotrophic lateral sclerosis 16, juvenile, 614373*
SIK3	*100%*	*100%*	*?Spondyloepimetaphyseal dysplasia, Krakow type, 618162*
SIL1	*100%*	*100%*	*Marinesco-Sjogren syndrome, 248800*
SIX6	*100%*	*100%*	*Optic disc anomalies with retinal and/or macular dystrophy, 212550*
SKIV2L	*100%*	*100%*	*Trichohepatoenteric syndrome 2, 614602*
TTC37	*100%*	*100%*	*Trichohepatoenteric syndrome 1, 222470*
SLC10A7	*100%*	*100%*	*Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363*
SLC11A2	*100%*	*100%*	*Anemia, hypochromic microcytic, with iron overload 1, 206100*
SLC12A1	*96%*	*96%*	*Bartter syndrome, type 1, 601678*
SLC12A2	*100%*	*100%*	*Kilquist syndrome, 619080* *Delpire-McNeill syndrome, 619083* *Deafness, autosomal dominant 78, 619081*
SLC12A3	*100%*	*100%*	*Gitelman syndrome, 263800*
SLC12A5	*100%*	*100%*	*Developmental and epileptic encephalopathy 34, 616645*
SLC12A6	*100%*	*100%*	*Agenesis of the corpus callosum with peripheral neuropathy, 218000* *Charcot-Marie-Tooth disease, axonal, type 2II, 620068*
SLC13A3	*100%*	*100%*	*Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384*
SLC13A5	*100%*	*100%*	*Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905*
SLC16A1	*100%*	*100%*	*Hyperinsulinemic hypoglycemia, familial, 7, 610021* *Erythrocyte lactate transporter defect, 245340* *Monocarboxylate transporter 1 deficiency, 616095*
SLC17A5	*100%*	*100%*	*Salla disease, 604369* *Sialic acid storage disorder, infantile, 269920*
SLC18A2	*100%*	*100%*	*?Parkinsonism-dystonia, infantile, 2, 618049*
SLC18A3	*100%*	*100%*	*Myasthenic syndrome, congenital, 21, presynaptic, 617239*
SLC19A2	*100%*	*100%*	*Thiamine-responsive megaloblastic anemia syndrome, 249270*
SLC19A3	*100%*	*98%*	*Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483*
SLC1A1	*100%*	*100%*	*Dicarboxylic aminoaciduria, 222730*

SLC1A4	*100%*	*100%*	*Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657*
SLC22A12	*100%*	*100%*	*Hypouricemia, renal, 220150*
SLC22A5	*100%*	*100%*	*Carnitine deficiency, systemic primary, 212140*
SLC24A1	*100%*	*100%*	*Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830*
SLC24A4	*100%*	*100%*	*Amelogenesis imperfecta, type IIA5, 615887*
SLC24A5	*100%*	*100%*	*Albinism, oculocutaneous, type VI, 113750*
SLC25A1	*100%*	*100%*	*Combined D-2- and L-2-hydroxyglutaric aciduria, 615182* *Myasthenic syndrome, congenital, 23, presynaptic, 618197*
SLC25A12	*100%*	*100%*	*Developmental and epileptic encephalopathy 39, 612949*
SLC25A13	*100%*	*100%*	*Citrullinemia, type II, neonatal-onset, 605814* *Citrullinemia, adult-onset type II, 603471*
SLC25A15	*100%*	*100%*	*Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970*
SLC25A19	*100%*	*100%*	*Microcephaly, Amish type, 607196* *Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710*
SLC25A20	*100%*	*100%*	*Carnitine-acylcarnitine translocase deficiency, 212138*
SLC25A22	*100%*	*100%*	*Developmental and epileptic encephalopathy 3, 609304*
SLC25A26	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 28, 616794*
SLC25A3	*100%*	*100%*	*Mitochondrial phosphate carrier deficiency, 610773*
SLC25A38	*100%*	*100%*	*Anemia, sideroblastic, 2, pyridoxine-refractory, 205950*
SLC25A4	*100%*	*100%*	*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*
SLC25A42	*100%*	*100%*	*Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416*
SLC25A46	*100%*	*100%*	*Neuropathy, hereditary motor and sensory, type VIB, 616505* *Pontocerebellar hypoplasia, type 1E, 619303*
SLC26A1	*100%*	*100%*	*?Nephrolithiasis, calcium oxalate, 167030*
SLC26A2	*100%*	*100%*	*Epiphyseal dysplasia, multiple, 4, 226900* *De la Chapelle dysplasia, 256050* *Diastrophic dysplasia, 222600* *Diastrophic dysplasia, broad bone-platyspondylic variant, 222600* *Achondrogenesis Ib, 600972* *Atelosteogenesis, type II, 256050*
SLC26A3	*100%*	*100%*	*Diarrhea 1, secretory chloride, congenital, 214700*

SLC26A4	*100%*	*100%*	*Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791* *Pendred syndrome, 274600*
SLC27A4	*100%*	*100%*	*Ichthyosis prematurity syndrome, 608649*
SLC29A3	*100%*	*100%*	*Histiocytosis-lymphadenopathy plus syndrome, 602782*
SLC2A1	*100%*	*100%*	*Dystonia 9, 601042* *GLUT1 deficiency syndrome 1, infantile onset, severe, 606777* *Stomatin-deficient cryohydrocytosis with neurologic defects, 608885* *GLUT1 deficiency syndrome 2, childhood onset, 612126*
SLC2A10	*100%*	*100%*	*Arterial tortuosity syndrome, 208050*
SLC2A2	*100%*	*100%*	*Fanconi-Bickel syndrome, 227810*
SLC2A9	*100%*	*100%*	*Hypouricemia, renal, 2, 612076*
SLC30A10	*100%*	*100%*	*Hypermanganesemia with dystonia 1, 613280*
SLC30A9	*100%*	*100%*	*Birk-Landau-Perez syndrome, 617595*
SLC33A1	*100%*	*100%*	*Spastic paraplegia 42, autosomal dominant, 612539* *Congenital cataracts, hearing loss, and neurodegeneration, 614482*
SLC34A1	*100%*	*100%*	*?Fanconi renotubular syndrome 2, 613388* *Hypercalcemia, infantile, 2, 616963* *Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286*
SLC34A2	*100%*	*100%*	*Pulmonary alveolar microlithiasis, 265100*
SLC34A3	*100%*	*100%*	*Hypophosphatemic rickets with hypercalciuria, 241530*
SLC35A1	*100%*	*100%*	*Congenital disorder of glycosylation, type IIf, 603585*
SLC35A3	*98%*	*93%*	*Arthrogryposis, impaired intellectual development, and seizures, 615553*
SLC35C1	*100%*	*100%*	*Congenital disorder of glycosylation, type IIc, 266265*
SLC35D1	*100%*	*100%*	*Schneckenbecken dysplasia, 269250*
SLC37A4	*100%*	*100%*	*Glycogen storage disease Ib, 232220* *Congenital disorder of glycosylation, type IIw, 619525* *Glycogen storage disease Ic, 232240*
SLC38A8	*100%*	*100%*	*Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218*
SLC39A13	*100%*	*100%*	*Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350*
SLC39A14	*94%*	*94%*	*?Hyperostosis cranalis interna, 144755* *Hypermanganesemia with dystonia 2, 617013*
SLC39A4	*100%*	*100%*	*Acrodermatitis enteropathica, 201100*
SLC39A8	*100%*	*100%*	*Congenital disorder of glycosylation, type IIn, 616721*
SLC3A1	*96%*	*96%*	*Cystinuria, 220100*
SLC44A1	*100%*	*100%*	*Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868*

SLC45A1	*100%*	*100%*	*Intellectual developmental disorder with neuropsychiatric features, 617532*
SLC45A2	*100%*	*100%*	*Albinism, oculocutaneous, type IV, 606574*
SLC46A1	*100%*	*100%*	*Folate malabsorption, hereditary, 229050*
SLC4A1	*100%*	*100%*	*Distal renal tubular acidosis 1, 179800* *Spherocytosis, type 4, 612653* *Distal renal tubular acidosis 4 with hemolytic anemia, 611590* *Cryohydrocytosis, 185020* *Ovalocytosis, SA type, 166900*
SLC4A11	*100%*	*100%*	*Corneal endothelial dystrophy, autosomal recessive, 217700* *Corneal dystrophy, Fuchs endothelial, 4, 613268* *Corneal endothelial dystrophy and perceptive deafness, 217400*
SLC4A4	*100%*	*100%*	*Renal tubular acidosis, proximal, with ocular abnormalities, 604278*
SLC52A2	*100%*	*100%*	*Brown-Vialetto-Van Laere syndrome 2, 614707*
SLC52A3	*100%*	*100%*	*?Fazio-Londe disease, 211500* *Brown-Vialetto-Van Laere syndrome 1, 211530*
SLC5A1	*100%*	*100%*	*Glucose/galactose malabsorption, 606824*
SLC5A2	*100%*	*100%*	*Renal glucosuria, 233100*
SLC5A5	*100%*	*100%*	*Thyroid dyshormonogenesis 1, 274400*
SLC5A7	*100%*	*100%*	*Neuronopathy, distal hereditary motor, type VIIA, 158580* *Myasthenic syndrome, congenital, 20, presynaptic, 617143*
SLC6A17	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 48, 616269*
SLC6A19	*100%*	*100%*	*Iminoglycinuria, digenic, 242600* *Hartnup disorder, 234500* *Hyperglycinuria, 138500*
SLC6A3	*100%*	*100%*	*Parkinsonism-dystonia, infantile, 1, 613135*
SLC6A5	*100%*	*100%*	*Hyperekplexia 3, 614618*
SLC6A9	*100%*	*100%*	*Glycine encephalopathy with normal serum glycine, 617301*
SLC7A14	*100%*	*100%*	*Retinitis pigmentosa 68, 615725*
SLC7A7	*100%*	*100%*	*Lysinuric protein intolerance, 222700*
SLC7A9	*100%*	*100%*	*Cystinuria, 220100*
SLC9A1	*100%*	*100%*	*Lichtenstein-Knorr syndrome, 616291*
SLC9A3	*100%*	*100%*	*Diarrhea 8, secretory sodium, congenital, 616868*
SLCO2A1	*100%*	*100%*	*Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100* *Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441*
SLITRK6	*100%*	*100%*	*Deafness and myopia, 221200*

SLURP1	*100%*	*100%*	*Meleda disease, 248300*
SLX4	*100%*	*100%*	*Fanconi anemia, complementation group P, 613951*
SMARCAL1	*100%*	*100%*	*Schimke immunoosseous dysplasia, 242900*
SMARCD2	*100%*	*100%*	*Specific granule deficiency 2, 617475*
SMG9	*100%*	*100%*	*Heart and brain malformation syndrome, 616920* *Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995*
SMN1	*94%*	*94%*	*Spinal muscular atrophy-2, 253550* *Spinal muscular atrophy-4, 271150* *Spinal muscular atrophy-3, 253400* *Spinal muscular atrophy-1, 253300*
SMO	*100%*	*100%*	*Pallister-Hall-like syndrome, 241800* *Basal cell carcinoma, somatic, 605462* *Curry-Jones syndrome, somatic mosaic, 601707*
SMOC1	*100%*	*100%*	*Microphthalmia with limb anomalies, 206920*
SMOC2	*100%*	*100%*	*Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400*
SMPD1	*100%*	*100%*	*Niemann-Pick disease, type B, 607616* *Niemann-Pick disease, type A, 257200*
SMPD4	*100%*	*100%*	*Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622*
SNAI2	*100%*	*100%*	*Waardenburg syndrome, type 2D, 608890* *Piebaldism, 172800*
SNAP29	*100%*	*100%*	*Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528*
SNIP1	*100%*	*100%*	*Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501*
SNORD118	*NC*	*NC*	*Leukoencephalopathy, brain calcifications, and cysts, 614561*
SNX10	*100%*	*100%*	*Osteopetrosis, autosomal recessive 8, 615085*
SNX14	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive 20, 616354*
SOBP	*100%*	*100%*	*Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671*
SOD1	*100%*	*100%*	*Spastic tetraplegia and axial hypotonia, progressive, 618598* *Amyotrophic lateral sclerosis 1, 105400*
SORD	*93%*	*90%*	*Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912*
SOST	*100%*	*100%*	*Sclerosteosis 1, 269500* *Craniodiaphyseal dysplasia, autosomal dominant, 122860*
SOX18	*100%*	*99%*	*Hypotrichosis-lymphedema-telangiectasia syndrome, 607823* *Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940*
SP110	*100%*	*100%*	*Hepatic venoocclusive disease with immunodeficiency, 235550*

SP7	*100%*	*100%*	*Osteogenesis imperfecta, type XII, 613849*
SPAG1	*100%*	*100%*	*Ciliary dyskinesia, primary, 28, 615505*
SPARC	*100%*	*100%*	*Osteogenesis imperfecta, type XVII, 616507*
SPART	*100%*	*100%*	*Troyer syndrome, 275900*
SPATA5	*100%*	*100%*	*Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577*
SPATA5L1	*100%*	*100%*	*Deafness, autosomal recessive 119, 619615* *Neurodevelopmental disorder with hearing loss and spasticity, 619616*
SPATA7	*100%*	*100%*	*Leber congenital amaurosis 3, 604232* *Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232*
SPEG	*100%*	*100%*	*Centronuclear myopathy 5, 615959*
SPG11	*100%*	*100%*	*Amyotrophic lateral sclerosis 5, juvenile, 602099* *Charcot-Marie-Tooth disease, axonal, type 2X, 616668* *Spastic paraplegia 11, autosomal recessive, 604360*
SPG21	*100%*	*100%*	*Mast syndrome, 248900*
SPG7	*100%*	*100%*	*Spastic paraplegia 7, autosomal recessive, 607259*
SPINK5	*100%*	*100%*	*Netherton syndrome, 256500*
SPINT2	*100%*	*100%*	*Diarrhea 3, secretory sodium, congenital, syndromic, 270420*
SPR	*100%*	*100%*	*Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716*
SPRED2	*100%*	*100%*	*Noonan syndrome 14, 619745*
SPRTN	*100%*	*100%*	*Ruijs-Aalfs syndrome, 616200*
SPTA1	*100%*	*100%*	*Spherocytosis, type 3, 270970* *Elliptocytosis-2, 130600* *Pyropoikilocytosis, 266140*
SPTB	*100%*	*100%*	*Anemia, neonatal hemolytic, fatal or near-fatal, 617948* *Elliptocytosis-3, 617948* *Spherocytosis, type 2, 616649*
SPTBN2	*100%*	*100%*	*Spinocerebellar ataxia 5, 600224* *Spinocerebellar ataxia, autosomal recessive 14, 615386*
SPTBN4	*100%*	*100%*	*Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519*
SQSTM1	*100%*	*100%*	*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* *Paget disease of bone 3, 167250*
SRD5A2	*100%*	*100%*	*Pseudovaginal perineoscrotal hypospadias, 264600*

SRD5A3	*100%*	*100%*	*Kahrizi syndrome, 612713* *Congenital disorder of glycosylation, type Iq, 612379*
ST14	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 11, 602400*
ST3GAL3	*97%*	*95%*	*Developmental and epileptic encephalopathy 15, 615006* *Intellectual developmental disorder, autosomal recessive 12, 611090*
ST3GAL5	*98%*	*98%*	*Salt and pepper developmental regression syndrome, 609056*
STAC3	*100%*	*100%*	*Myopathy, congenital, Baily-Bloch, 255995*
STAMBP	*100%*	*100%*	*Microcephaly-capillary malformation syndrome, 614261*
STAR	*100%*	*100%*	*Lipoid adrenal hyperplasia, 201710*
STAT1	*96%*	*96%*	*Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162* *Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892* *Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796*
STAT2	*100%*	*100%*	*Pseudo-TORCH syndrome 3, 618886* *Immunodeficiency 44, 616636*
STAT5B	*100%*	*100%*	*Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590* *Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985* *Leukemia, acute promyelocytic, somatic, 102578*
STIL	*100%*	*100%*	*Microcephaly 7, primary, autosomal recessive, 612703*
STIM1	*100%*	*100%*	*Myopathy, tubular aggregate, 1, 160565* *Stormorken syndrome, 185070* *Immunodeficiency 10, 612783*
STK4	*100%*	*100%*	*T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868*
STN1	*100%*	*100%*	*Cerebroretinal microangiopathy with calcifications and cysts 2, 617341*
STRA6	*100%*	*100%*	*Microphthalmia, syndromic 9, 601186* *Microphthalmia, isolated, with coloboma 8, 601186*
STRADA	*100%*	*100%*	*Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087*
STRC	*100%*	*100%*	*Deafness, autosomal recessive 16, 603720*
STT3A	*100%*	*100%*	*Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714* *Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596*
STT3B	*100%*	*100%*	*Congenital disorder of glycosylation, type Ix, 615597*
STUB1	*100%*	*100%*	*Spinocerebellar ataxia 48, 618093* *Spinocerebellar ataxia, autosomal recessive 16, 615768*
STX11	*100%*	*100%*	*Hemophagocytic lymphohistiocytosis, familial, 4, 603552*
STXBP2	*100%*	*100%*	*Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101*
SUCLA2	*100%*	*100%*	*Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073*

SUCLG1	*100%*	*100%*	*Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400*
SUFU	*100%*	*100%*	*Joubert syndrome 32, 617757* *Basal cell nevus syndrome, 109400*
SULT2B1	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 14, 617571*
SUMF1	*100%*	*100%*	*Multiple sulfatase deficiency, 272200*
SUOX	*100%*	*100%*	*Sulfite oxidase deficiency, 272300*
SURF1	*100%*	*100%*	*Charcot-Marie-Tooth disease, type 4K, 616684* *Mitochondrial complex IV deficiency, nuclear type 1, 220110*
SVBP	*100%*	*100%*	*Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569*
SYNE1	*100%*	*100%*	*Arthrogryposis multiplex congenita 3, myogenic type, 618484* *Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998* *Spinocerebellar ataxia, autosomal recessive 8, 610743*
SYNE4	*100%*	*100%*	*Deafness, autosomal recessive 76, 615540*
SYNJ1	*100%*	*100%*	*Parkinson disease 20, early-onset, 615530* *Developmental and epileptic encephalopathy 53, 617389*
SYT14	*100%*	*100%*	*?Spinocerebellar ataxia, autosomal recessive 11, 614229*
SZT2	*100%*	*100%*	*Developmental and epileptic encephalopathy 18, 615476*
TAC3	*100%*	*100%*	*Hypogonadotropic hypogonadism 10 with or without anosmia, 614839*
TACO1	*100%*	*100%*	*Mitochondrial complex IV deficiency, nuclear type 8, 619052*
TACR3	*100%*	*100%*	*Hypogonadotropic hypogonadism 11 with or without anosmia, 614840*
TACSTD2	*100%*	*100%*	*Corneal dystrophy, gelatinous drop-like, 204870*
TAF13	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 60, 617432*
TAF1C	*100%*	*100%*	*No OMIM disease ID*
TAF2	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 40, 615599*
TAF6	*100%*	*100%*	*Alazami-Yuan syndrome, 617126*
TALDO1	*100%*	*100%*	*Transaldolase deficiency, 606003*
TANGO2	*100%*	*100%*	*Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878*
TAP1	*100%*	*100%*	*Bare lymphocyte syndrome, type I, 604571*
TAP2	*100%*	*100%*	*Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571*
TAPBP	*96%*	*96%*	*Bare lymphocyte syndrome, type I, 604571*
TAPT1	*100%*	*100%*	*Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897*
TARS2	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 21, 615918*
TAT	*100%*	*100%*	*Tyrosinemia, type II, 276600*
TBC1D20	*100%*	*100%*	*Warburg micro syndrome 4, 615663*

TBC1D23	*100%*	*100%*	*Pontocerebellar hypoplasia, type 11, 617695*
TBC1D24	*100%*	*100%*	*Deafness, autosomal recessive 86, 614617* *Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105* *Myoclonic epilepsy, infantile, familial, 605021* *Deafness, autosomal dominant 65, 616044* *Developmental and epileptic encephalopathy 16, 615338* *DOORS syndrome, 220500*
TBC1D7	*100%*	*100%*	*Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000*
TBCD	*100%*	*100%*	*Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193*
TBCE	*100%*	*100%*	*Kenny-Caffey syndrome, type 1, 244460* *Hypoparathyroidism-retardation-dysmorphism syndrome, 241410* *Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207*
TBCK	*100%*	*100%*	*Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900*
TBX15	*100%*	*99%*	*Cousin syndrome, 260660*
TBX19	*100%*	*100%*	*Adrenocorticotropic hormone deficiency, 201400*
TBX6	*100%*	*100%*	*Spondylocostal dysostosis 5, 122600*
TBXAS1	*100%*	*100%*	*Ghosal hematodiaphyseal syndrome, 231095*
TBXT	*100%*	*100%*	*Sacral agenesis with vertebral anomalies, 615709*
TCAP	*100%*	*100%*	*Cardiomyopathy, hypertrophic, 25, 607487* *Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954*
TCIRG1	*100%*	*100%*	*Osteopetrosis, autosomal recessive 1, 259700*
TCN2	*100%*	*100%*	*Transcobalamin II deficiency, 275350*
TCTN1	*95%*	*94%*	*Joubert syndrome 13, 614173*
TCTN2	*100%*	*100%*	*Joubert syndrome 24, 616654* *?Meckel syndrome 8, 613885*
TCTN3	*100%*	*100%*	*Joubert syndrome 18, 614815* *Orofaciodigital syndrome IV, 258860*
TDP1	*100%*	*100%*	*?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250*
TDP2	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive 23, 616949*
TDRD7	*100%*	*100%*	*Cataract 36, 613887*
TECPR2	*100%*	*100%*	*Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031*
TECR	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 14, 614020*
TECRL	*100%*	*100%*	*Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021*
TECTA	*100%*	*100%*	*Deafness, autosomal dominant 8/12, 601543* *Deafness, autosomal recessive 21, 603629*

TELO2	*100%*	*100%*	*You-Hoover-Fong syndrome, 616954*
TENM3	*100%*	*100%*	*Microphthalmia, syndromic 15, 615145* *?Microphthalmia, isolated, with coloboma 9, 615145*
TENT5A	*100%*	*100%*	*Osteogenesis imperfecta, type XVIII, 617952*
TF	*100%*	*100%*	*Atransferrinemia, 209300*
TFAM	*100%*	*100%*	*?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156*
TFG	*100%*	*100%*	*?Spastic paraplegia 57, autosomal recessive, 615658* *Hereditary motor and sensory neuropathy, Okinawa type, 604484*
TFR2	*100%*	*100%*	*Hemochromatosis, type 3, 604250*
TFRC	*100%*	*100%*	*Immunodeficiency 46, 616740*
TG	*100%*	*100%*	*Thyroid dyshormonogenesis 3, 274700*
TGDS	*100%*	*100%*	*Catel-Manzke syndrome, 616145*
TGFB1	*100%*	*100%*	*Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213* *Camurati-Engelmann disease, 131300*
TGM1	*100%*	*100%*	*Ichthyosis, congenital, autosomal recessive 1, 242300*
TGM5	*100%*	*100%*	*Peeling skin syndrome 2, 609796*
TH	*100%*	*100%*	*Segawa syndrome, recessive, 605407*
THOC6	*100%*	*100%*	*Beaulieu-Boycott-Innes syndrome, 613680*
THRB	*100%*	*100%*	*Thyroid hormone resistance, autosomal recessive, 274300* *Thyroid hormone resistance, 188570* *Thyroid hormone resistance, selective pituitary, 145650*
THUMPD1	*100%*	*100%*	*Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989*
TIMM50	*100%*	*100%*	*3-methylglutaconic aciduria, type IX, 617698*
TIMMDC1	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 31, 618251*
TJP2	*100%*	*100%*	*Hypercholanemia, familial 1, 607748* *Cholestasis, progressive familial intrahepatic 4, 615878*
TK2	*100%*	*100%*	*Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560* *?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069*
TKT	*98%*	*98%*	*Short stature, developmental delay, and congenital heart defects, 617044*
TLE6	*100%*	*100%*	*Preimplantation embryonic lethality, 616814*
TMC1	*100%*	*100%*	*Deafness, autosomal dominant 36, 606705* *Deafness, autosomal recessive 7, 600974*
TMC6	*100%*	*100%*	*Epidermodysplasia verruciformis, 226400*
TMC8	*100%*	*100%*	*Epidermodysplasia verruciformis 2, 618231*

TMCO1	*88%*	*88%*	*Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980*
TMEM107	*100%*	*100%*	*Orofaciodigital syndrome XVI, 617563* *Meckel syndrome 13, 617562* *?Joubert syndrome 29, 617562*
TMEM126A	*100%*	*100%*	*Optic atrophy 7, 612989*
TMEM126B	*100%*	*100%*	*Mitochondrial complex I deficiency, nuclear type 29, 618250*
TMEM132E	*100%*	*100%*	*Deafness, autosomal recessive 99, 618481*
TMEM138	*100%*	*100%*	*Joubert syndrome 16, 614465*
TMEM165	*100%*	*100%*	*Congenital disorder of glycosylation, type IIk, 614727*
TMEM199	*100%*	*100%*	*Congenital disorder of glycosylation, type IIp, 616829*
TMEM216	*100%*	*100%*	*Joubert syndrome 2, 608091* *Meckel syndrome 2, 603194*
TMEM231	*100%*	*100%*	*Joubert syndrome 20, 614970* *Meckel syndrome 11, 615397*
TMEM237	*100%*	*100%*	*Joubert syndrome 14, 614424*
TMEM260	*100%*	*100%*	*Structural heart defects and renal anomalies syndrome, 617478*
TMEM38B	*100%*	*100%*	*Osteogenesis imperfecta, type XIV, 615066*
TMEM67	*100%*	*98%*	*Nephronophthisis 11, 613550* *Joubert syndrome 6, 610688* *Meckel syndrome 3, 607361* *?RHYNS syndrome, 602152* *COACH syndrome 1, 216360*
TMEM70	*100%*	*100%*	*Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052*
TMEM94	*100%*	*100%*	*Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316*
TMIE	*100%*	*100%*	*Deafness, autosomal recessive 6, 600971*
TMPRSS15	*100%*	*100%*	*Enterokinase deficiency, 226200*
TMPRSS3	*100%*	*100%*	*Deafness, autosomal recessive 8/10, 601072*
TMPRSS6	*100%*	*100%*	*Iron-refractory iron deficiency anemia, 206200*
TMTC3	*100%*	*100%*	*Lissencephaly 8, 617255*
TNFRSF11A	*100%*	*100%*	*Osteopetrosis, autosomal recessive 7, 612301* *Osteolysis, familial expansile, 174810*
TNFRSF11B	*100%*	*100%*	*Paget disease of bone 5, juvenile-onset, 239000*
TNFRSF13B	*100%*	*100%*	*Immunodeficiency, common variable, 2, 240500* *Immunoglobulin A deficiency 2, 609529*
TNFRSF13C	*100%*	*100%*	*Immunodeficiency, common variable, 4, 613494*

TNFRSF4	*100%*	*100%*	*?Immunodeficiency 16, 615593*
TNFSF11	*100%*	*100%*	*Osteopetrosis, autosomal recessive 2, 259710*
TNIK	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 54, 617028*
TNNI3	*100%*	*100%*	*?Cardiomyopathy, dilated, 2A, 611880* *Cardiomyopathy, hypertrophic, 7, 613690* *Cardiomyopathy, familial restrictive, 1, 115210* *Cardiomyopathy, dilated, 1FF, 613286*
TNNT1	*100%*	*100%*	*Nemaline myopathy 5, Amish type, 605355*
TNXB	*100%*	*100%*	*Ehlers-Danlos syndrome, classic-like, 1, 606408* *Vesicoureteral reflux 8, 615963*
TOE1	*100%*	*100%*	*Pontocerebellar hypoplasia, type 7, 614969*
TONSL	*100%*	*100%*	*Spondyloepimetaphyseal dysplasia, sponastrime type, 271510*
TOP3A	*100%*	*100%*	*?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098* *Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097*
TOR1AIP1	*100%*	*100%*	*?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072*
TP53RK	*100%*	*100%*	*Galloway-Mowat syndrome 4, 617730*
TP73	*100%*	*100%*	*Ciliary dyskinesia, primary, 47, and lissencephaly, 619466*
TPI1	*100%*	*100%*	*Hemolytic anemia due to triosephosphate isomerase deficiency, 615512*
TPK1	*100%*	*100%*	*Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458*
TPM3	*100%*	*100%*	*CAP myopathy 1, 609284* *Myopathy, congenital, with fiber-type disproportion, 255310* *Nemaline myopathy 1, autosomal dominant or recessive, 609284*
TPO	*100%*	*100%*	*Thyroid dyshormonogenesis 2A, 274500*
TPP1	*100%*	*100%*	*Ceroid lipofuscinosis, neuronal, 2, 204500* *Spinocerebellar ataxia, autosomal recessive 7, 609270*
TPRKB	*82%*	*81%*	*Galloway-Mowat syndrome 5, 617731*
TPRN	*97%*	*95%*	*Deafness, autosomal recessive 79, 613307*
TRAC	*100%*	*100%*	*Immunodeficiency 7, TCR-alpha/beta deficient, 615387*
TRAF3IP1	*100%*	*100%*	*Senior-Loken syndrome 9, 616629*
TRAIP	*100%*	*100%*	*Seckel syndrome 9, 616777*
TRAK1	*100%*	*100%*	*Developmental and epileptic encephalopathy 68, 618201*
TRAPPC11	*100%*	*100%*	*Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356*
TRAPPC12	*100%*	*100%*	*Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669*
MAP11	*100%*	*100%*	*?Microcephaly 25, primary, autosomal recessive, 618351*

TRAPPC2L	*100%*	*100%*	*Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331*
TRAPPC6B	*100%*	*100%*	*Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862*
TRAPPC9	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 13, 613192*
TRDN	*100%*	*100%*	*Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441*
TREM2	*100%*	*100%*	*Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193*
TREX1	*100%*	*100%*	*Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315* *Aicardi-Goutieres syndrome 1, dominant and recessive, 225750* *Chilblain lupus, 610448*
TRH	*100%*	*100%*	*No OMIM disease ID*
TRIM2	*94%*	*94%*	*Charcot-Marie-Tooth disease, type 2R, 615490*
TRIM32	*100%*	*100%*	*?Bardet-Biedl syndrome 11, 615988* *Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110*
TRIM37	*98%*	*98%*	*Mulibrey nanism, 253250*
TRIOBP	*100%*	*100%*	*Deafness, autosomal recessive 28, 609823*
TRIP11	*100%*	*100%*	*Odontochondrodysplasia 1, 184260* *Achondrogenesis, type IA, 200600*
TRIP13	*100%*	*100%*	*Oocyte maturation defect 9, 619011* *Mosaic variegated aneuploidy syndrome 3, 617598*
TRIP4	*100%*	*100%*	*?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066* *Spinal muscular atrophy with congenital bone fractures 1, 616866*
TRIT1	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 35, 617873*
TRMT1	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 68, 618302*
TRMT10A	*100%*	*100%*	*Microcephaly, short stature, and impaired glucose metabolism 1, 616033*
TRMT10C	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 30, 616974*
TRMT5	*100%*	*100%*	*Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539*
TRMU	*100%*	*100%*	*Liver failure, transient infantile, 613070*
TRNT1	*100%*	*100%*	*Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084* *Retinitis pigmentosa and erythrocytic microcytosis, 616959*
TRPM1	*100%*	*100%*	*Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216*
TRPM6	*100%*	*100%*	*Hypomagnesemia 1, intestinal, 602014*
TSEN15	*100%*	*100%*	*Pontocerebellar hypoplasia, type 2F, 617026*
TSEN2	*100%*	*100%*	*Pontocerebellar hypoplasia type 2B, 612389*
TSEN34	*100%*	*100%*	*?Pontocerebellar hypoplasia type 2C, 612390*

TSEN54	*100%*	*100%*	*Pontocerebellar hypoplasia type 2A, 277470* *Pontocerebellar hypoplasia type 4, 225753* *?Pontocerebellar hypoplasia type 5, 610204*
TSFM	*94%*	*94%*	*Combined oxidative phosphorylation deficiency 3, 610505*
TSHB	*100%*	*100%*	*Hypothyroidism, congenital, nongoitrous 4, 275100*
TSHR	*100%*	*100%*	*Hyperthyroidism, familial gestational, 603373* *Hyperthyroidism, nonautoimmune, 609152* *Hypothyroidism, congenital, nongoitrous, 1, 275200* *Thyroid adenoma, hyperfunctioning, somatic,* *Thyroid carcinoma with thyrotoxicosis, somatic,*
TSPAN12	*100%*	*100%*	*Exudative vitreoretinopathy 5, 613310*
TSPEAR	*100%*	*100%*	*?Deafness, autosomal recessive 98, 614861* *Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180*
TSPYL1	*100%*	*100%*	*Sudden infant death with dysgenesis of the testes syndrome, 608800*
TTC19	*100%*	*100%*	*Mitochondrial complex III deficiency, nuclear type 2, 615157*
TTC21B	*100%*	*100%*	*Short-rib thoracic dysplasia 4 with or without polydactyly, 613819* *Nephronophthisis 12, 613820*
TTC7A	*100%*	*100%*	*Gastrointestinal defects and immunodeficiency syndrome, 243150*
TTC8	*100%*	*100%*	*Bardet-Biedl syndrome 8, 615985* *?Retinitis pigmentosa 51, 613464*
TTI2	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 39, 615541*
TTLL5	*100%*	*100%*	*Cone-rod dystrophy 19, 615860*
TTPA	*100%*	*100%*	*Ataxia with isolated vitamin E deficiency, 277460*
TUB	*100%*	*100%*	*?Retinal dystrophy and obesity, 616188*
TUBA8	*100%*	*100%*	*Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840*
TUBGCP2	*97%*	*97%*	*Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737*
TUBGCP4	*100%*	*100%*	*Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335*
TUBGCP6	*100%*	*100%*	*Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270*
TUFM	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 4, 610678*
TULP1	*100%*	*100%*	*Leber congenital amaurosis 15, 613843* *Retinitis pigmentosa 14, 600132*
TUSC3	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 7, 611093*
TWIST2	*100%*	*100%*	*Ablepharon-macrostomia syndrome, 200110* *Barber-Say syndrome, 209885* *Focal facial dermal dysplasia 3, Setleis type, 227260*

TWNK	*100%*	*100%*	*Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245* *Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286* *Perrault syndrome 5, 616138*
TXNL4A	*100%*	*100%*	*Burn-McKeown syndrome, 608572*
TYK2	*100%*	*100%*	*Immunodeficiency 35, 611521*
TYMP	*100%*	*100%*	*Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041*
TYR	*100%*	*100%*	*Albinism, oculocutaneous, type IB, 606952* *Waardenburg syndrome/albinism, digenic, 103470* *Albinism, oculocutaneous, type IA, 203100*
TYROBP	*100%*	*100%*	*Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770*
TYRP1	*100%*	*100%*	*Albinism, oculocutaneous, type III, 203290*
UBA5	*100%*	*100%*	*?Spinocerebellar ataxia, autosomal recessive 24, 617133* *Developmental and epileptic encephalopathy 44, 617132*
UBE2T	*100%*	*100%*	*Fanconi anemia, complementation group T, 616435*
UBE3B	*100%*	*100%*	*Kaufman oculocerebrofacial syndrome, 244450*
UBR1	*98%*	*98%*	*Johanson-Blizzard syndrome, 243800*
UCHL1	*100%*	*100%*	*Spastic paraplegia 79, autosomal recessive, 615491*
UFC1	*100%*	*100%*	*Neurodevelopmental disorder with spasticity and poor growth, 618076*
UFM1	*100%*	*100%*	*Leukodystrophy, hypomyelinating, 14, 617899*
UGDH	*100%*	*100%*	*Developmental and epileptic encephalopathy 84, 618792*
UGT1A1	*100%*	*100%*	*Crigler-Najjar syndrome, type I, 218800* *Hyperbilirubinemia, familial transient neonatal, 237900* *Crigler-Najjar syndrome, type II, 606785*
UMPS	*100%*	*100%*	*Orotic aciduria, 258900*
UNC13D	*100%*	*100%*	*Hemophagocytic lymphohistiocytosis, familial, 3, 608898*
UNC80	*100%*	*100%*	*Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801*
UNG	*100%*	*100%*	*Immunodeficiency with hyper IgM, type 5, 608106*
UPB1	*100%*	*100%*	*Beta-ureidopropionase deficiency, 613161*
UQCC2	*100%*	*100%*	*Mitochondrial complex III deficiency, nuclear type 7, 615824*
UQCC3	*100%*	*100%*	*?Mitochondrial complex III deficiency, nuclear type 9, 616111*
UQCRB	*100%*	*100%*	*Mitochondrial complex III deficiency, nuclear type 3, 615158*
UQCRC2	*100%*	*100%*	*Mitochondrial complex III deficiency, nuclear type 5, 615160*
UQCRFS1	*100%*	*100%*	*Mitochondrial complex III deficiency, nuclear type 10, 618775*

UQCRQ	*100%*	*100%*	*Mitochondrial complex III deficiency, nuclear type 4, 615159*
UROC1	*100%*	*100%*	*?Urocanase deficiency, 276880*
UROD	*100%*	*100%*	*Porphyria, hepatoerythropoietic, 176100* *Porphyria cutanea tarda, 176100*
UROS	*100%*	*100%*	*Porphyria, congenital erythropoietic, 263700*
USB1	*100%*	*100%*	*Poikiloderma with neutropenia, 604173*
USH1C	*100%*	*100%*	*Usher syndrome, type 1C, 276904* *Deafness, autosomal recessive 18A, 602092*
USH1G	*100%*	*100%*	*Usher syndrome, type 1G, 606943*
USH2A	*100%*	*100%*	*Usher syndrome, type 2A, 276901* *Retinitis pigmentosa 39, 613809*
USP18	*100%*	*100%*	*Pseudo-TORCH syndrome 2, 617397*
USP45	*100%*	*100%*	*?Leber congenital amaurosis 19, 618513*
UVSSA	*100%*	*100%*	*UV-sensitive syndrome 3, 614640*
VAC14	*100%*	*100%*	*Striatonigral degeneration, childhood-onset, 617054*
VAMP1	*100%*	*100%*	*Myasthenic syndrome, congenital, 25, 618323* *Spastic ataxia 1, autosomal dominant, 108600*
VARS1	*100%*	*100%*	*Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802*
VARS2	*100%*	*100%*	*Combined oxidative phosphorylation deficiency 20, 615917*
VAX1	*100%*	*99%*	*?Microphthalmia, syndromic 11, 614402*
VDR	*100%*	*100%*	*Rickets, vitamin D-resistant, type IIA, 277440*
VHL	*100%*	*100%*	*Erythrocytosis, familial, 2, 263400* *von Hippel-Lindau syndrome, 193300* *Renal cell carcinoma, somatic, 144700* *Pheochromocytoma, 171300* *Hemangioblastoma, cerebellar, somatic,*
VIPAS39	*100%*	*100%*	*Arthrogryposis, renal dysfunction, and cholestasis 2, 613404*
VKORC1	*98%*	*93%*	*Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473* *Warfarin resistance, 122700*
VLDLR	*100%*	*100%*	*Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050*
VPS11	*100%*	*100%*	*?Dystonia 32, 619637* *Leukodystrophy, hypomyelinating, 12, 616683*
VPS13A	*100%*	*100%*	*Choreoacanthocytosis, 200150*
VPS13B	*100%*	*99%*	*Cohen syndrome, 216550*
VPS13C	*100%*	*100%*	*Parkinson disease 23, autosomal recessive, early onset, 616840*

VPS13D	*100%*	*100%*	*Spinocerebellar ataxia, autosomal recessive 4, 607317*
VPS33A	*90%*	*90%*	*Mucopolysaccharidosis-plus syndrome, 617303*
VPS33B	*100%*	*100%*	*Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009* *Cholestasis, progressive familial intrahepatic, 12, 620010* *Arthrogryposis, renal dysfunction, and cholestasis 1, 208085*
VPS37A	*100%*	*100%*	*Spastic paraplegia 53, autosomal recessive, 614898*
VPS45	*95%*	*95%*	*Neutropenia, severe congenital, 5, autosomal recessive, 615285*
VPS51	*100%*	*100%*	*Pontocerebellar hypoplasia, type 13, 618606*
VPS53	*100%*	*100%*	*Pontocerebellar hypoplasia, type 2E, 615851*
VRK1	*100%*	*100%*	*Pontocerebellar hypoplasia type 1A, 607596*
VSX2	*100%*	*100%*	*Microphthalmia, isolated 2, 610093* *Microphthalmia with coloboma 3, 610092*
VWA1	*100%*	*100%*	*Neuropathy, hereditary motor, with myopathic features, 619216*
VWA3B	*100%*	*100%*	*?Spinocerebellar ataxia, autosomal recessive 22, 616948*
VWF	*100%*	*100%*	*von Willebrand disease, type 1, 193400* *von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554* *von Willebrand disease, type 3, 277480*
WARS2	*100%*	*100%*	*Parkinsonism-dystonia 3, childhood-onset, 619738* *Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710*
WASHC4	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 43, 615817*
WASHC5	*100%*	*100%*	*Ritscher-Schinzel syndrome 1, 220210* *Spastic paraplegia 8, autosomal dominant, 603563*
WBP2	*100%*	*100%*	*Deafness, autosomal recessive 107, 617639*
WDPCP	*98%*	*97%*	*?Bardet-Biedl syndrome 15, 615992* *Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085*
WDR19	*100%*	*100%*	*Nephronophthisis 13, 614377* *Cranioectodermal dysplasia 4, 614378* *Senior-Loken syndrome 8, 616307* *Short-rib thoracic dysplasia 5 with or without polydactyly, 614376* *?Spermatogenic failure 72, 619867*
WDR35	*100%*	*100%*	*Short-rib thoracic dysplasia 7 with or without polydactyly, 614091* *Cranioectodermal dysplasia 2, 613610*
WDR4	*100%*	*100%*	*Galloway-Mowat syndrome 6, 618347* *Microcephaly, growth deficiency, seizures, and brain malformations, 618346*

WDR45B	*100%*	*100%*	*Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977*
WDR62	*100%*	*100%*	*Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317*
WDR72	*97%*	*97%*	*Amelogenesis imperfecta, type IIA3, 613211*
WDR73	*100%*	*100%*	*Galloway-Mowat syndrome 1, 251300*
WDR81	*100%*	*100%*	*Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185* *Hydrocephalus, congenital, 3, with brain anomalies, 617967*
WEE2	*100%*	*100%*	*Oocyte maturation defect 5, 617996*
WFS1	*100%*	*100%*	*Deafness, autosomal dominant 6/14/38, 600965* *?Cataract 41, 116400* *Wolfram-like syndrome, autosomal dominant, 614296* *Wolfram syndrome 1, 222300*
WHRN	*100%*	*100%*	*Deafness, autosomal recessive 31, 607084* *Usher syndrome, type 2D, 611383*
WIPF1	*100%*	*100%*	*Wiskott-Aldrich syndrome 2, 614493*
WIPI2	*100%*	*100%*	*?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453*
WNK1	*100%*	*100%*	*Neuropathy, hereditary sensory and autonomic, type II, 201300* *Pseudohypoaldosteronism, type IIC, 614492*
WNT1	*100%*	*100%*	*Osteogenesis imperfecta, type XV, 615220*
WNT10A	*100%*	*100%*	*Schopf-Schulz-Passarge syndrome, 224750* *Tooth agenesis, selective, 4, 150400* *Odontoonychodermal dysplasia, 257980*
WNT10B	*100%*	*100%*	*Tooth agenesis, selective, 8, 617073* *Split-hand/foot malformation 6, 225300*
WNT3	*100%*	*100%*	*?Tetra-amelia syndrome 1, 273395*
WNT4	*100%*	*100%*	*?SERKAL syndrome, 611812* *Mullerian aplasia and hyperandrogenism, 158330*
WNT7A	*100%*	*100%*	*Fuhrmann syndrome, 228930* *Ulna and fibula, absence of, with severe limb deficiency, 276820*
WRAP53	*100%*	*100%*	*Dyskeratosis congenita, autosomal recessive 3, 613988*
WRN	*100%*	*100%*	*Werner syndrome, 277700*
WWOX	*100%*	*100%*	*Esophageal squamous cell carcinoma, somatic, 133239* *Developmental and epileptic encephalopathy 28, 616211* *Spinocerebellar ataxia, autosomal recessive 12, 614322*
XDH	*100%*	*100%*	*Xanthinuria, type I, 278300*
XPA	*100%*	*100%*	*Xeroderma pigmentosum, group A, 278700*

XPC	*100%*	*100%*	*Xeroderma pigmentosum, group C, 278720*
XPNPEP3	*100%*	*100%*	*Nephronophthisis-like nephropathy 1, 613159*
XRCC1	*100%*	*100%*	*?Spinocerebellar ataxia, autosomal recessive 26, 617633*
XRCC2	*100%*	*100%*	*Spermatogenic failure 50, 619145* *?Premature ovarian failure 17, 619146* *?Fanconi anemia, complementation group U, 617247*
XRCC4	*100%*	*100%*	*Short stature, microcephaly, and endocrine dysfunction, 616541*
XYLT1	*100%*	*100%*	*Desbuquois dysplasia 2, 615777*
XYLT2	*100%*	*99%*	*Spondyloocular syndrome, 605822*
YARS2	*100%*	*100%*	*Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561*
YIF1B	*90%*	*90%*	*Kaya-Barakat-Masson syndrome, 619125*
YME1L1	*100%*	*100%*	*?Optic atrophy 11, 617302*
YY1AP1	*100%*	*100%*	*Grange syndrome, 602531*
ZAP70	*100%*	*100%*	*Immunodeficiency 48, 269840* *Autoimmune disease, multisystem, infantile-onset, 2, 617006*
ZBTB11	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 69, 618383*
ZBTB16	*100%*	*100%*	*Leukemia, acute promyelocytic, PL2F/RARA type,*
ZBTB24	*100%*	*100%*	*Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069*
ZBTB42	*100%*	*100%*	*?Lethal congenital contracture syndrome 6, 616248*
ZC3H14	*100%*	*100%*	*Intellectual developmental disorder, autosomal recessive 56, 617125*
ZFYVE26	*100%*	*100%*	*Spastic paraplegia 15, autosomal recessive, 270700*
ZMPSTE24	*100%*	*100%*	*Mandibuloacral dysplasia with type B lipodystrophy, 608612* *Restrictive dermopathy 1, 275210*
ZMYND10	*100%*	*100%*	*Ciliary dyskinesia, primary, 22, 615444*
ZNF142	*100%*	*100%*	*Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425*
ZNF335	*100%*	*100%*	*Microcephaly 10, primary, autosomal recessive, 615095*
ZNF341	*100%*	*100%*	*Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282*
ZNF408	*100%*	*100%*	*Retinitis pigmentosa 72, 616469* *?Exudative vitreoretinopathy 6, 616468*
ZNF423	*100%*	*100%*	*Nephronophthisis 14, 614844* *Joubert syndrome 19, 614844*
ZNF469	*100%*	*100%*	*Brittle cornea syndrome 1, 229200*
ZNF513	*100%*	*100%*	*?Retinitis pigmentosa 58, 613617*
ZNHIT3	*78%*	*76%*	*PEHO syndrome, 260565*

ZP1	*100%*	*100%*	*Oocyte maturation defect 1, 615774*

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST X2 is the chemistry used for WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 28th , 2022.
This list is accurate for panel version DG 3.5.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors