SecureCari®

Expanded Carrier Screening - 1784 Disorders Panel

Screens for 1,784 autosomal recessive and X-linked recessive disorders associated with 1,766 genes

This panel is available only for couple-based testing. We report pathogenic/ likely pathogenic variants only when both partners carry variants in the same gene, or when the female partner carries an X-linked variant. Single-carrier findings for autosomal recessive conditions and variants of uncertain significance are not disclosed in the final report.

Disorders	Genes	Inheritance
Achalasia-addisonianism-alacrimia syndrome	*AAAS*	AR
Combined oxidative phosphorylation deficiency 8	*AARS2*	AR
GABA-transaminase deficiency	*ABAT*	AR
Ichthyosis, congenital, autosomal recessive 4A	*ABCA12*	AR
Surfactant metabolism dysfunction, pulmonary, 3	*ABCA3*	AR
Cone-rod dystrophy 3	*ABCA4*	AR
Cholestasis, progressive familial intrahepatic 2	*ABCB11*	AR
Cholestasis, progressive familial intrahepatic 3	*ABCB4*	AR
Anemia, sideroblastic, with ataxia	*ABCB7*	XLR
Arterial calcification, generalized, of infancy, 2	*ABCC6*	AR
Hyperinsulinemic hypoglycemia, familial, 1	*ABCC8*	AD/ AR
Adrenoleukodystrophy	*ABCD1*	XLR
Sitosterolemia 2	*ABCG5*	AR
Sitosterolemia 1	*ABCG8*	AR
Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract	*ABHD12*	AR
Spastic paraplegia 86, autosomal recessive	*ABHD16A*	AR
Chanarin-Dorfman syndrome	*ABHD5*	AR
Mitochondrial complex I deficiency, nuclear type 20	*ACAD9*	AR
Acyl-CoA dehydrogenase, medium chain, deficiency of	*ACADM*	AR
2-methylbutyrylglycinuria	*ACADSB*	AR
VLCAD deficiency	*ACADVL*	AR
Spondyloepimetaphyseal dysplasia, aggrecan type	*ACAN*	AR
Alpha-methylacetoacetic aciduria	*ACAT1*	AR
Renal tubular dysgenesis	*ACE*	AR
Infantile cerebellar-retinal degeneration	*ACO2*	AR
Peroxisomal acyl-CoA oxidase deficiency	*ACOX1*	AR
Spondyloenchondrodysplasia with immune dysregulation	*ACP5*	AR
Combined malonic and methylmalonic aciduria	*ACSF3*	AR
Congenital myopathy 2B, severe infantile, autosomal recessive	*ACTA1*	AR
Developmental and epileptic encephalopathy 76	*ACTL6B*	AR
Aminoacylase 1 deficiency	*ACY1*	AR
Adenosine deaminase deficiency, partial	*ADA*	AR/ SMo
Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome	*ADA2*	AR
Inflammatory skin and bowel disease, neonatal, 1	*ADAM17*	AR
Cone-rod dystrophy 9	*ADAM9*	AR
Weill-Marchesani syndrome 1, recessive	*ADAMTS10*	AR
Thrombotic thrombocytopenic purpura, hereditary	*ADAMTS13*	AR
Weill-Marchesani 4 syndrome, recessive	*ADAMTS17*	AR
Microcornea, myopic chorioretinal atrophy, and telecanthus	*ADAMTS18*	AR
Ehlers-Danlos syndrome, dermatosparaxis type	*ADAMTS2*	AR
Geleophysic dysplasia 1	*ADAMTSL2*	AR
Aicardi-Goutieres syndrome 6	*ADAR*	AR
Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies	*ADAT3*	AR
Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal)	*ADGRG1*	AR
Usher syndrome, type 2C	*ADGRV1*	AR/ DD
Hypermethioninemia due to adenosine kinase deficiency	*ADK*	AR
Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures	*ADPRS*	AR
Adenylosuccinase deficiency	*ADSL*	AR
Ehlers-Danlos syndrome, classic-like, 2	*AEBP1*	AR
Intellectual developmental disorder, X-linked 109	*AFF2*	XLR
Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities	*AFG2A*	AR
Aspartylglucosaminuria	*AGA*	AR
Retinitis pigmentosa 75	*AGBL5*	AR
Sengers syndrome	*AGK*	AR
Glycogen storage disease IIIa	*AGL*	AR
Rhizomelic chondrodysplasia punctata, type 3	*AGPS*	AR
Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects	*AGRN*	AR
Renal tubular dysgenesis	*AGT*	AR
Neurodegeneration, childhood-onset, with cerebellar atrophy	*AGTPBP1*	AR
Renal tubular dysgenesis	*AGTR1*	AR
Hyperoxaluria, primary, type 1	*AGXT*	AR
Joubert syndrome 3	*AHI1*	AR
Immunodeficiency with hyper-IgM, type 2	*AICDA*	AR
Cowchock syndrome	*AIFM1*	XLR
Leukodystrophy, hypomyelinating, 3	*AIMP1*	AR
Leber congenital amaurosis 4	*AIPL1*	AD/ AR
Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia	*AIRE*	AD/ AR
Reticular dysgenesis	*AK2*	AR
Bile acid synthesis defect, congenital, 2	*AKR1D1*	AR
Porphyria, acute hepatic	*ALAD*	AR
Protoporphyria, erythropoietic, X-linked	*ALAS2*	XL
Spastic paraplegia 9B, autosomal recessive	*ALDH18A1*	AR
Microphthalmia, isolated 8	*ALDH1A3*	AR
Sjogren-Larsson syndrome	*ALDH3A2*	AR
Hyperprolinemia, type II	*ALDH4A1*	AR
Succinic semialdehyde dehydrogenase deficiency	*ALDH5A1*	AR

Disorders	Genes	Inheritance
Epilepsy, early-onset, 4, vitamin B6-dependent	*ALDH7A1*	AR
Fructose intolerance, hereditary	*ALDOB*	AR
Congenital disorder of glycosylation, type Ik	*ALG1*	AR
Congenital disorder of glycosylation, type Ip	*ALG11*	AR
Congenital disorder of glycosylation, type Ig	*ALG12*	AR
Developmental and epileptic encephalopathy 36	*ALG13*	XL
Myasthenic syndrome, congenital, 14, with tubular aggregates	*ALG2*	AR
Congenital disorder of glycosylation, type Id	*ALG3*	AR
Congenital disorder of glycosylation, type Ic	*ALG6*	AR
Congenital disorder of glycosylation, type Ih	*ALG8*	AR
Congenital disorder of glycosylation, type Il	*ALG9*	AR
Alstrom syndrome	*ALMS1*	AR
Ichthyosis, congenital, autosomal recessive 2	*ALOX12B*	AR
Ichthyosis, congenital, autosomal recessive 3	*ALOXE3*	AR
Cardiomyopathy, familial hypertrophic 27	*ALPK3*	AR
Hypophosphatasia, infantile	*ALPL*	AR
Primary lateral sclerosis, juvenile	*ALS2*	AR
Myopathy due to myoadenylate deaminase deficiency	*AMPD1*	AR
Pontocerebellar hypoplasia, type 9	*AMPD2*	AR
Glycine encephalopathy 2	*AMT*	-
Spherocytosis, type 1	*ANK1*	AD/ AR
Nephronophthisis 16	*ANKS6*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 12	*ANO5*	AR
GAPO syndrome	*ANTXR1*	AR
Hyaline fibromatosis syndrome	*ANTXR2*	AR
Dystonia 31	*AOPEP*	AR
Keratitis-ichthyosis-deafness syndrome, autosomal recessive	*AP1B1*	AR
MEDNIK syndrome	*AP1S1*	AR
Pettigrew syndrome	*AP1S2*	XLR
Hermansky-Pudlak syndrome 2	*AP3B1*	AR
Developmental and epileptic encephalopathy 48	*AP3B2*	AR
Spastic paraplegia 47, autosomal recessive	*AP4B1*	AR
Spastic paraplegia 51, autosomal recessive	*AP4E1*	AR
Spastic paraplegia 50, autosomal recessive	*AP4M1*	AR
Spastic paraplegia 52, autosomal recessive	*AP4S1*	AR
Spastic paraplegia 48, autosomal recessive	*AP5Z1*	AR
Cortical dysplasia, complex, with other brain malformations 10	*APC2*	AR
Hypobetalipoproteinemia	*APOB*	AR
Hyperlipoproteinemia, type Ib	*APOC2*	AR
Adenine phosphoribosyltransferase deficiency	*APRT*	AR
Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	*APTX*	AR
Diabetes insipidus, nephrogenic, 2	*AQP2*	AD/ AR
Androgen insensitivity	*AR*	XLR
Androgen insensitivity, partial, with or without breast cancer	*AR*	XLR
Periventricular heterotopia with microcephaly	*ARFGEF2*	AR
Argininemia	*ARG1*	AR

Disorders	Genes	Inheritance
Developmental and epileptic encephalopathy 8	*ARHGEF9*	XL
Joubert syndrome 8	*ARL13B*	AR
Retinitis pigmentosa with or without situs inversus	*ARL2BP*	AR
Bardet-Biedl syndrome 3	*ARL6*	AR
Spastic paraplegia 61, autosomal recessive	*ARL6IP1*	AR
Joubert syndrome 30	*ARMC9*	AR
Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia	*ARPC1B*	AR
Myopia 26, X-linked, female-limited	*ARR3*	XL
Metachromatic leukodystrophy	*ARSA*	AR
Mucopolysaccharidosis type VI (Maroteaux-Lamy)	*ARSB*	AR
Usher syndrome, type IV	*ARSG*	AR
Chondrodysplasia punctata, X-linked recessive	*ARSL*	XLR
Developmental and epileptic encephalopathy 38	*ARV1*	AR
Hydranencephaly with abnormal genitalia	*ARX*	XL
Farber lipogranulomatosis	*ASAH1*	AR
Spinal muscular atrophy with congenital bone fractures 2	*ASCC1*	AR
Argininosuccinic aciduria	*ASL*	AR
Asparagine synthetase deficiency	*ASNS*	AR
Canavan disease	*ASPA*	AR
Traboulsi syndrome	*ASPH*	AR
Microcephaly 5, primary, autosomal recessive	*ASPM*	AR
Citrullinemia	*ASS1*	AR
Hyperekplexia 4	*ATAD1*	AR
Harel-Yoon syndrome	*ATAD3A*	AD/ AR
Ataxia, cerebellar, Cayman type	*ATCAY*	AR
Achromatopsia 7	*ATF6*	AR
Spinocerebellar ataxia, autosomal recessive 31	*ATG7*	AR
Ataxia-telangiectasia	*ATM*	AR
Persistent hyperplastic primary vitreous, autosomal recessive	*ATOH7*	AR
Spastic paraplegia 78, autosomal recessive	*ATP13A2*	AR
Pulmonary hypertension, primary, 5	*ATP13A3*	AR
Immunodeficiency 47	*ATP6AP1*	XLR
Congenital disorder of glycosylation, type IIr	*ATP6AP2*	XLR
Intellectual developmental disorder, X-linked syndromic, Hedera type	*ATP6AP2*	XLR
Cutis laxa, autosomal recessive, type IIA	*ATP6V0A2*	AR
Distal renal tubular acidosis 3, with or without sensorineural hearing loss	*ATP6V0A4*	AR
Distal renal tubular acidosis 2 with progressive sensorineural hearing loss	*ATP6V1B1*	AR
Menkes disease	*ATP7A*	XLR
Wilson disease	*ATP7B*	AR
Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4	*ATP8A2*	AR
Cholestasis, progressive familial intrahepatic 1	*ATP8B1*	AR
Seckel syndrome 1	*ATR*	AR
Intellectual disability-hypotonic facies syndrome, X-linked	*ATRX*	XLR
3-methylglutaconic aciduria, type I	*AUH*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11	*B3GALNT2*	AR

Disorders	Genes	Inheritance
Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	*B3GALT6*	AR
Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects	*B3GAT3*	AR
Peters-plus syndrome	*B3GLCT*	AR
Spastic paraplegia 26, autosomal recessive	*B4GALNT1*	AR
Ehlers-Danlos syndrome, spondylodysplastic type, 1	*B4GALT7*	AR
Bardet-Biedl syndrome 1	*BBS1*	AR/ DR
Bardet-Biedl syndrome 10	*BBS10*	AR
Bardet-Biedl syndrome 12	*BBS12*	AR
Bardet-Biedl syndrome 2	*BBS2*	AR
Bardet-Biedl syndrome 4	*BBS4*	AR
Bardet-Biedl syndrome 5	*BBS5*	AR
Bardet-Biedl syndrome 7	*BBS7*	AR
Bardet-Biedl syndrome 9	*BBS9*	AR
Deafness, dystonia, and cerebral hypomyelination	*BCAP31*	XLR
Hengel-Maroofian-Schols syndrome	*BCAS3*	AR
Hypervalinemia or hyperleucine-isoleucinemia	*BCAT2*	AR
Maple syrup urine disease, type Ia	*BCKDHA*	AR
Maple syrup urine disease, type Ib	*BCKDHB*	AR
Branched-chain keto acid dehydrogenase kinase deficiency	*BCKDK*	-
Immunodeficiency 37	*BCL10*	AR
GRACILE syndrome	*BCS1L*	AR
Meester-Loeys syndrome	*BGN*	XL
Centronuclear myopathy 2	*BIN1*	AR
Bloom syndrome	*BLM*	AR
Agammaglobulinemia 4	*BLNK*	AR
Hermansky-Pudlak syndrome 9	*BLOC1S6*	AR
Alkuraya-Kucinskas syndrome	*BLTP1*	AR
Diaphanospondylodysostosis	*BMPER*	AR
Acromesomelic dysplasia 3	*BMPR1B*	AR
Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia	*BOLA3*	AR
Rigidity and multifocal seizure syndrome, lethal neonatal	*BRAT1*	AR
Fanconi anemia, complementation group S	*BRCA1*	AR
Fanconi anemia, complementation group D1	*BRCA2*	AR
Cerebellofaciodental syndrome	*BRF1*	AR
Intellectual developmental disorder, X-linked 93	*BRWD3*	XLR
Encephalopathy, progressive, with or without lipodystrophy	*BSCL2*	AR
Bartter syndrome, type 4a	*BSND*	AR
Biotinidase deficiency	*BTD*	AR
Isolated growth hormone deficiency, type III, with agammaglobulinemia	*BTK*	XLR
Mosaic variegated aneuploidy syndrome 1	*BUB1B*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 25	*BVES*	AR
Intellectual developmental disorder, autosomal recessive 66	*C12orf4*	AR
Temtamy syndrome	*C12orf57*	AR
Neurodegeneration with brain iron accumulation 4	*C19orf12*	AD/ AR
C1q deficiency 1	*C1QA*	AR

Disorders	Genes	Inheritance
C1q deficiency 2	*C1QB*	-
Combined oxidative phosphorylation deficiency 33	*C1QBP*	AR
C1q deficiency 3	*C1QC*	-
Orofaciodigital syndrome XIV	*C2CD3*	AR
Combined oxidative phosphorylation deficiency 53	*C2orf69*	AR
C3 deficiency	*C3*	AR
C5 deficiency	*C5*	AR
C6 deficiency	*C6*	AR
C7 deficiency	*C7*	.
C8 deficiency, type II	*C8B*	AR
Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	*CA2*	AR
Hyperammonemia due to carbonic anhydrase VA deficiency	*CA5A*	AR
Deafness, autosomal recessive 93	*CABP2*	AR
Cone-rod synaptic disorder, congenital nonprogressive	*CABP4*	AR
Cone-rod dystrophy, X-linked, 3	*CACNA1F*	XLR
Retinal cone dystrophy 4	*CACNA2D4*	AR
Desbuquois dysplasia 1	*CANT1*	AR
Spastic paraplegia 76, autosomal recessive	*CAPN1*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 1	*CAPN3*	AR
Immunodeficiency 11A	*CARD11*	AR
Immunodeficiency 103, susceptibility to fungal infection	*CARD9*	AR
Combined oxidative phosphorylation deficiency 27	*CARS2*	AR
Intellectual developmental disorder, with or without nystagmus	*CASK*	XLR
Ventricular tachycardia, catecholaminergic polymorphic, 2	*CASQ2*	AR
Hyperparathyroidism, neonatal	*CASR*	AD/ AR
Lipodystrophy, congenital generalized, type 4	*CAVIN1*	AR
Homocystinuria, B6-responsive and nonresponsive types	*CBS*	AR
Intellectual developmental disorder, autosomal recessive 3	*CC2D1A*	AR
Joubert syndrome 9	*CC2D2A*	AR
Hennekam lymphangiectasia-lymphedema syndrome 1	*CCBE1*	AR
Ciliary dyskinesia, primary, 17	*CCDC103*	AR
Congenital disorder of glycosylation, type IIo	*CCDC115*	AR
Ciliary dyskinesia, primary, 14	*CCDC39*	AR
Ciliary dyskinesia, primary, 15	*CCDC40*	AR
3-M syndrome 3	*CCDC8*	AR
Hydrocephalus, congenital, 1	*CCDC88C*	AR
Progressive pseudorheumatoid dysplasia	*CCN6*	AR
Ciliary dyskinesia, primary, 29	*CCNO*	AR
Immunodeficiency, common variable, 3	*CD19*	AR
Immunodeficiency 25	*CD247*	AR
Lymphoproliferative syndrome 2	*CD27*	AR
Methylmalonic aciduria, transient, due to transcobalamin receptor defect	*CD320*	AR
Platelet glycoprotein IV deficiency	*CD36*	AR
Immunodeficiency 19, severe combined	*CD3D*	AR

Disorders	Genes	Inheritance
Immunodeficiency 18	*CD3E*	AR
Immunodeficiency 17, CD3 gamma deficient	*CD3G*	AR
Immunodeficiency with hyper-IgM, type 3	*CD40*	AR
Immunodeficiency, X-linked, with hyper-IgM	*CD40LG*	XLR
Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy	*CD55*	AR
Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	*CD59*	AR
Lymphoproliferative syndrome 3	*CD70*	AR
Agammaglobulinemia 3	*CD79A*	AR
Agammaglobulinemia 6	*CD79B*	AR
Immunodeficiency, common variable, 6	*CD81*	AR
Dyserythropoietic anemia, congenital, type Ia	*CDAN1*	AR
Meier-Gorlin syndrome 7	*CDC45*	AR
Elsahy-Waters syndrome	*CDH11*	AR
Usher syndrome, type 1D	*CDH23*	AR/DR
Ectodermal dysplasia, ectrodactyly, and macular dystrophy	*CDH3*	AR
Al Kaissi syndrome	*CDK10*	AR
Microcephaly 3, primary, autosomal recessive	*CDK5RAP2*	AR
Meier-Gorlin syndrome 4	*CDT1*	AR
Deafness, autosomal recessive 113	*CEACAM16*	AR
Stromme syndrome	*CENPF*	AR
Microcephaly 6, primary, autosomal recessive	*CENPJ*	AR
Short-rib thoracic dysplasia 13 with or without polydactyly	*CEP120*	AR
Microcephaly 8, primary, autosomal recessive	*CEP135*	AR
Seckel syndrome 5	*CEP152*	AR
Nephronophthisis 15	*CEP164*	AR
Cone-rod dystrophy and hearing loss 2	*CEP250*	AR
Joubert syndrome 5	*CEP290*	AR
Joubert syndrome 15	*CEP41*	AR
Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly	*CEP55*	AR
Mosaic variegated aneuploidy syndrome 2	*CEP57*	AR
Cone-rod dystrophy and hearing loss	*CEP78*	AR
Retinitis pigmentosa 26	*CERKL*	AR
Ichthyosis, congenital, autosomal recessive 9	*CERS3*	AR
Ciliary dyskinesia, primary, 38	*CFAP300*	AR
Retinal dystrophy with macular staphyloma	*CFAP410*	AR
Cone-rod dystrophy 16	*CFAP418*	AR
Complement factor D deficiency	*CFD*	AR
Complement factor H deficiency	*CFH*	AD/ AR
Complement factor I deficiency	*CFI*	AR
Nemaline myopathy 7, autosomal recessive	*CFL2*	AR
Properdin deficiency, X-linked	*CFP*	XLR
Cystic fibrosis	*CFTR*	AR
Myasthenic syndrome, congenital, 6, presynaptic	*CHAT*	AR
Muscular dystrophy, congenital, megaconial type	*CHKB*	AR
Choroideremia	*CHM*	XL
Pontocerebellar hypoplasia, type 8	*CHMP1A*	AR
Multiple pterygium syndrome, lethal type	*CHRNA1*	AR

Disorders	Genes	Inheritance
Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	*CHRNB1*	AR
Myasthenic syndrome, congenital, 3B, fast-channel	*CHRND*	AR
Myasthenic syndrome, congenital, 4A, slow-channel	*CHRNE*	AD/ AR
Escobar syndrome	*CHRNG*	AR
Ehlers-Danlos syndrome, musculocontractural type 1	*CHST14*	AR
Spondyloepiphyseal dysplasia with congenital joint dislocations	*CHST3*	AR
Temtamy preaxial brachydactyly syndrome	*CHSY1*	AR
Usher syndrome, type IJ	*CIB2*	AR
Bare lymphocyte syndrome, type II, complementation group A	*CIITA*	AR
Wolfram syndrome 2	*CISD2*	AR
Microcephaly 17, primary, autosomal recessive	*CIT*	AR
Filippi syndrome	*CKAP2L*	AR
Cold-induced sweating syndrome 2	*CLCF1*	AR
Myotonia congenita, recessive	*CLCN1*	AR
Leukoencephalopathy with ataxia	*CLCN2*	AR
Dent disease 1	*CLCN5*	XLR
Osteopetrosis, autosomal recessive 4	*CLCN7*	AR
Bartter syndrome, type 4b, digenic	*CLCNKB*	DR
Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	*CLDN1*	AR
HELIX syndrome	*CLDN10*	AR
Deafness, autosomal recessive 29	*CLDN14*	AR
Hypomagnesemia 5, renal, with ocular involvement	*CLDN19*	AR
Congenital short bowel syndrome	*CLMP*	AR
Ceroid lipofuscinosis, neuronal, 3	*CLN3*	AR
Ceroid lipofuscinosis, neuronal, 5	*CLN5*	AR
Ceroid lipofuscinosis, neuronal, 6A	*CLN6*	AR
Ceroid lipofuscinosis, neuronal, 8	*CLN8*	AR
Pontocerebellar hypoplasia, type 10	*CLP1*	AR
3-methylglutaconic aciduria, type VIIB, autosomal recessive	*CLPB*	AR
Perrault syndrome 3	*CLPP*	AR
Usher syndrome, type 3A	*CLRN1*	AR
Retinitis pigmentosa 49	*CNGA1*	AR
Achromatopsia 2	*CNGA3*	AR
Retinitis pigmentosa 45	*CNGB1*	AR
Achromatopsia 3	*CNGB3*	AR
Intellectual developmental disorder, X-linked syndromic, Houge type	*CNKSR2*	XL
Hypomagnesemia, seizures, and impaired intellectual development 1	*CNNM2*	AD/ AR
Jalili syndrome	*CNNM4*	AR
Lethal congenital contracture syndrome 7	*CNTNAP1*	AR
Pitt-Hopkins like syndrome 1	*CNTNAP2*	AR
Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3	*COA7*	AR
Mitochondrial complex IV deficiency, nuclear type 17	*COA8*	AR
Congenital disorder of glycosylation, type IIj	*COG4*	AR
Congenital disorder of glycosylation, type IIi	*COG5*	AR
Congenital disorder of glycosylation, type III	*COG6*	AR

Disorders	Genes	Inheritance
Congenital disorder of glycosylation, type IIe	*COG7*	AR
Fibrochondrogenesis 1	*COL11A1*	AR
Fibrochondrogenesis 2	*COL11A2*	AD/ AR
Epidermolysis bullosa, junctional 4, intermediate	*COL17A1*	AR
Knobloch syndrome, type 1	*COL18A1*	AR
Ehlers-Danlos syndrome, cardiac valvular type	*COL1A2*	AR
Steel syndrome	*COL27A1*	AR
Polymicrogyria with or without vascular-type EDS	*COL3A1*	AR
Alport syndrome 3B, autosomal recessive	*COL4A3*	-
Alport syndrome 2, autosomal recessive	*COL4A4*	AR
Ullrich congenital muscular dystrophy 1	*COL6A1*	AD/AR
Ullrich congenital muscular dystrophy 1	*COL6A2*	AD/AR
Ullrich congenital muscular dystrophy 1	*COL6A3*	AD/AR
Epidermolysis bullosa dystrophica, autosomal recessive	*COL7A1*	AR
Stickler syndrome, type V	*COL9A2*	AR
Stickler syndrome, type VI	*COL9A3*	AR
3MC syndrome 2	*COLEC11*	AR
Myasthenic syndrome, congenital, 5	*COLQ*	AR
Coenzyme Q10 deficiency, primary, 1	*COQ2*	AR
Coenzyme Q10 deficiency, primary, 7	*COQ4*	AR
Coenzyme Q10 deficiency, primary, 6	*COQ6*	AR
Coenzyme Q10 deficiency, primary, 4	*COQ8A*	AR
Nephrotic syndrome, type 9	*COQ8B*	AR
Coenzyme Q10 deficiency, primary, 5	*COQ9*	AR
Immunodeficiency 8	*CORO1A*	AR
Mitochondrial complex IV deficiency, nuclear type 3	*COX10*	AR
Mitochondrial complex IV deficiency, nuclear type 6	*COX15*	AR
Mitochondrial complex IV deficiency, nuclear type 11	*COX20*	AR
Mitochondrial complex IV deficiency, nuclear type 7	*COX6B1*	AR
Aceruloplasminemia	*CP*	AR
Anterior segment dysgenesis 8	*CPAMD8*	AR
Joubert syndrome 17	*CPLANE1*	AR
Carbamoylphosphate synthetase I deficiency	*CPS1*	AR
CPT deficiency, hepatic, type IA	*CPT1A*	AR
CPT II deficiency, lethal neonatal	*CPT2*	AR
Immunodeficiency, common variable, 7	*CR2*	AR
Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly	*CRADD*	AR
Leber congenital amaurosis 8	*CRB1*	AR
Ventriculomegaly with cystic kidney disease	*CRB2*	AR
Cold-induced sweating syndrome 1	*CRLF1*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7	*CRPPA*	AR
Osteogenesis imperfecta, type VII	*CRTAP*	AR
Neutropenia, severe congenital, 7, autosomal recessive	*CSF3R*	AR
Joubert syndrome 21	*CSPP1*	AR
Peeling skin syndrome 4	*CSTA*	AR
Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	*CSTB*	AR

Disorders	Genes	Inheritance
Cerebroretinal microangiopathy with calcifications and cysts	*CTC1*	AR
Cystinosis, nephropathic	*CTNS*	AR
Immunodeficiency 24	*CTPS1*	AR
Galactosialidosis	*CTSA*	AR
Papillon-Lefevre syndrome	*CTSC*	AR
Ceroid lipofuscinosis, neuronal, 10	*CTSD*	AR
Ceroid lipofuscinosis, neuronal, 13 (Kufs type)	*CTSF*	AR
Pycnodysostosis	*CTSK*	AR
Imerslund-Grasbeck syndrome 1	*CUBN*	AR
Intellectual developmental disorder, X-linked syndromic, Cabezas type	*CUL4B*	XLR
3-M syndrome 1	*CUL7*	AR
Retinitis pigmentosa with or without skeletal anomalies	*CWC27*	AR
Methemoglobinemia, type I	*CYB5R3*	AR
Chronic granulomatous disease 4, autosomal recessive	*CYBA*	AR
Chronic granulomatous disease, X-linked	*CYBB*	XLR
Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	*CYP11A1*	-
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	*CYP11B1*	AR
Hypoaldosteronism, congenital, due to CMO I deficiency	*CYP11B2*	AR
17,20-lyase deficiency, isolated	*CYP17A1*	AR
Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset	*CYP1B1*	AR
Hypercalcemia, infantile, 1	*CYP24A1*	AR
Focal facial dermal dysplasia 4	*CYP26C1*	AR
Cerebrotendinous xanthomatosis	*CYP27A1*	AR
Vitamin D-dependent rickets, type I	*CYP27B1*	AR
Rickets due to defect in vitamin D 25-hydroxylation deficiency	*CYP2R1*	AR
Spastic paraplegia 56, autosomal recessive	*CYP2U1*	AR
Ichthyosis, congenital, autosomal recessive 5	*CYP4F22*	AR
Bietti crystalline corneoretinal dystrophy	*CYP4V2*	AR
Bile acid synthesis defect, congenital, 3	*CYP7B1*	AR
D-2-hydroxyglutaric aciduria	*D2HGDH*	AR
Hypomyelination with brainstem and spinal cord involvement and leg spasticity	*DARS1*	AR
Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	*DARS2*	AR
Orthostatic hypotension 1, due to DBH deficiency	*DBH*	AR
Maple syrup urine disease, type II	*DBT*	AR
Woodhouse-Sakati syndrome	*DCAF17*	AR
Nephronophthisis 19	*DCDC2*	AR
Van Maldergem syndrome 1	*DCHS1*	AR
Severe combined immunodeficiency, Athabascan type	*DCLRE1C*	AR
Lissencephaly, X-linked	*DCX*	XL
Xeroderma pigmentosum, group E, DDB-negative subtype	*DDB2*	AR
Aromatic L-amino acid decarboxylase deficiency	*DDC*	AR
Spastic paraplegia 28, autosomal recessive	*DDHD1*	AR

Disorders	Genes	Inheritance
Spastic paraplegia 54, autosomal recessive	*DDHD2*	AR
Spondylometaepiphyseal dysplasia, short limb-hand type	*DDR2*	AR
Warsaw breakage syndrome	*DDX11*	AR
Intellectual developmental disorder, X-linked syndromic, Snijders Blok type	*DDX3X*	XLD/ XLR
Orofaciodigital syndrome V	*DDX59*	AR
Leukodystrophy, hypomyelinating, 18	*DEGS1*	AR
Developmental and epileptic encephalopathy 49	*DENND5A*	AR
Diarrhea 7, protein-losing enteropathy type	*DGAT1*	AR
Nephrotic syndrome, type 7	*DGKE*	AR
Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	*DGUOK*	AR
Desmosterolosis	*DHCR24*	AR
Smith-Lemli-Opitz syndrome	*DHCR7*	AR
Retinitis pigmentosa 59	*DHDDS*	AR
Miller syndrome	*DHODH*	AR
Perlman syndrome	*DIS3L2*	AR
Dyskeratosis congenita, X-linked	*DKC1*	XLR
Dihydrolipoamide dehydrogenase deficiency	*DLD*	AR
Intellectual developmental disorder, X-linked 90	*DLG3*	XLR
Spondylocostal dysostosis 1, autosomal recessive	*DLL3*	AR
Duchenne muscular dystrophy	*DMD*	XLR
Hypophosphatemic rickets, AR	*DMP1*	AR
Ciliary dyskinesia, primary, 13	*DNAAF1*	AR
Ciliary dyskinesia, primary, 19	*DNAAF11*	AR
Ciliary dyskinesia, primary, 10	*DNAAF2*	AR
Ciliary dyskinesia, primary, 2	*DNAAF3*	AR
Ciliary dyskinesia, primary, 25	*DNAAF4*	AR
Ciliary dyskinesia, primary, 18	*DNAAF5*	AR
Ciliary dyskinesia, primary, 36, X-linked	*DNAAF6*	XLR
Ciliary dyskinesia, primary, 7, with or without situs inversus	*DNAH11*	AR
Ciliary dyskinesia, primary, 3, with or without situs inversus	*DNAH5*	AR
Ciliary dyskinesia, primary, 40	*DNAH9*	AR
Ciliary dyskinesia, primary, 1, with or without situs inversus	*DNAI1*	AR
Ciliary dyskinesia, primary, 9, with or without situs inversus	*DNAI2*	AR
Neuronopathy, distal hereditary motor, autosomal recessive 5	*DNAJB2*	AR
Hyperphenylalaninemia, mild, non-BH4-deficient	*DNAJC12*	AR
3-methylglutaconic aciduria, type V	*DNAJC19*	AR
Bone marrow failure syndrome 3	*DNAJC21*	AR
Parkinson disease 19a, juvenile-onset	*DNAJC6*	AR
Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1	*DNM1L*	AD/ AR
Immunodeficiency-centromeric instability-facial anomalies syndrome 1	*DNMT3B*	AR
Immunodeficiency 40	*DOCK2*	AR
Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia	*DOCK3*	AR
Adams-Oliver syndrome 2	*DOCK6*	AR

Disorders	Genes	Inheritance
Developmental and epileptic encephalopathy 23	*DOCK7*	AR
Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections	*DOCK8*	AR
Myasthenic syndrome, congenital, 10	*DOK7*	AR
Congenital disorder of glycosylation, type Im	*DOLK*	AR
Microcephaly, short stature, and limb abnormalities	*DONSON*	AR
Myasthenic syndrome, congenital, 13, with tubular aggregates	*DPAGT1*	AR
Developmental delay with short stature, dysmorphic facial features, and sparse hair	*DPH1*	AR
Cone-rod dystrophy 21	*DRAM2*	AR
Ciliary dyskinesia, primary, 21	*DRC1*	AR
Ehlers-Danlos syndrome, musculocontractural type 2	*DSE*	AR
Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE	*DSG1*	AR
Cardiomyopathy, dilated, with woolly hair and keratoderma	*DSP*	AR
Neuropathy, hereditary sensory and autonomic, type VI	*DST*	AR
Spastic paraplegia 23, autosomal recessive	*DSTYK*	AR
Hermansky-Pudlak syndrome 7	*DTNBP1*	AR
Dyggve-Melchior-Clausen disease	*DYM*	AR
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*	AR/ DR
Short-rib thoracic dysplasia 8 with or without polydactyly	*DYNC2I1*	AR
Short-rib thoracic dysplasia 11 with or without polydactyly	*DYNC2I2*	AR
Short-rib thoracic dysplasia 15 with polydactyly	*DYNC2LI1*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 2	*DYSF*	AR
Polycystic kidney disease 5	*DZIP1L*	AR
Combined oxidative phosphorylation deficiency 12	*EARS2*	AR
MEND syndrome	*EBP*	XLR
Arthrogryposis, distal, type 5D	*ECEL1*	AR
Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	*ECHS1*	AR
Urbach-Wiethe disease	*ECM1*	AR
Ectodermal dysplasia 1, hypohidrotic, X-linked	*EDA*	XLR
Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive	*EDAR*	AR
Congenital disorder of glycosylation, type IIv	*EDEM3*	AR
Waardenburg syndrome, type 4B	*EDN3*	AD/ AR
Cutis laxa, autosomal recessive, type IB	*EFEMP2*	AR
Shwachman-Diamond syndrome 2	*EFL1*	AR
Wolcott-Rallison syndrome	*EIF2AK3*	AR
Pulmonary venoocclusive disease 2	*EIF2AK4*	AR
Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure	*EIF2B1*	AR
Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure	*EIF2B5*	-
MEHMO syndrome	*EIF2S3*	XLR
Combined oxidative phosphorylation deficiency 17	*ELAC2*	AR
Ichthyosis, spastic quadriplegia, and impaired intellectual development	*ELOVL4*	AR
Dysautonomia, familial	*ELP1*	AR

Disorders	Genes	Inheritance
Intellectual developmental disorder, autosomal recessive 58	*ELP2*	AR
Neurodevelopmental disorder with dysmorphic facies and variable seizures	*EMC10*	AR
Emery-Dreifuss muscular dystrophy 1, X-linked	*EMD*	XLR
Bowen-Conradi syndrome	*EMG1*	AR
Band heterotopia	*EML1*	AR
Hypophosphatemic rickets, autosomal recessive, 2	*ENPP1*	AR
Spastic paraplegia 64, autosomal recessive	*ENTPD1*	AR
Adams-Oliver syndrome 4	*EOGT*	AR
Diarrhea 5, with tufting enteropathy, congenital	*EPCAM*	AR
Vici syndrome	*EPG5*	AR
Epilepsy, progressive myoclonic 2A (Lafora)	*EPM2A*	AR
Lethal congenital contractural syndrome 2	*ERBB3*	AR
Cerebrooculofacioskeletal syndrome 2	*ERCC2*	AR
Xeroderma pigmentosum, group B	*ERCC3*	AR
Fanconi anemia, complementation group Q	*ERCC4*	AR
Xeroderma pigmentosum, group G	*ERCC5*	AR
Cockayne syndrome, type B	*ERCC6*	AR
Bone marrow failure syndrome 2	*ERCC6L2*	AR
Cockayne syndrome, type A	*ERCC8*	AR
Spastic paraplegia 18B, autosomal recessive	*ERLIN2*	AR
Roberts-SC phocomelia syndrome	*ESCO2*	AR
Deafness, autosomal recessive 36	*ESPN*	AR
Deafness, autosomal recessive 35	*ESRRB*	AR
Glutaric acidemia IIA	*ETFA*	AR
Glutaric acidemia IIB	*ETFB*	AR
Glutaric acidemia IIC	*ETFDH*	AR
Ethylmalonic encephalopathy	*ETHE1*	AR
Ellis-van Creveld syndrome	*EVC*	AR
Ellis-van Creveld syndrome	*EVC2*	AR
Pontocerebellar hypoplasia, type 1B	*EXOSC3*	AR
Pontocerebellar hypoplasia, type 1C	*EXOSC8*	AR
Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive	*EXPH5*	AR
Immunoskeletal dysplasia with neurodevelopmental abnormalities	*EXTL3*	AR
Retinitis pigmentosa 25	*EYS*	AR
Factor X deficiency	*F10*	AR
Factor XII deficiency	*F12*	AR
Factor XIIIA deficiency	*F13A1*	AR
Factor XIIIB deficiency	*F13B*	AR
Dysprothrombinemia	*F2*	AR
Factor V deficiency	*F5*	AR
Factor VII deficiency	*F7*	AR
Hemophilia A	*F8*	XLR
Hemophilia B	*F9*	XLR
Spastic paraplegia 35, autosomal recessive	*FA2H*	AR
Tyrosinemia, type I	*FAH*	AR
Retinitis pigmentosa 28	*FAM161A*	AR
Raine syndrome	*FAM20C*	AR

Disorders	Genes	Inheritance
Fanconi anemia, complementation group A	*FANCA*	AR
Fanconi anemia, complementation group B	*FANCB*	XLR
Fanconi anemia, complementation group C	*FANCC*	AR
Fanconi anemia, complementation group D2	*FANCD2*	AR
Fanconi anemia, complementation group E	*FANCE*	AR
Fanconi anemia, complementation group F	*FANCF*	AR
Fanconi anemia, complementation group G	*FANCG*	AR
Fanconi anemia, complementation group I	*FANCI*	AR
Fanconi anemia, complementation group L	*FANCL*	AR
Combined oxidative phosphorylation deficiency 14	*FARS2*	AR
Combined oxidative phosphorylation deficiency 44	*FASTKD2*	AR
Hennekam lymphangiectasia-lymphedema syndrome 2	*FAT4*	AR
Cutis laxa, autosomal recessive, type IA	*FBLN5*	AR
Fructose-1,6-bisphosphatase deficiency	*FBP1*	AR
Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	*FBXL4*	AR
Parkinson disease 15, autosomal recessive	*FBXO7*	AR
Kindler syndrome	*FERMT1*	AR
Leukocyte adhesion deficiency, type III	*FERMT3*	AR
Afibrinogenemia, congenital	*FGA*	AR
Afibrinogenemia, congenital	*FGB*	AR
Aarskog-Scott syndrome	*FGD1*	XLR
Intellectual developmental disorder, X-linked syndromic 16	*FGD1*	XLR
Charcot-Marie-Tooth disease, type 4H	*FGD4*	AR
Tumoral calcinosis, hyperphosphatemic, familial, 2	*FGF23*	AR
Deafness, congenital with inner ear agenesis, microtia, and microdontia	*FGF3*	AR
Afibrinogenemia, congenital	*FGG*	AR
Fumarase deficiency	*FH*	AR
Emery-Dreifuss muscular dystrophy 6, X-linked	*FHL1*	XLR
Yunis-Varon syndrome	*FIG4*	AR
Siddiqi syndrome	*FITM2*	AR
Bruck syndrome 1	*FKBP10*	AR
Ehlers-Danlos syndrome, kyphoscoliotic type, 2	*FKBP14*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	*FKRP*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	*FKTN*	AR
Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency	*FLAD1*	AR
Peeling skin syndrome 6	*FLG2*	AR
Bleeding disorder, platelet-type, 21	*FLI1*	AD/AR
FG syndrome 2	*FLNA*	XL
Spondylocarpotarsal synostosis syndrome	*FLNB*	AR
Ataxia, posterior column, with retinitis pigmentosa	*FLVCR1*	AR
Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	*FLVCR2*	AR
Trimethylaminuria	*FMO3*	AR
Fragile X syndrome	*FMR1*	XLD
Immunodeficiency 93 and hypertrophic cardiomyopathy	*FNIP1*	AR

Disorders	Genes	Inheritance
Neurodegeneration due to cerebral folate transport deficiency	*FOLR1*	AR
Anterior segment dysgenesis 2, multiple subtypes	*FOXE3*	AR
T-cell immunodeficiency, congenital alopecia, and nail dystrophy	*FOXN1*	AR
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	*FOXP3*	XLR
Mitochondrial complex I deficiency, nuclear type 19	*FOXRED1*	AR
Fraser syndrome 1	*FRAS1*	AR
Bifid nose with or without anorectal and renal anomalies	*FREM1*	AR
Intellectual developmental disorder, X-linked 104	*FRMPD4*	XL
Developmental and epileptic encephalopathy 37	*FRRS1L*	AR
Glutamate formiminotransferase deficiency	*FTCD*	AR
Growth retardation, developmental delay, facial dysmorphism	*FTO*	AR
Intellectual developmental disorder, X-linked 9	*FTSJ1*	XLR
Fucosidosis	*FUCA1*	AR
Congenital disorder of glycosylation with defective fucosylation 1	*FUT8*	AR
Cataract 18, autosomal recessive	*FYCO1*	AR
Glycogen storage disease Ia	*G6PC1*	AR
Dursun syndrome	*G6PC3*	AR
Hemolytic anemia, G6PD deficient (favism)	*G6PD*	XL
Glycogen storage disease II	*GAA*	AR
Developmental and epileptic encephalopathy 89	*GAD1*	AR
Krabbe disease	*GALC*	AR
Galactose epimerase deficiency	*GALE*	AR
Galactokinase deficiency with cataracts	*GALK1*	AR
Mucopolysaccharidosis IVA	*GALNS*	AR
Congenital disorder of glycosylation, type IIt	*GALNT2*	AR
Tumoral calcinosis, hyperphosphatemic, familial, 1	*GALNT3*	AR
Galactosemia	*GALT*	AR
Cerebral creatine deficiency syndrome 2	*GAMT*	AR
Giant axonal neuropathy-1	*GAN*	AR
Ciliary dyskinesia, primary, 33	*GAS8*	AR
Anemia, X-linked, with/without neutropenia and/or platelet abnormalities	*GATA1*	XLR
Thrombocytopenia with beta-thalassemia, X-linked	*GATA1*	XLR
Thrombocytopenia, X-linked, with or without dyserythropoietic anemia	*GATA1*	XLR
Cerebral creatine deficiency syndrome 3	*GATM*	AR
Gaucher disease, perinatal lethal	*GBA1*	AR
Spastic paraplegia 46, autosomal recessive	*GBA2*	AR
Glycogen storage disease IV	*GBE1*	AR
Glutaricaciduria, type I	*GCDH*	AR
Dystonia, DOPA-responsive	*GCH1*	AD/ AR
Charcot-Marie-Tooth disease, recessive intermediate, A	*GDAP1*	AR
Spinocerebellar ataxia, autosomal recessive 27	*GDAP2*	AR
Right atrial isomerism (Ivemark)	*GDF1*	AR
Acromesomelic dysplasia 2A	*GDF5*	AR

Disorders	Genes	Inheritance
Combined oxidative phosphorylation deficiency 1	*GFM1*	AR
Myasthenia, congenital, 12, with tubular aggregates	*GFPT1*	AR
Vitamin K-dependent clotting factors, combined deficiency of, 1	*GGCX*	AR
Laron dwarfism	*GHR*	AR
Deafness, autosomal recessive 15	*GIPC3*	AR
Deafness, autosomal recessive 1A	*GJB2*	AR/ DD
Leukodystrophy, hypomyelinating, 2	*GJC2*	AR
Glycerol kinase deficiency	*GK*	XLR
Fabry disease	*GLA*	XL
Mucopolysaccharidosis type IVB (Morquio)	*GLB1*	AR
Glycine encephalopathy1	*GLDC*	AR
Lethal congenital contracture syndrome 11	*GLDN*	AR
Congenital arthrogryposis with anterior horn cell disease	*GLE1*	AR
Diabetes mellitus, neonatal, with congenital hypothyroidism	*GLIS3*	AR
Hyperekplexia 2	*GLRB*	AR
Developmental and epileptic encephalopathy 71	*GLS*	AR
Global developmental delay, progressive ataxia, and elevated glutamine	*GLS*	AR
D-glyceric aciduria	*GLYCTK*	AR
GM2-gangliosidosis, AB variant	*GM2A*	AR
Alacrima, achalasia, and impaired intellectual development syndrome	*GMPPA*	AR
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14	*GMPPB*	AR
Achromatopsia 4	*GNAT2*	-
Lodder-Merla syndrome, type 1, with impaired intellectual development and cardiac arrhythmia	*GNB5*	AR
Nonaka myopathy	*GNE*	AR
Rhizomelic chondrodysplasia punctata, type 2	*GNPAT*	AR
Mucolipidosis III alpha/beta	*GNPTAB*	AR
Mucolipidosis III gamma	*GNPTG*	AR
Mucopolysaccharidosis type IIID	*GNS*	AR
Geroderma osteodysplasticum	*GORAB*	AR
Epilepsy, progressive myoclonic 6	*GOSR2*	AR
Bernard-Soulier syndrome, type A1 (recessive)	*GP1BA*	AR
Bernard-Soulier syndrome, type B	*GP1BB*	AR
Bleeding disorder, platelet-type, 11	*GP6*	AR
Bernard-Soulier syndrome, type C	*GP9*	AR
Glycosylphosphatidylinositol biosynthesis defect 15	*GPAA1*	AR
Simpson-Golabi-Behmel syndrome, type 1	*GPC3*	XLR
Keipert syndrome	*GPC4*	XLR
Omodysplasia 1	*GPC6*	AR
Hypertriglyceridemia, transient infantile	*GPD1*	AR
Molybdenum cofactor deficiency C	*GPHN*	AR
Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency	*GPI*	AR
Hyperlipoproteinemia, type 1D	*GPIHBP1*	AR
Ocular albinism, type I, Nettleship-Falls type	*GPR143*	XL

Disorders	Genes	Inheritance
Night blindness, congenital stationary (complete), 1E, autosomal recessive	*GPR179*	AR
Chudley-McCullough syndrome	*GPSM2*	AR
Neurodevelopmental disorder with microcephaly and spastic paraplegia	*GPT2*	AR
Intellectual developmental disorder, X-linked syndromic, Wu type	*GRIA3*	XLR
Intellectual developmental disorder, autosomal recessive 6	*GRIK2*	AR
Fraser syndrome 3	*GRIP1*	AR
Spinocerebellar ataxia, autosomal recessive 13	*GRM1*	AR
Night blindness, congenital stationary (complete), 1B, autosomal recessive	*GRM6*	AR
Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities	*GRM7*	AR
Ceroid lipofuscinosis, neuronal, 11	*GRN*	AR
Deafness, autosomal recessive 25	*GRXCR1*	AR
Glutathione synthetase deficiency	*GSS*	AR
Trichothiodystrophy 3, photosensitive	*GTF2H5*	AR
Jaberi-Elahi syndrome	*GTPBP2*	AR
Combined oxidative phosphorylation deficiency 23	*GTPBP3*	AR
Moyamoya 6 with achalasia	*GUCY1A1*	AR
Meconium ileus	*GUCY2C*	AR
Leber congenital amaurosis 1	*GUCY2D*	AR
Mucopolysaccharidosis VII	*GUSB*	AR
Glycogen storage disease XV	*GYG1*	AR
Polyglucosan body myopathy 2	*GYG1*	AR
Glycogen storage disease 0, liver	*GYS2*	AR
Vertebral, cardiac, renal, and limb defects syndrome 1	*HAAO*	AR
Congenital myopathy 11	*HACD1*	AR
Spastic paraplegia and psychomotor retardation with or without seizures	*HACE1*	AR
3-hydroxyacyl-CoA dehydrogenase deficiency	*HADH*	AR
LCHAD deficiency	*HADHA*	AR
Mitochondrial trifunctional protein deficiency 2	*HADHB*	-
Hemochromatosis, type 2B	*HAMP*	AR
Neutropenia, severe congenital 3, autosomal recessive	*HAX1*	AR
Thalassemia, alpha	*HBA1*	AR
	HBA2	AR
Thalassemia, beta	*HBB*	.
Methylmalonic aciduria and homocysteinemia, cblX type	*HCFC1*	XLR
Megalencephalic leukoencephalopathy with subcortical cysts 2A	*HEPACAM*	AR
Macrocephaly, dysmorphic facies, and psychomotor retardation	*HERC1*	AR
Intellectual developmental disorder, autosomal recessive 38	*HERC2*	AR
Spondylocostal dysostosis 4, autosomal recessive	*HES7*	AR
Septooptic dysplasia	*HESX1*	AD/ AR
Tay-Sachs disease	*HEXA*	AR
Sandhoff disease, infantile, juvenile, and adult forms	*HEXB*	AR
Hemochromatosis, type 1	*HFE*	AR

Disorders	Genes	Inheritance
Mucopolysaccharidosis type IIIC (Sanfilippo C)	*HGSNAT*	AR
3-hydroxyisobutryl-CoA hydrolase deficiency	*HIBCH*	AR
Neuromyotonia and axonal neuropathy, autosomal recessive	*HINT1*	AR
Hemochromatosis, type 2A	*HJV*	AR
Neuropathy, hereditary motor and sensory, Russe type	*HK1*	AR
Holocarboxylase synthetase deficiency	*HLCS*	AR
HMG-CoA lyase deficiency	*HMGCL*	AR
HMG-CoA synthase-2 deficiency	*HMGCS2*	AR
Heme oxygenase-1 deficiency	*HMOX1*	AR
Oculoauricular syndrome	*HMX1*	AR
Athabaskan brainstem dysgenesis syndrome	*HOXA1*	AR
Tyrosinemia, type III	*HPD*	AR
Cranioosteoarthropathy	*HPGD*	AR
Lesch-Nyhan syndrome	*HPRT1*	XLR
Hermansky-Pudlak syndrome 1	*HPS1*	AR
Hermansky-Pudlak syndrome 3	*HPS3*	AR
Hermansky-Pudlak syndrome 4	*HPS4*	AR
Hermansky-Pudlak syndrome 5	*HPS5*	AR
Hermansky-Pudlak syndrome 6	*HPS6*	AR
Urofacial syndrome 1	*HPSE2*	AR
Apparent mineralocorticoid excess	*HSD11B2*	AR
D-bifunctional protein deficiency	*HSD17B4*	AR
Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	*HSD3B2*	AR
Bile acid synthesis defect, congenital, 1	*HSD3B7*	AR
Leukodystrophy, hypomyelinating, 4	*HSPD1*	AR
Schwartz-Jampel syndrome, type 1	*HSPG2*	AR
CARASIL syndrome	*HTRA1*	AR
3-methylglutaconic aciduria, type VIII	*HTRA2*	AR
Intellectual developmental disorder, X-linked syndromic, Turner type	*HUWE1*	XL
Leukodystrophy, hypomyelinating, 5	*HYCC1*	AR
Ciliary dyskinesia, primary, 5	*HYDIN*	AR
Hydrolethalus syndrome	*HYLS1*	AR
Growth retardation, impaired intellectual development, hypotonia, and hepatopathy	*IARS1*	AR
Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	*IARS2*	AR
Multiple mitochondrial dysfunctions syndrome 3	*IBA57*	AR
Immunodeficiency, common variable, 1	*ICOS*	AR
Mucopolysaccharidosis II	*IDS*	XLR
Mucopolysaccharidosis Ih	*IDUA*	AR
Microcephaly, epilepsy, and diabetes syndrome	*IER3IP1*	AR
Immunodeficiency 95	*IFIH1*	AR
Immunodeficiency 106, susceptibility to viral infections	*IFNAR1*	AR
Immunodeficiency 27A, mycobacteriosis, AR	*IFNGR1*	AR
Immunodeficiency 28, mycobacteriosis	*IFNGR2*	AR
Cranioectodermal dysplasia 1	*IFT122*	AR

Disorders	Genes	Inheritance
Short-rib thoracic dysplasia 9 with or without polydactyly	*IFT140*	AR
Short-rib thoracic dysplasia 10 with or without polydactyly	*IFT172*	AR
Short-rib thoracic dysplasia 2 with or without polydactyly	*IFT80*	AR
Short-rib thoracic dysplasia 19 with or without polydactyly	*IFT81*	AR
Insulin-like growth factor I deficiency	*IGF1*	AR
Insulin-like growth factor I, resistance to	*IGF1R*	AD/ AR
Retinal arterial macroaneurysm with supravalvular pulmonic stenosis	*IGFBP7*	AR
Agammaglobulinemia 1	*IGHM*	AR
Neuronopathy, distal hereditary motor, autosomal recessive 1	*IGHMBP2*	AR
Immunodeficiency 15B	*IKBKB*	AR
Ectodermal dysplasia and immunodeficiency 1	*IKBKG*	XLR
Immunodeficiency 33	*IKBKG*	XLR
Inflammatory bowel disease 28, early onset, autosomal recessive	*IL10RA*	AR
Inflammatory bowel disease 25, early onset, autosomal recessive	*IL10RB*	AR
Craniosynostosis and dental anomalies	*IL11RA*	AR
Immunodeficiency 29, mycobacteriosis	*IL12B*	AR
Immunodeficiency 30	*IL12RB1*	AR
Immunodeficiency 51	*IL17RA*	AR
Intellectual developmental disorder, X-linked 21	*IL1RAPL1*	XLR
Interleukin 1 receptor antagonist deficiency	*IL1RN*	AR
Immunodeficiency 56	*IL21R*	AR
Immunodeficiency 41 with lymphoproliferation and autoimmunity	*IL2RA*	AR
Severe combined immunodeficiency, X-linked	*IL2RG*	XLR
Immunodeficiency 104, severe combined	*IL7R*	AR
Deafness, autosomal recessive 42	*ILDR1*	AR
Macular dystrophy, vitelliform, 4	*IMPG1*	AD/ AR
Retinitis pigmentosa 56	*IMPG2*	AR
Joubert syndrome 1	*INPP5E*	AR
Muscular dystrophy, congenital, with cataracts and intellectual disability	*INPP5K*	AR
Opsismodysplasia	*INPPL1*	AR
Donohue syndrome	*INSR*	AR
Nephronophthisis 2, infantile	*INVS*	AR
Senior-Loken syndrome 5	*IQCB1*	AR
Multiple mitochondrial dysfunctions syndrome 4	*ISCA2*	AR
Autoimmune disease, multisystem, with facial dysmorphism	*ITCH*	AR
Glanzmann thrombasthenia 1	*ITGA2B*	AR
Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome	*ITGA3*	AR
Muscular dystrophy, congenital, due to ITGA7 deficiency	*ITGA7*	AR
Renal hypodysplasia/aplasia 1	*ITGA8*	AR
Leukocyte adhesion deficiency	*ITGB2*	AR
Glanzmann thrombasthenia 2	*ITGB3*	AR
Epidermolysis bullosa, junctional 5B, with pyloric atresia	*ITGB4*	AR

Disorders	Genes	Inheritance
Lymphoproliferative syndrome 1	*ITK*	AR
Developmental and epileptic encephalopathy 35	*ITPA*	AR
Gillespie syndrome	*ITPR1*	AD/ AR
Isovaleric acidemia	*IVD*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 27	*JAG2*	AR
Neutropenia, severe congenital, 6, autosomal recessive	*JAGN1*	AR
SCID, autosomal recessive, T-negative/B-positive type	*JAK3*	AR
Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	*JAM3*	AR
Naxos disease	*JUP*	AR
Leukoencephalopathy, progressive, infantile-onset, with or without deafness	*KARS1*	AR
Lissencephaly 6, with microcephaly	*KATNB1*	AR
Jervell and Lange-Nielsen syndrome 2	*KCNE1*	AR
Bartter syndrome, type 2	*KCNJ1*	AR
SESAME syndrome	*KCNJ10*	AR
Hyperinsulinemic hypoglycemia, familial, 2	*KCNJ11*	AD/AR
Jervell and Lange-Nielsen syndrome	*KCNQ1*	AR
Retinal cone dystrophy 3B	*KCNV2*	AR
Epilepsy, progressive myoclonic 3, with or without intracellular inclusions	*KCTD7*	AR
Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type	*KDM5C*	XLR
Erythrokeratodermia variabilis et progressiva 4	*KDSR*	AR
Cornea plana 2, autosomal recessive	*KERA*	AR
Short-rib thoracic dysplasia 14 with polydactyly	*KIAA0586*	AR
Joubert syndrome 38	*KIAA0753*	AR
Orofaciodigital syndrome XV	*KIAA0753*	AR
Short-rib thoracic dysplasia 21 without polydactyly	*KIAA0753*	AR
Microcephaly 20, primary, autosomal recessive	*KIF14*	AR
Spastic paraplegia 30, autosomal recessive	*KIF1A*	AD/ AR
Spastic ataxia 2, autosomal recessive	*KIF1C*	AR
Hydrolethalus syndrome 2	*KIF7*	AR
Goldberg-Shprintzen megacolon syndrome	*KIFBP*	AR
Retinitis pigmentosa 69	*KIZ*	AR
Nemaline myopathy 8, autosomal recessive	*KLHL40*	AR
Nemaline myopathy 9	*KLHL41*	AR
PERCHING syndrome	*KLHL7*	AR
Fletcher factor (prekallikrein) deficiency	*KLKB1*	AR
High molecular weight kininogen deficiency	*KNG1*	AR
Microcephaly 4, primary, autosomal recessive	*KNL1*	AR
Intellectual developmental disorder, autosomal recessive 41	*KPTN*	AR
Epidermolytic hyperkeratosis 2	*KRT10*	-
Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive	*KRT14*	AR
Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive	*KRT5*	AR
None	*KRT8*	-
Ectodermal dysplasia 4, hair/nail type	*KRT85*	AR
Myopathy, myofibrillar, 7	*KY*	AR

Disorders	Genes	Inheritance
Hydroxykynureninuria	*KYNU*	AR
Vertebral, cardiac, renal, and limb defects syndrome 2	*KYNU*	AR
MASA syndrome	*L1CAM*	XLR
L-2-hydroxyglutaric aciduria	*L2HGDH*	AR
Poretti-Boltshauser syndrome	*LAMA1*	AR
Muscular dystrophy, congenital, merosin deficient or partially deficient	*LAMA2*	AR
Lissencephaly 5	*LAMB1*	AR
Pierson syndrome	*LAMB2*	AR
Epidermolysis bullosa, junctional 1B, severe	*LAMB3*	AR
Cortical malformations, occipital	*LAMC3*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6	*LARGE1*	AR
Alazami syndrome	*LARP7*	AR
Infantile liver failure syndrome 1	*LARS1*	AR
Perrault syndrome 4	*LARS2*	AR
Immunodeficiency 52	*LAT*	AR
Greenberg skeletal dysplasia	*LBR*	AR
Leber congenital amaurosis 5	*LCA5*	AR
Norum disease	*LCAT*	AR
Lactase deficiency, congenital	*LCT*	AR
Glycogen storage disease XI	*LDHA*	AR
Lactate dehydrogenase-B deficiency	*LDHB*	.
Hypercholesterolemia, familial, 1	*LDLR*	AD/ AR
LDL cholesterol level QTL2	*LDLR*	AD/ AR
Hypercholesterolemia, familial, 4	*LDLRAP1*	AR
Obesity, morbid, due to leptin deficiency	*LEP*	AR
Spondylocostal dysostosis 3, autosomal recessive	*LFNG*	AR
Intellectual developmental disorder with muscle tone abnormalities and distal skeletal defects	*LGI3*	AR
Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect	*LGI4*	AR
Deafness, autosomal recessive 67	*LHFPL5*	AR
Pituitary hormone deficiency, combined, 3	*LHX3*	AR
Hyperglycinemia, lactic acidosis, and seizures	*LIAS*	AR
Stuve-Wiedemann syndrome/ Schwartz-Jampel type 2 syndrome	*LIFR*	AR
LIG4 syndrome	*LIG4*	AR
Intellectual developmental disorder, autosomal recessive 27	*LINS1*	AR
Cholesteryl ester storage disease	*LIPA*	AR
Hepatic lipase deficiency	*LIPC*	AR
Lipodystrophy, familial partial, type 6	*LIPE*	AR
Lipoyltransferase 1 deficiency	*LIPT1*	AR
Combined factor V and VIII deficiency	*LMAN1*	AR
Acheiropody	*LMBR1*	AR
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*	AR
Lipase deficiency, combined	*LMF1*	AR
Nemaline myopathy 10	*LMOD3*	AR
CODAS syndrome	*LONP1*	AR
Deafness, autosomal recessive 77	*LOXHD1*	AR
Myoglobinuria, acute recurrent, autosomal recessive	*LPIN1*	AR

Disorders	Genes	Inheritance
Myoglobinuria, acute recurrent, autosomal recessive	*LPIN1*	AR
Majeed syndrome	*LPIN2*	.
Lipoprotein lipase deficiency	*LPL*	AR
Leber congenital amaurosis 14	*LRAT*	AR
Immunodeficiency, common variable, 8, with autoimmunity	*LRBA*	AR
Urofacial syndrome 2	*LRIG2*	AR
Albinism, oculocutaneous, type VII	*LRMDA*	AR
Donnai-Barrow syndrome	*LRP2*	AR
Cenani-Lenz syndactyly syndrome	*LRP4*	AR
Osteoporosis-pseudoglioma syndrome	*LRP5*	AR
Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)	*LRPPRC*	AR
Osteosclerotic metaphyseal dysplasia	*LRRK1*	AR
Charcot-Marie-Tooth disease, axonal, type 2P	*LRSAM1*	AD/ AR
Deafness, autosomal recessive 63	*LRTOMT*	AR
Weill-Marchesani syndrome 3, recessive	*LTBP2*	AR
Dental anomalies and short stature	*LTBP3*	AR
Cutis laxa, autosomal recessive, type IC	*LTBP4*	AR
Mitochondrial complex III deficiency, nuclear type 8	*LYRM7*	AR
Chediak-Higashi syndrome	*LYST*	AR
Bardet-Biedl syndrome 17	*LZTFL1*	AR
Noonan syndrome 2	*LZTR1*	AR
Cerebellar, ocular, craniofacial, and genital syndrome	*MAB21L1*	AR
Bartter syndrome, type 5, antenatal, transient	*MAGED2*	XLR
Nephrotic syndrome, type 15	*MAGI2*	AR
Retinitis pigmentosa 62	*MAK*	AR
Immunodeficiency 12	*MALT1*	AR
Hypospadias 2, X-linked	*MAMLD1*	XLR
Rafiq syndrome	*MAN1B1*	AR
Mannosidosis, alpha-, types I and II	*MAN2B1*	AR
Mannosidosis, beta	*MANBA*	AR
Brunner syndrome	*MAOA*	XLR
Centronuclear myopathy 6 with fiber-type disproportion	*MAP3K20*	AR
Nephronophthisis 20	*MAPKBP1*	AR
Interstitial lung and liver disease	*MARS1*	AR
Spastic ataxia 3, autosomal recessive	*MARS2*	AR
Deafness, autosomal recessive 49	*MARVELD2*	AR
3MC syndrome 1	*MASP1*	AR
Methionine adenosyltransferase deficiency, autosomal recessive	*MAT1A*	AD/ AR
Intellectual developmental disorder, autosomal recessive 57	*MBOAT7*	AR
IFAP syndrome with or without BRESHECK syndrome	*MBTPS2*	XLR
Albinism, oculocutaneous, type II, modifier of	*MC1R*	AR
Glucocorticoid deficiency, due to ACTH unresponsiveness	*MC2R*	AR
3-Methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*	AR
Methylmalonyl-CoA epimerase deficiency	*MCEE*	AR
Factor V and factor VIII, combined deficiency of	*MCFD2*	-

Disorders	Genes	Inheritance
Ciliary dyskinesia, primary, 42	*MCIDAS*	AR
Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development	*MCM3AP*	AR
Immunodeficiency 54	*MCM4*	AR
Mucolipidosis IV	*MCOLN1*	AR
Microcephaly 1, primary, autosomal recessive	*MCPH1*	AR
Encephalopathy, neonatal severe	*MECP2*	XLR
Lujan-Fryns syndrome	*MED12*	XLR
Microcephaly, postnatal progressive, with seizures and brain atrophy	*MED17*	AR
Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy	*MED23*	AR
Basel-Vanagait-Smirin-Yosef syndrome	*MED25*	AR
Familial Mediterranean fever, AR	*MEFV*	AR
Congenital myopathy 10A, severe variant	*MEGF10*	AR
Carpenter syndrome 2	*MEGF8*	AR
Retinitis pigmentosa 38	*MERTK*	AR
Osteogenesis imperfecta, type XX	*MESD*	AR
Spondylocostal dysostosis 2, autosomal recessive	*MESP2*	AR
Intellectual developmental disorder, autosomal recessive 44	*METTL23*	AR
Charcot-Marie-Tooth disease, axonal, type 2A2B	*MFN2*	AR
Microphthalmia, isolated 5	*MFRP*	AR
Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities	*MFSD2A*	AR
Ceroid lipofuscinosis, neuronal, 7	*MFSD8*	AR
Congenital disorder of glycosylation, type IIa	*MGAT2*	AR
Mitochondrial DNA depletion syndrome 11	*MGME1*	AR
Keutel syndrome	*MGP*	AR
Combined oxidative phosphorylation deficiency 37	*MICOS13*	AR
Myopathy with extrapyramidal signs	*MICU1*	AR
Opitz GBBB syndrome	*MID1*	XLR
Pontocerebellar hypoplasia, type 16	*MINPP1*	AR
COMMAD syndrome	*MITF*	AR
McKusick-Kaufman syndrome	*MKKS*	AR
Meckel syndrome 1	*MKS1*	AR
Megalencephalic leukoencephalopathy with subcortical cysts 1	*MLC1*	AR
Mismatch repair cancer syndrome 1	*MLH1*	AR
Malonyl-CoA decarboxylase deficiency	*MLYCD*	AR
Methylmalonic aciduria, vitamin B12-responsive, cblA type	*MMAA*	AR
Methylmalonic aciduria, vitamin B12-responsive, cblB type	*MMAB*	AR
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	AR
Methylmalonic aciduria and homocystinuria, cblD type	*MMADHC*	AR
Charcot-Marie-Tooth disease, axonal, type 2T	*MME*	AD/ AR
Multicentric osteolysis, nodulosis, and arthropathy	*MMP2*	AR
Heterotaxy, visceral, 7, autosomal	*MMP21*	AR
Methylmalonic aciduria, mut(0) type	*MMUT*	AR
Molybdenum cofactor deficiency A	*MOCS1*	AR
Molybdenum cofactor deficiency B	*MOCS2*	AR
Congenital disorder of glycosylation, type IIb	*MOGS*	AR

Disorders	Genes	Inheritance
Congenital disorder of glycosylation, type If	*MPDU1*	AR
Hydrocephalus, congenital, 2, with or without brain or eye anomalies	*MPDZ*	AR
Congenital disorder of glycosylation, type Ib	*MPI*	AR
Thrombocytopenia, anemia, and myelofibrosis	*MPIG6B*	AR
Thrombocytopenia, congenital amegakaryocytic	*MPL*	AR
Trichothiodystrophy 4, nonphotosensitive	*MPLKIP*	AR
Myeloperoxidase deficiency	*MPO*	AR
Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	*MPV17*	AR
Dejerine-Sottas disease	*MPZ*	AD/ AR
Deafness, autosomal recessive 111	*MPZL2*	AR
Glucocorticoid deficiency 2	*MRAP*	AR
Ataxia-telangiectasia-like disorder 1	*MRE11*	AR
Combined oxidative phosphorylation deficiency 16	*MRPL44*	AR
Combined oxidative phosphorylation deficiency 5	*MRPS22*	AR
Mismatch repair cancer syndrome 2	*MSH2*	AR
Mismatch repair cancer syndrome 3	*MSH6*	AR
Immunodeficiency 50	*MSN*	XLR
Deafness, autosomal recessive 74	*MSRB3*	AR
Myopathy, mitochondrial, and ataxia	*MSTO1*	AD/AR
Combined oxidative phosphorylation deficiency 15	*MTFMT*	AR
Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	*MTHFD1*	AR
Homocystinuria due to MTHFR deficiency	*MTHFR*	AR
Myopathy, centronuclear, X-linked	*MTM1*	XLR
Charcot-Marie-Tooth disease, type 4B1	*MTMR2*	AR
Combined oxidative phosphorylation deficiency 10	*MTO1*	AR
Homocystinuria-megaloblastic anemia, cblG complementation type	*MTR*	AR
Combined oxidative phosphorylation deficiency 7	*MTRFR*	AR
Homocystinuria-megaloblastic anemia, cbl E type	*MTRR*	AR
Abetalipoproteinemia	*MTTP*	AR
Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency	*MUSK*	AR
Mevalonic aciduria	*MVK*	AR
Immunodeficiency 68	*MYD88*	AR
Congenital myopathy 6 with ophthalmoplegia	*MYH2*	AD/ AR
Carey-Fineman-Ziter syndrome	*MYMK*	AR
Deafness, autosomal recessive 3	*MYO15A*	AR
Glomerulosclerosis, focal segmental, 6	*MYO1E*	AR
Deafness, autosomal recessive 30	*MYO3A*	AR
Griscelli syndrome, type 1	*MYO5A*	AR
Diarrhea 2, with microvillus atrophy, with or without cholestasis	*MYO5B*	AR
Usher syndrome, type 1B	*MYO7A*	AR
Basal ganglia calcification, idiopathic, 7, autosomal recessive	*MYORG*	AR
Congenital myopathy 24	*MYPN*	AR
Bone marrow failure syndrome 4	*MYSM1*	AR
Ogden syndrome	*NAA10*	XLD/ XLR
Vertebral, cardiac, renal, and limb defects syndrome 3	*NADSYN1*	AR

Disorders	Genes	Inheritance
Schindler disease, type I	*NAGA*	AR
Mucopolysaccharidosis type IIIB (Sanfilippo B)	*NAGLU*	AR
N-acetylglutamate synthase deficiency	*NAGS*	AR
Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	*NALCN*	AR
Spondyloepimetaphyseal dysplasia, Camera-Genesvieve type	*NANS*	AR
Combined oxidative phosphorylation deficiency 24	*NARS2*	AR
Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2	*NAXD*	AR
Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy	*NAXE*	AR
Short stature, optic nerve atrophy, and Pelger-Huet anomaly	*NBAS*	AR
Gray platelet syndrome	*NBEAL2*	AR
Nijmegen breakage syndrome	*NBN*	AR
Chronic granulomatous disease 1, autosomal recessive	*NCF1*	AR
Chronic granulomatous disease 2, autosomal recessive	*NCF2*	AR
Chronic granulomatous disease 3, autosomal recessive	*NCF4*	AR
Lissencephaly 4 (with microcephaly)	*NDE1*	AR
Norrie disease	*NDP*	XLR
Charcot-Marie-Tooth disease, type 4D	*NDRG1*	AR
Mitochondrial complex I deficiency, nuclear type 12	*NDUFA1*	XLR
Mitochondrial complex I deficiency, nuclear type 22	*NDUFA10*	AR
Mitochondrial complex I deficiency, nuclear type 14	*NDUFA11*	AR
Mitochondrial complex I deficiency, nuclear type 23	*NDUFA12*	AR
Mitochondrial complex I deficiency, nuclear type 10	*NDUFAF2*	AR
Mitochondrial complex I deficiency, nuclear type 16	*NDUFAF5*	AR
Mitochondrial complex I deficiency, nuclear type 17	*NDUFAF6*	AR
Mitochondrial complex I deficiency, nuclear type 30	*NDUFB11*	XL
Mitochondrial complex I deficiency, nuclear type 25	*NDUFB3*	AR
Mitochondrial complex I deficiency, nuclear type 5	*NDUFS1*	AR
Mitochondrial complex I deficiency, nuclear type 6	*NDUFS2*	AR
Mitochondrial complex I deficiency, nuclear type 1	*NDUFS4*	AR
Mitochondrial complex I deficiency, nuclear type 9	*NDUFS6*	AR
Mitochondrial complex I deficiency, nuclear type 3	*NDUFS7*	AR
Mitochondrial complex I deficiency, nuclear type 2	*NDUFS8*	AR
Mitochondrial complex I deficiency, nuclear type 4	*NDUFV1*	AR
Mitochondrial complex I deficiency, nuclear type 7	*NDUFV2*	AR
Nemaline myopathy 2, autosomal recessive	*NEB*	AR
Cleft lip/palate-ectodermal dysplasia syndrome	*NECTIN1*	AR
Ectodermal dysplasia-syndactyly syndrome 1	*NECTIN4*	AR
Charcot-Marie-Tooth disease, type 1F	*NEFL*	AD/ AR
Short-rib thoracic dysplasia 6 with or without polydactyly	*NEK1*	AR/ DR
Renal-hepatic-pancreatic dysplasia 2	*NEK8*	AR
Sialidosis, type I	*NEU1*	AR
Diarrhea 4, malabsorptive, congenital	*NEUROG3*	AR
Multiple mitochondrial dysfunctions syndrome 1	*NFU1*	AR
Neuropathy, hereditary sensory and autonomic, type V	*NGF*	AR

Disorders	Genes	Inheritance
Congenital disorder of deglycosylation 1	*NGLY1*	AR
Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	*NHEJ1*	-
Epilepsy, progressive myoclonic 2B (Lafora)	*NHLRC1*	AR
FINCA syndrome	*NHLRC2*	AR
Cataract 40, X-linked	*NHS*	XL
Ichthyosis, congenital, autosomal recessive 6	*NIPAL4*	AR
Spondylo-megaepiphyseal-metaphyseal dysplasia	*NKX3-2*	AR
Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy	*NKX6-2*	AR
Intellectual developmental disorder, X-linked	*NLGN4X*	XL
Leber congenital amaurosis 9	*NMNAT1*	AR
Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency	*NNT*	AR
Niemann-Pick disease, type C1	*NPC1*	AR
Niemann-pick disease, type C2	*NPC2*	AR
Joubert syndrome 4	*NPHP1*	AR
Meckel syndrome 7	*NPHP3*	AR
Senior-Loken syndrome 4	*NPHP4*	AR
Nephrotic syndrome, type 1	*NPHS1*	AR
Nephrotic syndrome, type 2	*NPHS2*	AR
Acromesomelic dysplasia 1, Maroteaux type	*NPR2*	AR
Boudin-Mortier syndrome	*NPR3*	AR
46XY sex reversal 2, dosage-sensitive	*NR0B1*	XL
Cholestasis, progressive familial intrahepatic, 5	*NR1H4*	AR
Enhanced S-cone syndrome	*NR2E3*	AR
Neurodevelopmental disorder with neuromuscular and skeletal abnormalities	*NRCAM*	AR
CK syndrome	*NSDHL*	XLR
Intellectual developmental disorder, autosomal recessive 5	*NSUN2*	AR
Spastic paraplegia 45, autosomal recessive	*NT5C2*	AR
Anemia, hemolytic, due to UMPH1 deficiency	*NT5C3A*	AR
Calcification of joints and arteries	*NT5E*	AR
Familial adenomatous polyposis 3	*NTHL1*	AR
Insensitivity to pain, congenital, with anhidrosis	*NTRK1*	AR
Mitochondrial complex I deficiency, nuclear type 21	*NUBPL*	AR
Nephrotic syndrome, type 11	*NUP107*	AR
Striatonigral degeneration, infantile	*NUP62*	AR
Nephrotic syndrome, type 12	*NUP93*	AR
Night blindness, congenital stationary (complete), 1A, X-linked	*NYX*	XLR
Gyrate atrophy of choroid and retina with or without ornithinemia	*OAT*	AR
3-M syndrome 2	*OBSL1*	AR
Albinism, brown oculocutaneous	*OCA2*	AR
Pseudo-TORCH syndrome 1	*OCLN*	AR
Lowe syndrome	*OCRL*	XLR
Ciliary dyskinesia, primary, 20	*ODAD1*	AR
Ciliary dyskinesia, primary, 23	*ODAD2*	AR
Ciliary dyskinesia, primary, 30	*ODAD3*	AR

Disorders	Genes	Inheritance
Behr syndrome	*OPA1*	AR
3-methylglutaconic aciduria, type III	*OPA3*	AR
Intellectual developmental disorder, X-linked syndromic, Billuart type	*OPHN1*	XLR
Blue cone monochromacy	*OPN1LW*	XLR
Immunodeficiency 9	*ORAI1*	AR
Meier-Gorlin syndrome 1	*ORC1*	AR
Meier-Gorlin syndrome 3	*ORC6*	AR
Galloway-Mowat syndrome 3	*OSGEP*	AR
Osteopetrosis, autosomal recessive 5	*OSTM1*	AR
Ornithine transcarbamylase deficiency	*OTC*	XL
Deafness, autosomal recessive 22	*OTOA*	AR
Auditory neuropathy, autosomal recessive, 1	*OTOF*	AR
Deafness, autosomal recessive 9	*OTOF*	AR
Deafness, autosomal recessive 18B	*OTOG*	AR
Deafness, autosomal recessive 84B	*OTOGL*	AR
Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies	*OTUD6B*	AR
Osteogenesis imperfecta, type VIII	*P3H1*	AR
Myopia, high, with cataract and vitreoretinal degeneration	*P3H2*	AR
Phenylketonuria	*PAH*	AR
Intellectual developmental disorder, X-linked 30	*PAK3*	XLR
Neurodegeneration with brain iron accumulation 1	*PANK2*	AR
Brachyolmia 4 with mild epiphyseal and metaphyseal changes	*PAPSS2*	AR
Dyskeratosis congenita, autosomal recessive 6	*PARN*	AR
Waardenburg syndrome, type 3	*PAX3*	AD/ AR
Pyruvate carboxylase deficiency	*PC*	AR
Retinitis pigmentosa 54	*PCARE*	AR
Hyperphenylalaninemia, BH4-deficient, D	*PCBD1*	AR
Propionicacidemia	*PCCA*	AR
Propionicacidemia	*PCCB*	AR
Diencephalic-mesencephalic junction dysplasia syndrome 1	*PCDH12*	AR
Usher syndrome, type 1F	*PCDH15*	AR
Developmental and epileptic encephalopathy 9	*PCDH19*	XL
Microcephalic osteodysplastic primordial dwarfism, type II	*PCNT*	AR
Endocrinopathy due to proprotein convertase 1/3 deficiency	*PCSK1*	AR
Low density lipoprotein cholesterol level QTL 1	*PCSK9*	AD
Hypercholesterolemia, familial, 3	*PCSK9*	AD
Spondylometaphyseal dysplasia with cone-rod dystrophy	*PCYT1A*	AR
Retinitis pigmentosa-40	*PDE6B*	AR
Cone dystrophy 4	*PDE6C*	AR
Retinal cone dystrophy 3	*PDE6H*	AD/ AR
Pyruvate dehydrogenase E1-beta deficiency	*PDHB*	AR
Lacticacidemia due to PDX1 deficiency	*PDHX*	AR
Pyruvate dehydrogenase phosphatase deficiency	*PDP1*	AR
Deafness, autosomal recessive 57	*PDZD7*	AR
Prolidase deficiency	*PEPD*	AR

Disorders	Genes	Inheritance
Mitochondrial complex IV deficiency, nuclear type 12	*PET100*	AR
Peroxisome biogenesis disorder 1A (Zellweger)	*PEX1*	AR
Peroxisome biogenesis disorder 6A (Zellweger)	*PEX10*	AR
Peroxisome biogenesis disorder 14B	*PEX11B*	AR
Peroxisome biogenesis disorder 3A (Zellweger)	*PEX12*	AR
Peroxisome biogenesis disorder 11A (Zellweger)	*PEX13*	AR
Peroxisome biogenesis disorder 13A (Zellweger)	*PEX14*	AR
Peroxisome biogenesis disorder 8A (Zellweger)	*PEX16*	AR
Peroxisome biogenesis disorder 12A (Zellweger)	*PEX19*	AR
Peroxisome biogenesis disorder 5A (Zellweger)	*PEX2*	AR
Peroxisome biogenesis disorder 7A (Zellweger)	*PEX26*	AR
Peroxisome biogenesis disorder 10A (Zellweger)	*PEX3*	AR
Peroxisome biogenesis disorder 2A (Zellweger)	*PEX5*	AR
Peroxisome biogenesis disorder 4A (Zellweger)	*PEX6*	AR
Rhizomelic chondrodysplasia punctata, type 1	*PEX7*	AR
Glycogen storage disease VII	*PFKM*	AR
Glycogen storage disease X	*PGAM2*	AR
Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities	*PGAP1*	AR
Hyperphosphatasia with impaired intellectual development syndrome 3	*PGAP2*	AR
Hyperphosphatasia with impaired intellectual development syndrome 4	*PGAP3*	AR
Phosphoglycerate kinase 1 deficiency	*PGK1*	XLR
Congenital disorder of glycosylation, type It	*PGM1*	AR
Immunodeficiency 23	*PGM3*	AR
Borjeson-Forssman-Lehmann syndrome	*PHF6*	XLR
Intellectual developmental disorder, X-linked syndromic, Siderius type	*PHF8*	XLR
Neu-Laxova syndrome 1	*PHGDH*	AR
Muscle glycogenosis	*PHKA1*	XLR
Glycogen storage disease, type IXa1	*PHKA2*	XLR
Glycogen storage disease, type IXa2	*PHKA2*	XLR
Phosphorylase kinase deficiency of liver and muscle, autosomal recessive	*PHKB*	AR
Glycogen storage disease IXc	*PHKG2*	AR
Refsum disease	*PHYH*	AR
Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis	*PI4KA*	AR
Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly	*PIDD1*	AR
Arthrogryposis, distal, with impaired proprioception and touch	*PIEZO2*	AR
Multiple congenital anomalies-hypotonia-seizures syndrome 2	*PIGA*	XLR
Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy	*PIGG*	AR
Glycosylphosphatidylinositol biosynthesis defect 17	*PIGH*	AR
Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures	*PIGK*	AR
CHIME syndrome	*PIGL*	AR
Multiple congenital anomalies-hypotonia-seizures syndrome 1	*PIGN*	AR
Hyperphosphatasia with impaired intellectual development syndrome 2	*PIGO*	AR

Disorders	Genes	Inheritance
Multiple congenital anomalies-hypotonia-seizures syndrome 4	*PIGQ*	AR
Multiple congenital anomalies-hypotonia-seizures syndrome 3	*PIGT*	AR
Hyperphosphatasia with impaired intellectual development syndrome 1	*PIGV*	AR
Immunodeficiency 14B, autosomal recessive	*PIK3CD*	AR
Lethal congenital contractural syndrome 3	*PIP5K1C*	AR
Deafness, autosomal recessive 59	*PJVK*	AR
Heterotaxy, visceral, 8, autosomal	*PKD1L1*	AR
Polycystic kidney disease 4, with or without hepatic disease	*PKHD1*	AR
Pyruvate kinase deficiency	*PKLR*	AR
Ectodermal dysplasia/skin fragility syndrome	*PKP1*	AR
Neurodegeneration with brain iron accumulation 2B	*PLA2G6*	AR
Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	*PLAA*	AR
Developmental and epileptic encephalopathy 12	*PLCB1*	AR
Nephrotic syndrome, type 3	*PLCE1*	AR
Epidermolysis bullosa simplex 5C, with pyloric atresia	*PLEC*	AR
Plasminogen deficiency, type I	*PLG*	AR
Microcephaly and chorioretinopathy, autosomal recessive, 2	*PLK4*	AR
Ehlers-Danlos syndrome, kyphoscoliotic type, 1	*PLOD1*	AR
Bruck syndrome 2	*PLOD2*	AR
Pelizaeus-Merzbacher disease	*PLP1*	XLR
Epilepsy, early-onset, 1, vitamin B6-dependent	*PLPBP*	AR
Congenital disorder of glycosylation, type Ia	*PMM2*	AR
Spinocerebellar ataxia, autosomal recessive 2	*PMPCA*	AR
Mismatch repair cancer syndrome 4	*PMS2*	AR
Microcephaly, seizures, and developmental delay	*PNKP*	AR
Immunodeficiency due to purine nucleoside phosphorylase deficiency	*PNP*	AR
Ichthyosis, congenital, autosomal recessive 10	*PNPLA1*	AR
Neutral lipid storage disease with myopathy	*PNPLA2*	AR
Boucher-Neuhauser syndrome	*PNPLA6*	AR
Mitochondrial myopathy with lactic acidosis	*PNPLA8*	AR
Pyridoxamine 5'-phosphate oxidase deficiency	*PNPO*	AR
Combined oxidative phosphorylation deficiency 13	*PNPT1*	AR
Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	*POC1A*	AR
Cone-rod dystrophy 20	*POC1B*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 21	*POGLUT1*	AR
Pigmentary disorder, reticulate, with systemic manifestations, X-linked	*POLA1*	XLR
Mitochondrial DNA depletion syndrome 4A (Alpers type)	*POLG*	AR
Treacher Collins syndrome 3	*POLR1C*	AR
Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism	*POLR3A*	AR
Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	*POLR3B*	AR
Obesity, adrenal insufficiency, and red hair due to POMC deficiency	*POMC*	AR

Disorders	Genes	Inheritance
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	*POMGNT1*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8	*POMGNT2*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12	*POMK*	AR
Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	*POMP*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	*POMT1*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	*POMT2*	AR
Anauxetic dysplasia 2	*POP1*	AR
Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	*POR*	AR
Pituitary hormone deficiency, combined or isolated, 1	*POU1F1*	AD/AR
Deafness, X-linked 2	*POU3F4*	XLR
Sudden cardiac failure, infantile	*PPA2*	AR
Pontocerebellar hypoplasia, type 14	*PPIL1*	AR
Ceroid lipofuscinosis, neuronal, 1	*PPT1*	AR
Renpenning syndrome	*PQBP1*	XLR
Neuropathy, hereditary sensory and autonomic, type VIII	*PRDM12*	AR
Brittle cornea syndrome 2	*PRDM5*	AR
Spinocerebellar ataxia, autosomal recessive 32	*PRDX3*	AR
Hemophagocytic lymphohistiocytosis, familial, 2	*PRF1*	AR
Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	*PRG4*	AR
Epilepsy, progressive myoclonic 1B	*PRICKLE1*	AR
Autoimmune lymphoproliferative syndrome, type III	*PRKCD*	AR
Immunodeficiency 26, with or without neurologic abnormalities	*PRKDC*	AR
Dystonia 16	*PRKRA*	AR
Thrombophilia 3 due to protein C deficiency, autosomal recessive	*PROC*	AR
Hyperprolinemia, type I	*PRODH*	AR
Pituitary hormone deficiency, combined, 2	*PROP1*	AR
Thrombophilia 5 due to protein S deficiency, autosomal recessive	*PROS1*	AR
Arts syndrome	*PRPS1*	XLR
Microphthalmia, isolated 6	*PRSS56*	AR
Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies	*PRUNE1*	AR
Dejerine-Sottas disease	*PRX*	AD/ AR
Metachromatic leukodystrophy due to SAP-b deficiency	*PSAP*	AR
Neu-Laxova syndrome 2	*PSAT1*	AR
Proteasome-associated autoinflammatory syndrome 1 and digenic forms	*PSMB8*	AR
Phosphoserine phosphatase deficiency	*PSPH*	AR
Chondrodysplasia, Blomstrand type	*PTH1R*	AR
Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	*PTPN23*	AR
Immunodeficiency 105, severe combined	*PTPRC*	AR
Deafness, autosomal recessive 84A	*PTPRQ*	AR

Disorders	Genes	Inheritance
Infantile-onset multisystem neurologic, endocrine, and pancreatic disease	*PTRH2*	AR
Hyperphenylalaninemia, BH4-deficient, A	*PTS*	AR
Myopathy, lactic acidosis, and sideroblastic anemia 1	*PUS1*	AR
Neurodevelopmental disorder with microcephaly and gray sclerae	*PUS3*	AR
Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature	*PUS7*	AR
Anterior segment dysgenesis 7, with sclerocornea	*PXDN*	AR
Cutis laxa, autosomal recessive, type IIB	*PYCR1*	AR
Leukodystrophy, hypomyelinating, 10	*PYCR2*	AR
Glycogen storage disease VI	*PYGL*	AR
McArdle disease	*PYGM*	AR
Myopathy, myofibrillar, 8	*PYROXD1*	AR
Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	*QARS1*	AR
Hyperphenylalaninemia, BH4-deficient, C	*QDPR*	AR
Combined oxidative phosphorylation deficiency 40	*QRSL1*	AR
Warburg micro syndrome 3	*RAB18*	AR
Carpenter syndrome	*RAB23*	AR
Griscelli syndrome, type 2	*RAB27A*	AR
Cone-rod dystrophy 18	*RAB28*	AR
Smith-McCort dysplasia 2	*RAB33B*	AR
Intellectual developmental disorder, X-linked 72	*RAB39B*	XLR
Warburg micro syndrome 1	*RAB3GAP1*	AR
Warburg micro syndrome 2	*RAB3GAP2*	AR
Nijmegen breakage syndrome-like disorder	*RAD50*	AR
Severe combined immunodeficiency, B cell-negative	*RAG1*	AR
Severe combined immunodeficiency, B cell-negative	*RAG2*	AR
Fetal akinesia deformation sequence 2	*RAPSN*	AR
Microphthalmia, syndromic 12	*RARB*	AD/AR
Leukodystrophy, hypomyelinating, 9	*RARS1*	AR
Pontocerebellar hypoplasia, type 6	*RARS2*	AR
Immunodeficiency 64	*RASGRP1*	AR
Bleeding disorder, platelet-type, 18	*RASGRP2*	AR
Microphthalmia, syndromic 16	*RAX*	AR
Retinitis pigmentosa 95	*RAX2*	AR
Seckel syndrome 2	*RBBP8*	AR
Polyglucosan body myopathy 1 with or without immunodeficiency	*RBCK1*	AR
TARP syndrome	*RBM10*	XLR
Retinal dystrophy with or without extraocular anomalies	*RCBTB1*	AR
Leber congenital amaurosis 12	*RD3*	AR
Leber congenital amaurosis 13	*RDH12*	AD/ AR
Fundus albipunctatus	*RDH5*	AD/ AR
Deafness, autosomal recessive 24	*RDX*	AR
Baller-Gerold syndrome	*RECQL4*	AR
Retinitis pigmentosa 77	*REEP6*	AR
Lissencephaly 2 (Norman-Roberts type)	*RELN*	AR
Renal tubular dysgenesis	*REN*	AR
Neuropathy, hereditary sensory and autonomic, type IIB	*RETREG1*	AR

Disorders	Genes	Inheritance
Congenital disorder of glycosylation, type In	*RFT1*	AR
Mitchell-Riley syndrome	*RFX6*	AR
Bare lymphocyte syndrome, type II, complementation group B	*RFXANK*	AR
Bare lymphocyte syndrome, type II, complementation group D	*RFXAP*	AR
Macrocephaly, alopecia, cutis laxa, and scoliosis	*RIN2*	AR
Immunodeficiency 57 with autoinflammation	*RIPK1*	AR
Popliteal pterygium syndrome, Bartsocas-Papas type 1	*RIPK4*	AR
Bothnia retinal dystrophy	*RLBP1*	AR
Tonne-Kalscheuer syndrome	*RLIM*	XL
Combined oxidative phosphorylation deficiency 11	*RMND1*	AR
Cartilage-hair hypoplasia	*RMRP*	AR
Aicardi-Goutieres syndrome 4	*RNASEH2A*	AR
Aicardi-Goutieres syndrome 2	*RNASEH2B*	AR
Aicardi-Goutieres syndrome 3	*RNASEH2C*	AR
Leukoencephalopathy, cystic, without megalencephaly	*RNASET2*	AR
RIDDLE syndrome	*RNF168*	AR
Cerebellar ataxia and hypogonadotropic hypogonadism	*RNF216*	AR
Microcephalic osteodysplastic primordial dwarfism, type I	*RNU4ATAC*	AR
Gaze palsy, familial horizontal, with progressive scoliosis, 1	*ROBO3*	AR
Kohlschutter-Tonz syndrome	*ROGDI*	AR
Retinitis pigmentosa 7, digenic form	*ROM1*	AD/ AR/ DD
Robinow syndrome, autosomal recessive	*ROR2*	AR
Immunodeficiency 42	*RORC*	AR
Retinitis pigmentosa 1	*RP1*	AD/ AR
Retinitis pigmentosa 2	*RP2*	XL
Leber congenital amaurosis 2	*RPE65*	AR
Macular degeneration, X-linked atrophic	*RPGR*	XLR
Cone-rod dystrophy 13	*RPGRIP1*	AR
Meckel syndrome 5	*RPGRIP1L*	AR
Intellectual developmental disorder, X-linked syndromic 35	*RPL10*	XLR
Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	*RRM2B*	AR
Retinoschisis	*RS1*	XLR
Ciliary dyskinesia, primary, 24	*RSPH1*	AR
Ciliary dyskinesia, primary, 32	*RSPH3*	AR
Ciliary dyskinesia, primary, 11	*RSPH4A*	AR
Ciliary dyskinesia, primary, 12	*RSPH9*	AR
Intellectual developmental disorder, autosomal recessive 70	*RSRC1*	AR
Dyskeratosis congenita, autosomal recessive 5	*RTEL1*	AD/ AR
Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures	*RTN4IP1*	AR
Microcephaly, short stature, and polymicrogyria with seizures	*RTTN*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10	*RXYLT1*	AR
Congenital myopathy 1B, autosomal recessive	*RYR1*	AR

Disorders	Genes	Inheritance
Deafness, autosomal recessive 68	*S1PR2*	AR
Spastic ataxia, Charlevoix-Saguenay type	*SACS*	AR
Tumoral calcinosis, familial, normophosphatemic	*SAMD9*	AR
Aicardi-Goutieres syndrome 5	*SAMHD1*	AR
Chylomicron retention disease	*SAR1B*	AR
Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	*SARS2*	AR
Shwachman-Diamond syndrome 1	*SBDS*	AR
Charcot-Marie-Tooth disease, type 4B3	*SBF1*	AR
Charcot-Marie-Tooth disease, type 4B2	*SBF2*	AR
Lathosterolosis	*SC5D*	AR
Epilepsy, progressive myoclonic 4, with or without renal failure	*SCARB2*	AR
Van den Ende-Gupta syndrome	*SCARF2*	AR
Developmental and epileptic encephalopathy 52	*SCN1B*	AR
Myasthenic syndrome, congenital, 16	*SCN4A*	AR
Insensitivity to pain, congenital	*SCN9A*	AR
Pseudohypoaldosteronism, type IB1, autosomal recessive	*SCNN1A*	AR
Pseudohypoaldosteronism, type IB2, autosomal recessive	*SCNN1B*	AR
Mitochondrial complex IV deficiency, nuclear type 4	*SCO1*	AR
Mitochondrial complex IV deficiency, nuclear type 2	*SCO2*	AR
Spinocerebellar ataxia, autosomal recessive 21	*SCYL1*	AR
Bardet-Biedl syndrome 16	*SDCCAG8*	AR
Mitochondrial complex II deficiency, nuclear type 1	*SDHA*	AR
Mitochondrial complex II deficiency, nuclear type 2	*SDHAF1*	AR
Mitochondrial complex II deficiency, nuclear type 4	*SDHB*	AR
Ichthyosis, congenital, autosomal recessive 13	*SDR9C7*	AR
Craniolenticulosutural dysplasia	*SEC23A*	AD/ AR
Dyserythropoietic anemia, congenital, type II	*SEC23B*	AR
Cole-Carpenter syndrome 2	*SEC24D*	AR
Thyroid hormone metabolism, abnormal, 1	*SECISBP2*	AR
Congenital myopathy 3 with rigid spine	*SELENON*	AR
Cone-rod dystrophy 10	*SEMA4A*	AR
Pontocerebellar hypoplasia type 2D	*SEPSECS*	AR
3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	*SERAC1*	AR
Emphysema-cirrhosis, due to AAT deficiency	*SERPINA1*	AR
Thrombophilia 7 due to antithrombin III deficiency	*SERPINC1*	AD/ AR
Osteogenesis imperfecta, type VI	*SERPINF1*	AR
Alpha-2-plasmin inhibitor deficiency	*SERPINF2*	AR
Osteogenesis imperfecta, type X	*SERPINH1*	AR
Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2	*SETX*	AR
Pyle disease	*SFRP4*	AR
Surfactant metabolism dysfunction, pulmonary, 1	*SFTPB*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 3	*SGCA*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 4	*SGCB*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 6	*SGCD*	AR

Disorders	Genes	Inheritance
Muscular dystrophy, limb-girdle, autosomal recessive 5	*SGCG*	AR
Chronic atrial and intestinal dysrhythmia	*SGO1*	AR
RENI syndrome	*SGPL1*	AR
Mucopolysaccharidosis type IIIA (Sanfilippo A)	*SGSH*	AR
Lymphoproliferative syndrome, X-linked, 1	*SH2D1A*	XLR
Frank-ter Haar syndrome	*SH3PXD2B*	AR
Charcot-Marie-Tooth disease, type 4C	*SH3TC2*	AR
Langer mesomelic dysplasia	*SHOX*	PR
Marinesco-Sjogren syndrome	*SIL1*	AR
Trichohepatoenteric syndrome 2	*SKIC2*	AR
Trichohepatoenteric syndrome 1	*SKIC3*	AR
Bartter syndrome, type 1	*SLC12A1*	AR
Kilquist syndrome	*SLC12A2*	AR
Developmental and epileptic encephalopathy 34	*SLC12A5*	AR
Agenesis of the corpus callosum with peripheral neuropathy	*SLC12A6*	AR
Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta	*SLC13A5*	AR
Monocarboxylate transporter 1 deficiency	*SLC16A1*	AD/ AR
Allan-Herndon-Dudley syndrome	*SLC16A2*	XL
Sialic acid storage disorder, infantile	*SLC17A5*	AR
Myasthenic syndrome, congenital, 21, presynaptic	*SLC18A3*	AR
Thiamine-responsive megaloblastic anemia syndrome	*SLC19A2*	AR
Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	*SLC19A3*	AR
Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	*SLC1A4*	AR
Carnitine deficiency, systemic primary	*SLC22A5*	AR
Albinism, oculocutaneous, type VI	*SLC24A5*	AR
Combined D-2- and L-2-hydroxyglutaric aciduria	*SLC25A1*	AR
Developmental and epileptic encephalopathy 39	*SLC25A12*	AR
Citrullinemia, type II, neonatal-onset	*SLC25A13*	AR
Hyperornithinemia-hyperammonemia-mocitrullinemia syndrome	*SLC25A15*	AR
Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)	*SLC25A19*	AR
Carnitine-acylcarnitine translocase deficiency	*SLC25A20*	AR
Developmental and epileptic encephalopathy 3	*SLC25A22*	AR
Anemia, sideroblastic, 2, pyridoxine-refractory	*SLC25A38*	AR
Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR	*SLC25A4*	AR
Neuropathy, hereditary motor and sensory, type VIB	*SLC25A46*	AR
Achondrogenesis Ib	*SLC26A2*	AR
Diarrhea 1, secretory chloride, congenital	*SLC26A3*	AR
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	*SLC26A4*	AR
Pendred syndrome	*SLC26A4*	AR
Ichthyosis prematurity syndrome	*SLC27A4*	AR
Histiocytosis-lymphadenopathy plus syndrome	*SLC29A3*	AR
Arterial tortuosity syndrome	*SLC2A10*	AR
Fanconi-Bickel syndrome	*SLC2A2*	AR
Hypermanganesemia with dystonia 1	*SLC30A10*	AR

Disorders	Genes	Inheritance
Huppke-Brendel syndrome	*SLC33A1*	AR
Pulmonary alveolar microlithiasis	*SLC34A2*	AR
Arthrogryposis, impaired intellectual development, and seizures	*SLC35A3*	AR
Congenital disorder of glycosylation, type IIc	*SLC35C1*	AR
Schneckenbecken dysplasia	*SLC35D1*	AR
Glycogen storage disease Ib	*SLC37A4*	AR
Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis	*SLC38A8*	AR
Ehlers-Danlos syndrome, spondylodysplastic type, 3	*SLC39A13*	AR
Hypermanganesemia with dystonia 2	*SLC39A14*	AR
Acrodermatitis enteropathica	*SLC39A4*	AR
Agammaglobulinemia 9, autosomal recessive	*SLC39A7*	AR
Congenital disorder of glycosylation, type IIn	*SLC39A8*	AR
Cystinuria	*SLC3A1*	AD/ AR
Albinism, oculocutaneous, type IV	*SLC45A2*	AR
Folate malabsorption, hereditary	*SLC46A1*	AR
Distal renal tubular acidosis 4 with hemolytic anemia	*SLC4A1*	AR
Renal tubular acidosis, proximal, with ocular abnormalities	*SLC4A4*	AR
Brown-Vialetto-Van Laere syndrome 2	*SLC52A2*	AR
Brown-Vialetto-Van Laere syndrome 1	*SLC52A3*	AR
Glucose/galactose malabsorption	*SLC5A1*	AR
Sodium-dependent multivitamin transporter deficiency	*SLC5A6*	AR
Myasthenic syndrome, congenital, 20, presynaptic	*SLC5A7*	AR
Hartnup disorder	*SLC6A19*	AR
Parkinsonism-dystonia, infantile, 1	*SLC6A3*	AR
Hyperekplexia 3	*SLC6A5*	AD/ AR
Cerebral creatine deficiency syndrome 1	*SLC6A8*	XLR
Glycine encephalopathy with normal serum glycine	*SLC6A9*	AR
Lysinuric protein intolerance	*SLC7A7*	AR
Diarrhea 8, secretory sodium, congenital	*SLC9A3*	AR
Intellectual developmental disorder, X-linked syndromic, Christianson type	*SLC9A6*	XL
Hypertrophic osteoarthropathy, primary, autosomal recessive 2	*SLCO2A1*	AR
Deafness and myopia	*SLITRK6*	AR
Fanconi anemia, complementation group P	*SLX4*	AR
Schimke immunoosseous dysplasia	*SMARCAL1*	AR
Specific granule deficiency 2	*SMARCD2*	AR
Spinal muscular atrophy-1	*SMN1*	AR
Pallister-Hall-like syndrome	*SMO*	AR
Microphthalmia with limb anomalies	*SMOC1*	AR
Niemann-Pick disease, type A	*SMPD1*	AR
Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type	*SMS*	XLR
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	*SNAP29*	AR
Leukoencephalopathy, brain calcifications, and cysts	*SNORD118*	AR
Osteopetrosis, autosomal recessive 8	*SNX10*	AR
Spinocerebellar ataxia, autosomal recessive 20	*SNX14*	AR
Neuronopathy, distal hereditary motor, autosomal recessive 8	*SORD*	AR

Disorders	Genes	Inheritance
Sclerosteosis 1	*SOST*	AR
Hepatic venoocclusive disease with immunodeficiency	*SP110*	AR
Ciliary dyskinesia, primary, 28	*SPAG1*	AR
Troyer syndrome	*SPART*	AR
Leber congenital amaurosis 3	*SPATA7*	AR
Centronuclear myopathy 5	*SPEG*	AR
Spastic paraplegia 11, autosomal recessive	*SPG11*	AR
Netherton syndrome	*SPINK5*	AR
Diarrhea 3, secretory sodium, congenital, syndromic	*SPINT2*	AR
Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	*SPR*	AD/ AR
Neurodevelopmental disorder with hypotonia, neuropathy, and deafness	*SPTBN4*	AR
Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	*SQSTM1*	AR
Congenital disorder of glycosylation, type Iq	*SRD5A3*	AR
Congenital disorder of glycosylation, type Iy	*SSR4*	XLR
Ichthyosis, congenital, autosomal recessive 11	*ST14*	AR
Salt and pepper developmental regression syndrome	*ST3GAL5*	AR
Congenital myopathy 13	*STAC3*	AR
Mullegama-Klein-Martinez syndrome	*STAG2*	XL
Microcephaly-capillary malformation syndrome	*STAMBP*	AR
Lipoid adrenal hyperplasia	*STAR*	AR
Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive	*STAT1*	AR
Immunodeficiency 44	*STAT2*	AR
Growth hormone insensitivity with immune dysregulation 1, autosomal recessive	*STAT5B*	AR
Microcephaly 7, primary, autosomal recessive	*STIL*	AR
Immunodeficiency 10	*STIM1*	AR
Microphthalmia, isolated, with coloboma 8	*STRA6*	AR
Polyhydramnios, megalencephaly, and symptomatic epilepsy	*STRADA*	AR
Deafness, autosomal recessive 16	*STRC*	AR
Ichthyosis, X-linked	*STS*	XLR
Spinocerebellar ataxia, autosomal recessive 16	*STUB1*	AR
Hemophagocytic lymphohistiocytosis, familial, 4	*STX11*	AR
Developmental and epileptic encephalopathy 4	*STXBP1*	AD/ AR
Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease	*STXBP2*	AR
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	*SUCLA2*	AR
Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	*SUCLG1*	AR
Multiple sulfatase deficiency	*SUMF1*	AR
Sulfite oxidase deficiency	*SUOX*	AR
Mitochondrial complex IV deficiency, nuclear type 1	*SURF1*	AR
Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders	*SYN1*	XL
Spinocerebellar ataxia, autosomal recessive 8	*SYNE1*	AR
Developmental and epileptic encephalopathy 53	*SYNJ1*	AR
Intellectual developmental disorder, X-linked 96	*SYP*	XLR
Developmental and epileptic encephalopathy 18	*SZT2*	AR

Disorders	Genes	Inheritance
Dystonia-Parkinsonism, X-linked	*TAF1*	XLR
Intellectual developmental disorder, X-linked syndromic 33	*TAF1*	XLR
Barth syndrome	*TAFAZZIN*	XLR
Transaldolase deficiency	*TALDO1*	AR
Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	*TANGO2*	AR
Bare lymphocyte syndrome, type I	*TAP1*	AR
Tyrosinemia, type II	*TAT*	AR
Pontocerebellar hypoplasia, type 11	*TBC1D23*	AR
Developmental and epileptic encephalopathy 16	*TBC1D24*	AR
Nephrotic syndrome, type 20	*TBC1D8B*	XL
Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	*TBCD*	AR
Kenny-Caffey syndrome, type 1	*TBCE*	AR
Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	*TBCK*	AR
Adrenocorticotropic hormone deficiency	*TBX19*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 7	*TCAP*	AR
Osteopetrosis, autosomal recessive 1	*TCIRG1*	AR
Transcobalamin II deficiency	*TCN2*	AR
Joubert syndrome 13	*TCTN1*	AR
Joubert syndrome 24	*TCTN2*	AR
Joubert syndrome 18	*TCTN3*	AR
Spinocerebellar ataxia, autosomal recessive 23	*TDP2*	AR
Cataract 36	*TDRD7*	AR
Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay	*TECPR2*	AR
Ventricular tachycardia, catecholaminergic polymorphic, 3	*TECRL*	AR
Deafness, autosomal recessive 21	*TECTA*	AR
You-Hoover-Fong syndrome	*TELO2*	AR
Microphthalmia, isolated, with coloboma 9	*TENM3*	AR
Microphthalmia, syndromic 15	*TENM3*	AR
Beck-Fahrner syndrome	*TET3*	AD/AR
Atransferrinemia	*TF*	AR
Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies	*TFE3*	XL
Hemochromatosis, type 3	*TFR2*	AR
Ichthyosis, congenital, autosomal recessive 1	*TGM1*	AR
Segawa syndrome, recessive	*TH*	AR
Intellectual developmental disorder, X-linked 12	*THOC2*	XLR
Beaulieu-Boycott-Innes syndrome	*THOC6*	AR
Neurodevelopmental disorder with speech delay and variable ocular anomalies	*THUMPD1*	AR
3-methylglutaconic aciduria, type IX	*TIMM50*	AR
Mohr-Tranebjaerg syndrome	*TIMM8A*	XLR
Cholestasis, progressive familial intrahepatic 4	*TJP2*	AR
Mitochondrial DNA depletion syndrome 2 (myopathic type)	*TK2*	AR
Immunodeficiency 74, COVID19-related, X-linked	*TLR7*	XLR
Deafness, autosomal recessive 7	*TMC1*	AR

Disorders	Genes	Inheritance
Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1	*TMCO1*	AR
Orofaciodigital syndrome XVI	*TMEM107*	AR
Optic atrophy 7	*TMEM126A*	AR
Mitochondrial complex I deficiency, nuclear type 29	*TMEM126B*	AR
Joubert syndrome 16	*TMEM138*	AR
Congenital disorder of glycosylation, type IIk	*TMEM165*	AR
Joubert syndrome 2	*TMEM216*	AR
Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities	*TMEM222*	AR
Joubert syndrome 20	*TMEM231*	AR
Joubert syndrome 14	*TMEM237*	AR
Structural heart defects and renal anomalies syndrome	*TMEM260*	AR
Osteogenesis imperfecta, type XIV	*TMEM38B*	AR
Joubert syndrome 6	*TMEM67*	AR
Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	*TMEM70*	AR
Deafness, autosomal recessive 6	*TMIE*	AR
Enterokinase deficiency	*TMPRSS15*	AR
Deafness, autosomal recessive 8/10	*TMPRSS3*	AR
Lissencephaly 8	*TMTC3*	AR
Osteopetrosis, autosomal recessive 7	*TNFRSF11A*	AR
Paget disease of bone 5, juvenile-onset	*TNFRSF11B*	AR
Immunodeficiency, common variable, 2	*TNFRSF13B*	AD/ AR
Osteopetrosis, autosomal recessive 2	*TNFSF11*	AR
Nemaline myopathy 5A, autosomal recessive, severe infantile	*TNNT1*	AR
Ehlers-Danlos syndrome, classic-like, 1	*TNXB*	AR
Pontocerebellar hypoplasia, type 7	*TOE1*	AR
Joubert syndrome 37	*TOGARAM1*	AR
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5	*TOP3A*	AR
Microcephaly, growth restriction, and increased sister chromatid exchange 2	*TOP3A*	AR
Arthrogryposis multiplex congenita 5	*TOR1A*	AR
Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures	*TOR1AIP1*	AR
Galloway-Mowat syndrome 4	*TP53RK*	AR
Hemolytic anemia due to triosephosphate isomerase deficiency	*TPI1*	AR
Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	*TPK1*	AR
Congenital myopathy 4B, autosomal recessive	*TPM3*	AR
Ceroid lipofuscinosis, neuronal, 2	*TPP1*	AR
Immunodeficiency 78 with autoimmunity and developmental delay	*TPP2*	AR
Deafness, autosomal recessive 79	*TPRN*	AR
Immunodeficiency 7, TCR-alpha/beta deficient	*TRAC*	AR
Senior-Loken syndrome 9	*TRAF3IP1*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 18	*TRAPPC11*	AR
Spondyloepiphyseal dysplasia tarda	*TRAPPC2*	XLR

Disorders	Genes	Inheritance
Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy	*TRAPPC6B*	AR
Intellectual developmental disorder, autosomal recessive 13	*TRAPPC9*	AR
Cardiac arrhythmia syndrome, with or without skeletal muscle weakness	*TRDN*	AR
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2	*TREM2*	AR
Aicardi-Goutieres syndrome 1, dominant and recessive	*TREX1*	AD/ AR
Muscular dystrophy, limb-girdle, autosomal recessive 8	*TRIM32*	AR
Mulibrey nanism	*TRIM37*	AR
Deafness, autosomal recessive 28	*TRIOBP*	AR
Achondrogenesis, type IA	*TRIP11*	AR
Combined oxidative phosphorylation deficiency 35	*TRIT1*	AR
Microcephaly, short stature, and impaired glucose metabolism 1	*TRMT10A*	AR
Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay	*TRMT5*	AR
Liver failure, transient infantile	*TRMU*	AR
Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay	*TRNT1*	AR
Hypomagnesemia 1, intestinal	*TRPM6*	AR
Pontocerebellar hypoplasia type 2B	*TSEN2*	AR
Pontocerebellar hypoplasia type 2A	*TSEN54*	AR
Combined oxidative phosphorylation deficiency 3	*TSFM*	AR
Hypothyroidism, congenital, nongoitrous 4	*TSHB*	AR
Intellectual developmental disorder, X-linked 58	*TSPAN7*	XLR
Sudden infant death with dysgenesis of the testes syndrome	*TSPYL1*	AR
Mitochondrial complex III deficiency, nuclear type 2	*TTC19*	AR
Short-rib thoracic dysplasia 4 with or without polydactyly	*TTC21B*	AR
Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism	*TTC5*	AR
Gastrointestinal defects and immunodeficiency syndrome	*TTC7A*	AR
Bardet-Biedl syndrome 8	*TTC8*	AR
Intellectual developmental disorder, autosomal recessive 39	*TTI2*	AR
Congenital myopathy 5 with cardiomyopathy	*TTN*	AR
Ataxia with isolated vitamin E deficiency	*TTPA*	AR
Microcephaly and chorioretinopathy, autosomal recessive, 3	*TUBGCP4*	AR
Microcephaly and chorioretinopathy, autosomal recessive, 1	*TUBGCP6*	AR
Combined oxidative phosphorylation deficiency 4	*TUFM*	AR
Retinitis pigmentosa 14	*TULP1*	AR
Hepatorenocardiac degenerative fibrosis	*TULP3*	AR
Intellectual developmental disorder, autosomal recessive 7	*TUSC3*	AR
Focal facial dermal dysplasia 3, Setleis type	*TWIST2*	AR
Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	*TWNK*	AR
Burn-McKeown syndrome	*TXNL4A*	AR
Immunodeficiency 35	*TYK2*	AR

Disorders	Genes	Inheritance
Mitochondrial DNA depletion syndrome 1 (MNGIE type)	*TYMP*	AR
Albinism, oculocutaneous, type IA	*TYR*	AR
Albinism, oculocutaneous, type III	*TYRP1*	AR
Spinal muscular atrophy, X-linked 2, infantile	*UBA1*	XLR
Developmental and epileptic encephalopathy 44	*UBA5*	AR
Intellectual developmental disorder, X-linked syndromic, Nascimento type	*UBE2A*	XLR
Fanconi anemia, complementation group T	*UBE2T*	AR
Kaufman oculocerebrofacial syndrome	*UBE3B*	AR
Johanson-Blizzard syndrome	*UBR1*	AR
Li-Campeau syndrome	*UBR7*	AR
Leukodystrophy, hypomyelinating, 14	*UFM1*	AR
Crigler-Najjar syndrome, type I	*UGT1A1*	AR
Orotic aciduria	*UMPS*	AR
Hemophagocytic lymphohistiocytosis, familial, 3	*UNC13D*	AR
Osteootohepatoenteric syndrome	*UNC45A*	AR
Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	*UNC80*	AR
Immunodeficiency with hyper IgM, type 5	*UNG*	AR
Beta-ureidopropionase deficiency	*UPB1*	AR
Intellectual developmental disorder, X-linked syndromic 14	*UPF3B*	XLR
Mitochondrial complex III deficiency, nuclear type 5	*UQCRC2*	AR
Mitochondrial complex III deficiency, nuclear type 4	*UQCRQ*	AR
Porphyria, congenital erythropoietic	*UROS*	AR
Poikiloderma with neutropenia	*USB1*	AR
Usher syndrome, type 1C	*USH1C*	AR
Usher syndrome, type 1G	*USH1G*	AR
Usher syndrome, type 2A	*USH2A*	AR
Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss	*USP53*	AR
Intellectual developmental disorder, X-linked 99	*USP9X*	XLR
Myasthenic syndrome, congenital, 25	*VAMP1*	AR
Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy	*VARS1*	AR
Combined oxidative phosphorylation deficiency 20	*VARS2*	AR
Arthrogryposis, renal dysfunction, and cholestasis 2	*VIPAS39*	AR
Vitamin K-dependent clotting factors, combined deficiency of, 2	*VKORC1*	AR
Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1	*VLDLR*	AR
Myopathy, X-linked, with excessive autophagy	*VMA21*	XLR
Leukodystrophy, hypomyelinating, 12	*VPS11*	AR
Choreoacanthocytosis	*VPS13A*	AR
Cohen syndrome	*VPS13B*	AR
Spinocerebellar ataxia, autosomal recessive 4	*VPS13D*	AR
Arthrogryposis, renal dysfunction, and cholestasis 1	*VPS33B*	AR
Spastic paraplegia 53, autosomal recessive	*VPS37A*	AR
Neutropenia, severe congenital, 5, autosomal recessive	*VPS45*	AR
Pontocerebellar hypoplasia, type 2E	*VPS53*	AR
Pontocerebellar hypoplasia type 1A	*VRK1*	AR

Disorders	Genes	Inheritance
Microphthalmia with coloboma 3	*VSX2*	AR
Neuronopathy, distal hereditary motor, autosomal recessive 7	*VWA1*	AR
von Willebrand disease, types 2A, 2B, 2M, and 2N	*VWF*	AD/ AR
Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures	*WARS2*	AR
Wiskott-Aldrich syndrome	*WAS*	XLR
Senior-Loken syndrome 8	*WDR19*	AR
Short-rib thoracic dysplasia 7 with or without polydactyly	*WDR35*	AR
Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures	*WDR45B*	AR
Microcephaly 2, primary, autosomal recessive, with or without cortical malformations	*WDR62*	AR
Galloway-Mowat syndrome 1	*WDR73*	AR
Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2	*WDR81*	AR
Wolfram syndrome 1	*WFS1*	AR
Usher syndrome, type 2D	*WHRN*	AR
Wiskott-Aldrich syndrome 2	*WIPF1*	AR
Neuropathy, hereditary sensory and autonomic, type II	*WNK1*	AR
Osteogenesis imperfecta, type XV	*WNT1*	AR
Split-hand/foot malformation 6	*WNT10B*	AR
Ulna and fibula, absence of, with severe limb deficiency	*WNT7A*	AR
Dyskeratosis congenita, autosomal recessive 3	*WRAP53*	AR
Werner syndrome	*WRN*	AR
Developmental and epileptic encephalopathy 28	*WWOX*	AR
Lymphoproliferative syndrome, X-linked, 2	*XIAP*	XLR
Xeroderma pigmentosum, group A	*XPA*	AR
Xeroderma pigmentosum, group C	*XPC*	AR
Nephronophthisis-like nephropathy 1	*XPNPEP3*	AR
Fanconi anemia, complementation group U	*XRCC2*	AR
Short stature, microcephaly, and endocrine dysfunction	*XRCC4*	AR
Desbuquois dysplasia 2	*XYLT1*	AR
Spondyloocular syndrome	*XYLT2*	AR
Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2	*YARS1*	AR
Myopathy, lactic acidosis, and sideroblastic anemia 2	*YARS2*	AR
Kaya-Barakat-Masson syndrome	*YIF1B*	AR
Grange syndrome	*YY1AP1*	AR
Immunodeficiency 48	*ZAP70*	AR
Immunodeficiency-centromeric instability-facial anomalies syndrome 2	*ZBTB24*	AR
Wieacker-Wolff syndrome	*ZC4H2*	XLR
Intellectual developmental disorder, X-linked syndromic, Raymond type	*ZDHHC9*	XL
Cholestasis, progressive familial intrahepatic, 9	*ZFYVE19*	AR
Spastic paraplegia 15, autosomal recessive	*ZFYVE26*	AR
Congenital heart defects, nonsyndromic, 1, X-linked	*ZIC3*	XLR
Restrictive dermopathy 1	*ZMPSTE24*	AR
Ciliary dyskinesia, primary, 22	*ZMYND10*	AR
Neurodevelopmental disorder with impaired speech and hyperkinetic movements	*ZNF142*	AR
Microcephaly 10, primary, autosomal recessive	*ZNF335*	AR
Brittle cornea syndrome 1	*ZNF469*	AR
DEGCAGS syndrome	*ZNF699*	AR
Intellectual developmental disorder, X-linked 97	*ZNF711*	XL
Immunodeficiency 91 and hyperinflammation	*ZNFX1*	AR
PEHO syndrome	*ZNHIT3*	AR

Note: AD: Autosomal dominant; AR: Autosomal recessive; XL: X-linked; XLD: X-linked dominant; XLR: X-linked recessive; DD: Digenic dominant; SMo: Somatic mosaicism
'Albinism, oculocutaneous, type II, modifier of' and 'Low density lipoprotein cholesterol level QTL 1' indicate mutations that contribute to susceptibility to multifactorial disorders or to susceptibility to infection.
'High molecular weight kininogen deficiency' and 'Lactate dehydrogenase-B deficiency' are nondiseases, mainly genetic variations that lead to apparently abnormal laboratory test values.