SecureCari™

Expanded Carrier Screening 535-Gene Panel

Screens for pathogenic/ likely pathogenic variants in 535 genes associated with 734 X-linked and autosomal recessive disorders

Disorders	Genes	Inheritance
Developmental and epileptic encephalopathy 29	*AARS1*	AR
Trichothiodystrophy 8, nonphotosensitive	*AARS1*	AR
Ichthyosis, congenital, autosomal recessive 4A	*ABCA12*	AR
Ichthyosis, congenital, autosomal recessive 4B (harlequin)	*ABCA12*	AR
Surfactant metabolism dysfunction, pulmonary, 3	*ABCA3*	AR
Cholestasis, benign recurrent intrahepatic, 2	*ABCB11*	AR
Cholestasis, progressive familial intrahepatic 2	*ABCB11*	AR
Cholestasis, progressive familial intrahepatic 3	*ABCB4*	AR
Arterial calcification, generalized, of infancy, 2	*ABCC6*	AR
Hyperinsulinemic hypoglycemia, familial, 1	*ABCC8*	AD/AR
Diabetes mellitus, permanent neonatal 3, with or without neurologic features	*ABCC8*	AD/AR
Adrenoleukodystrophy	*ABCD1*	XLR
Methylmalonic aciduria and homocystinuria, cblJ type	*ABCD4*	AR
Sitosterolemia 2	*ABCG5*	AR
Sitosterolemia 1	*ABCG8*	AR
Isobutyryl-CoA dehydrogenase deficiency	*ACAD8*	AR
Acyl-CoA dehydrogenase, medium chain, deficiency of	*ACADM*	AR
Acyl-CoA dehydrogenase, short-chain, deficiency of	*ACADS*	AR
2-methylbutyrylglycinuria	*ACADSB*	AR
VLCAD deficiency	*ACADVL*	AR
Alpha-methylacetoacetic aciduria	*ACAT1*	AR
Renal tubular dysgenesis	*ACE*	AR
Spondyloenchondrodysplasia with immune dysregulation	*ACP5*	AR
Combined malonic and methylmalonic aciduria	*ACSF3*	AR
Aminoacylase 1 deficiency	*ACY1*	AR
Adenosine deaminase deficiency, partial	*ADA*	AR/SMo
Severe combined immunodeficiency due to ADA deficiency	*ADA*	AR/SMo
Thrombotic thrombocytopenic purpura, hereditary	*ADAMTS13*	AR
Usher syndrome, type 2C	*ADGRV1*	AR/DD
Adenylosuccinase deficiency	*ADSL*	AR
Intellectual developmental disorder, X-linked 109	*AFF2*	XLR
Aspartylglucosaminuria	*AGA*	AR
Glycogen storage disease IIIa	*AGL*	AR
Glycogen storage disease IIIb	*AGL*	AR
Rhizomelic chondrodysplasia punctata, type 3	*AGPS*	AR
Renal tubular dysgenesis	*AGT*	AR
Hyperoxaluria, primary, type 1	*AGXT*	AR
Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	*AHCY*	AR
Joubert syndrome 3	*AHI1*	AR
Leber congenital amaurosis 4	*AIPL1*	AD/AR
Cone-rod dystrophy	*AIPL1*	AD/AR
Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia	*AIRE*	AD/AR
Anemia, congenital, nonspherocytic hemolytic, 3, adenylate kinase deficient	*AK1*	AR
Reticular dysgenesis	*AK2*	AR
Sjogren-Larsson syndrome	*ALDH3A2*	AR
Hyperprolinemia, type II	*ALDH4A1*	AR
Succinic semialdehyde dehydrogenase deficiency	*ALDH5A1*	AR
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 Ic	*ALG6*	AR
Alstrom syndrome	*ALMS1*	AR
Ichthyosis, congenital, autosomal recessive 2	*ALOX12B*	AR
Hypophosphatasia, childhood	*ALPL*	AR
Hypophosphatasia, infantile	*ALPL*	AR
Glycine encephalopathy 2	*AMT*	AR
Intellectual developmental disorder, autosomal recessive 37	*ANK3*	AR
Nephronophthisis 16	*ANKS6*	AR
Spinocerebellar ataxia, autosomal recessive 10	*ANO10*	AR
Hypobetalipoproteinemia	*APOB*	AR
Androgen insensitivity, partial, with or without breast cancer	*AR*	XLR
Androgen insensitivity	*AR*	XLR
Argininemia	*ARG1*	AR
Metachromatic leukodystrophy	*ARSA*	AR
Mucopolysaccharidosis type VI (Maroteaux-Lamy)	*ARSB*	AR
Chondrodysplasia punctata, X-linked recessive	*ARSL*	XLR
Partington syndrome	*ARX*	XLR
Proud syndrome	*ARX*	XL
Developmental and epileptic encephalopathy 1	*ARX*	XLR
Hydranencephaly with abnormal genitalia	*ARX*	XL
Lissencephaly, X-linked 2	*ARX*	XL
Intellectual developmental disorder, X-linked 29	*ARX*	XLR
Argininosuccinic aciduria	*ASL*	AR
Canavan disease	*ASPA*	AR
Microcephaly 5, primary, autosomal recessive	*ASPM*	AR
Citrullinemia	*ASS1*	AR
Achromatopsia 7	*ATF6*	AR
Ataxia-telangiectasia	*ATM*	AR
Wilson disease	*ATP7B*	AR
Cholestasis, progressive familial intrahepatic 1	*ATP8B1*	AR
Intellectual disability-hypotonic facies syndrome, X-linked	*ATRX*	XLR
3-methylglutaconic aciduria, type I	*AUH*	AR

Disorders	Genes	Inheritance
Diabetes insipidus, nephrogenic, 1	*AVPR2*	XLR
Nephrogenic syndrome of inappropriate antidiuresis	*AVPR2*	XLR
Ehlers-Danlos syndrome, spondylodysplastic type, 2	*B3GALT6*	AR
Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	*B3GALT6*	AR
Peters-plus syndrome	*B3GLCT*	AR
Combined low LDL and fibrinogen	*B4GALT1*	AR
Congenital disorder of glycosylation, type IId	*B4GALT1*	AR
Bardet-Biedl syndrome 1	*BBS1*	AR/DR
Bardet-Biedl syndrome 10	*BBS10*	AR
Bardet-Biedl syndrome 2	*BBS2*	AR
Retinitis pigmentosa 74	*BBS2*	AR
Bardet-Biedl syndrome 9	*BBS9*	AR
Butyrylcholinesterase deficiency	*BCHE*	AR
Maple syrup urine disease, type Ia	*BCKDHA*	AR
Maple syrup urine disease, type Ib	*BCKDHB*	AR
GRACILE syndrome	*BCS1L*	AR
Mitochondrial complex III deficiency, nuclear type 1	*BCS1L*	AR
Bloom syndrome	*BLM*	AR
Osteogenesis imperfecta, type XIII	*BMP1*	AR
Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	*BRAT1*	AR
Rigidity and multifocal seizure syndrome, lethal neonatal	*BRAT1*	AR
Biotinidase deficiency	*BTD*	AR
Isolated growth hormone deficiency, type III, with agammaglobulinemia	*BTK*	XLR
Agammaglobulinemia, X-linked 1	*BTK*	XLR
Orofaciodigital syndrome XIV	*C2CD3*	AR
C6 deficiency	*C6*	AR
Cone-rod dystrophy, X-linked, 3	*CACNA1F*	XLR
Muscular dystrophy, limb-girdle, autosomal recessive 1	*CAPN3*	AR
Homocystinuria, B6-responsive and nonresponsive types	*CBS*	AR
Thrombosis, hyperhomocysteinemic	*CBS*	AR
COACH syndrome 2	*CC2D2A*	AR
Joubert syndrome 9	*CC2D2A*	AR
Meckel syndrome 6	*CC2D2A*	AR
Retinitis pigmentosa 93	*CC2D2A*	AR
Hydrocephalus, congenital, 1	*CCDC88C*	AR
Immunodeficiency, X-linked, with hyper-IgM	*CD40LG*	XLR
Dyserythropoietic anemia, congenital, type Ia	*CDAN1*	AR
Usher syndrome, type 1D	*CDH23*	AR/DR
Deafness, autosomal recessive 12	*CDH23*	AR
Microcephaly 8, primary, autosomal recessive	*CEP135*	AR
Seckel syndrome 5	*CEP152*	AR
Microcephaly 9, primary, autosomal recessive	*CEP152*	AR
Joubert syndrome 5	*CEP290*	AR
Meckel syndrome 4	*CEP290*	AR
Leber congenital amaurosis 10	*CEP290*	.
Cystic fibrosis	*CFTR*	AR

Disorders	Genes	Inheritance
Myasthenic syndrome, congenital, 6, presynaptic	*CHAT*	AR
Myasthenic syndrome, congenital, 1B, fast-channel	*CHRNA1*	AD/AR
Multiple pterygium syndrome, lethal type	*CHRNA1*	AR
Myasthenic syndrome, congenital, 4B, fast-channel	*CHRNE*	AR
Myasthenic syndrome, congenital, 4A, slow-channel	*CHRNE*	AD/AR
Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	*CHRNE*	AR
Usher syndrome, type IJ	*CIB2*	AR
Deafness, autosomal recessive 48	*CIB2*	AR
Myotonia congenita, recessive	*CLCN1*	AR
Dent disease 1	*CLCN5*	XLR
Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis	*CLCN5*	XLR
Hypophosphatemic rickets	*CLCN5*	XLR
Nephrolithiasis, type I	*CLCN5*	XLR
Osteopetrosis, autosomal recessive 4	*CLCN7*	AR
Bartter syndrome, type 3	*CLCNKB*	AR
Ceroid lipofuscinosis, neuronal, 3	*CLN3*	AR
Ceroid lipofuscinosis, neuronal, 5	*CLN5*	AR
Ceroid lipofuscinosis, neuronal, 6A	*CLN6*	AR
Ceroid lipofuscinosis, neuronal, 6B (Kufs type)	*CLN6*	AR
Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant	*CLN8*	AR
Ceroid lipofuscinosis, neuronal, 8	*CLN8*	AR
3-methylglutaconic aciduria, type VIIB, autosomal recessive	*CLPB*	AR
Usher syndrome, type 3A	*CLRN1*	AR
Achromatopsia 2	*CNGA3*	AR
Achromatopsia 3	*CNGB3*	AR
Hypomyelinating neuropathy, congenital, 3	*CNTNAP1*	AR
Lethal congenital contracture syndrome 7	*CNTNAP1*	AR
Neurodegeneration with brain iron accumulation 6	*COASY*	AR
Pontocerebellar hypoplasia, type 12	*COASY*	AR
Congenital disorder of glycosylation, type IIi	*COG5*	AR
Epidermolysis bullosa, junctional 4, intermediate	*COL17A1*	AR
Ehlers-Danlos syndrome, cardiac valvular type	*COL1A2*	AR
Steel syndrome	*COL27A1*	AR
Alport syndrome 3B, autosomal recessive	*COL4A3*	.
Alport syndrome 2, autosomal recessive	*COL4A4*	AR
Ullrich congenital muscular dystrophy 1B	*COL6A2*	AD/AR
Epidermolysis bullosa dystrophica inversa	*COL7A1*	AR
Epidermolysis bullosa dystrophica, autosomal recessive	*COL7A1*	AR
Epidermolysis bullosa dystrophica, localisata variant	*COL7A1*	AR
Myasthenic syndrome, congenital, 5	*COLQ*	AR
Spastic ataxia 10, autosomal recessive	*COQ4*	AR
Coenzyme Q10 deficiency, primary, 7	*COQ4*	AR
Nephrotic syndrome, type 9	*COQ8B*	AR
Joubert syndrome 17	*CPLANE1*	AR
Orofaciodigital syndrome VI	*CPLANE1*	AR
Carbamoylphosphate synthetase I deficiency	*CPS1*	AR

Disorders	Genes	Inheritance
CPT deficiency, hepatic, type IA	*CPT1A*	AR
CPT II deficiency, lethal neonatal	*CPT2*	AR
CPT II deficiency, myopathic, stress-induced	*CPT2*	AD/AR
CPT II deficiency, infantile	*CPT2*	AR
Leber congenital amaurosis 8	*CRB1*	AR
Retinitis pigmentosa-12	*CRB1*	AR
Osteogenesis imperfecta, type XVI	*CREB3L1*	AR
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7	*CRPPA*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7	*CRPPA*	AR
Osteogenesis imperfecta, type VII	*CRTAP*	AR
Cataract 9, multiple types	*CRYAA*	AD/AR
Joubert syndrome 21	*CSPP1*	AR
Congenital cataracts, facial dysmorphism, and neuropathy	*CTDP1*	AR
Cystinosis, atypical nephropathic	*CTNS*	AR
Cystinosis, nephropathic	*CTNS*	AR
Cystinosis, late-onset juvenile or adolescent nephropathic	*CTNS*	AR
Cystinosis, ocular nonnephropathic	*CTNS*	AR
Galactosialidosis	*CTSA*	AR
Ceroid lipofuscinosis, neuronal, 10	*CTSD*	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
Chronic granulomatous disease 4, autosomal recessive	*CYBA*	AR
Immunodeficiency 34, mycobacteriosis, 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
17-alpha-hydroxylase/17,20-lyase deficiency	*CYP17A1*	AR
17,20-lyase deficiency, isolated	*CYP17A1*	AR
Anterior segment dysgenesis 6, multiple subtypes	*CYP1B1*	AR
Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset	*CYP1B1*	AR
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	*CYP21A2*	AR
Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	*CYP21A2*	AR
Hypercalcemia, infantile, 1	*CYP24A1*	AR
Cerebrotendinous xanthomatosis	*CYP27A1*	AR
Vitamin D-dependent rickets, type I	*CYP27B1*	AR
Maple syrup urine disease, type II	*DBT*	AR
Van Maldergem syndrome 1	*DCHS1*	AR
Xeroderma pigmentosum, group E, DDB-negative subtype	*DDB2*	AR
Smith-Lemli-Opitz syndrome	*DHCR7*	AR
Retinitis pigmentosa 59	*DHDDS*	AR
Dyskeratosis congenita, X-linked	*DKC1*	XLR
Dihydrolipoamide dehydrogenase deficiency	*DLD*	AR

Disorders	Genes	Inheritance
Becker muscular dystrophy	*DMD*	XLR
Duchenne muscular dystrophy	*DMD*	XLR
Hypophosphatemic rickets, AR	*DMP1*	AR
Ciliary dyskinesia, primary, 18	*DNAAF5*	AR
Ciliary dyskinesia, primary, 40	*DNAH9*	AR
3-methylglutaconic aciduria, type V	*DNAJC19*	AR
Immunodeficiency-centromeric instability-facial anomalies syndrome 1	*DNMT3B*	AR
Developmental and epileptic encephalopathy 23	*DOCK7*	AR
Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections	*DOCK8*	AR
Fetal akinesia deformation sequence 3	*DOK7*	AR
Myasthenic syndrome, congenital, 10	*DOK7*	AR
Congenital disorder of glycosylation, type Im	*DOLK*	AR
Myasthenic syndrome, congenital, 13, with tubular aggregates	*DPAGT1*	AR
Congenital disorder of glycosylation, type Ij	*DPAGT1*	AR
Congenital disorder of glycosylation, type Ie	*DPM1*	AR
5-fluorouracil toxicity	*DPYD*	AR
Dihydropyrimidine dehydrogenase deficiency	*DPYD*	AR
Neuropathy, hereditary sensory and autonomic, type VI	*DST*	AR
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*	AR/DR
Short-rib thoracic dysplasia 8 with or without polydactyly	*DYNC2I1*	AR
Myopathy, distal, with anterior tibial onset	*DYSF*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 2	*DYSF*	AR
Arthrogryposis, distal, type 5D	*ECEL1*	AR
Ectodermal dysplasia 1, hypohidrotic, X-linked	*EDA*	XLR
Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure	*EIF2B5*	AR
Dysautonomia, familial	*ELP1*	AR
Hypophosphatemic rickets, autosomal recessive, 2	*ENPP1*	AR
Arterial calcification, generalized, of infancy, 1	*ENPP1*	AR
Cerebrooculofacioskeletal syndrome 2	*ERCC2*	AR
Xeroderma pigmentosum, group D	*ERCC2*	AR
Trichothiodystrophy 1, photosensitive	*ERCC2*	AR
Cockayne syndrome, type A	*ERCC8*	AR
UV-sensitive syndrome 2	*ERCC8*	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
Retinitis pigmentosa 25	*EYS*	AR
Factor XI deficiency, autosomal recessive	*F11*	.
Hypoprothrombinemia	*F2*	AR
Dysprothrombinemia	*F2*	AR
Factor V deficiency	*F5*	AR
Factor VII deficiency	*F7*	AR

Disorders	Genes	Inheritance
Hemophilia A	*F8*	XLR
Hemophilia B	*F9*	XLR
Tyrosinemia, type I	*FAH*	AR
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 G	*FANCG*	AR
Fanconi anemia, complementation group I	*FANCI*	AR
Hennekam lymphangiectasia-lymphedema syndrome 2	*FAT4*	AR
Van Maldergem syndrome 2	*FAT4*	AR
Aarskog-Scott syndrome	*FGD1*	XLR
Intellectual developmental disorder, X-linked syndromic 16	*FGD1*	XLR
Tumoral calcinosis, hyperphosphatemic, familial, 2	*FGF23*	AR
Bruck syndrome 1	*FKBP10*	AR
Osteogenesis imperfecta, type XI	*FKBP10*	AR
Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5	*FKRP*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	*FKRP*	AR
Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4	*FKTN*	AR
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4	*FKTN*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	*FKTN*	AR
Cardiomyopathy, dilated, 1X	*FKTN*	AR
Trimethylaminuria	*FMO3*	AR
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	*FOXP3*	XLR
Cryptophthalmos, unilateral or bilateral, isolated	*FREM2*	AR
Fraser syndrome 2	*FREM2*	AR
Fucosidosis	*FUCA1*	AR
Glycogen storage disease Ia	*G6PC1*	AR
Dursun syndrome	*G6PC3*	AR
Neutropenia, severe congenital 4, autosomal recessive	*G6PC3*	AR
Glycogen storage disease II	*GAA*	AR
Krabbe disease	*GALC*	AR
Mucopolysaccharidosis IVA	*GALNS*	AR
Galactosemia	*GALT*	AR
Gaucher disease, type IIIC	*GBA1*	AR
Gaucher disease, type III	*GBA1*	AR
Gaucher disease, type II	*GBA1*	AR
Gaucher disease, type I	*GBA1*	AR
Gaucher disease, perinatal lethal	*GBA1*	AR
Polyglucosan body disease, adult form	*GBE1*	AR
Glycogen storage disease IV	*GBE1*	AR
Glutaricaciduria, type I	*GCDH*	AR
Hyperphenylalaninemia, BH4-deficient, B	*GCH1*	AR
Laron dwarfism	*GHR*	AR

Disorders	Genes	Inheritance
Deafness, autosomal recessive 1A	*GJB2*	AR/DD
Erythrokeratodermia variabilis et progressiva 1	*GJB3*	AD/AR
Fabry disease	*GLA*	XL
Fabry disease, cardiac variant	*GLA*	XL
GM1-gangliosidosis, type III	*GLB1*	AR
GM1-gangliosidosis, type II	*GLB1*	AR
GM1-gangliosidosis, type I	*GLB1*	AR
Mucopolysaccharidosis type IVB (Morquio)	*GLB1*	AR
Glycine encephalopathy1	*GLDC*	AR
Congenital arthrogryposis with anterior horn cell disease	*GLE1*	AR
Lethal congenital contracture syndrome 1	*GLE1*	AR
Achromatopsia 4	*GNAT2*	.
Lodder-Merla syndrome, type 1, with impaired intellectual development and cardiac arrhythmia	*GNB5*	AR
Lodder-Merla syndrome, type 2, with developmental delay and with or without cardiac arrhythmia	*GNB5*	AR
Nonaka myopathy	*GNE*	AR
Glycine N-methyltransferase deficiency	*GNMT*	AR
Mucolipidosis III alpha/beta	*GNPTAB*	AR
Mucolipidosis II alpha/beta	*GNPTAB*	AR
Mucopolysaccharidosis type IIID	*GNS*	AR
Geroderma osteodysplasticum	*GORAB*	AR
Anemia, congenital, nonspherocytic hemolytic, 4, glucose phosphate isomerase deficient	*GPI*	AR
Hyperoxaluria, primary, type II	*GRHPR*	AR
Intellectual developmental disorder, X-linked syndromic, Wu type	*GRIA3*	XLR
Fraser syndrome 3	*GRIP1*	AR
Leber congenital amaurosis 1	*GUCY2D*	AR
Cone-rod dystrophy 6	*GUCY2D*	AD/AR
Mucopolysaccharidosis VII	*GUSB*	AR
3-hydroxyacyl-CoA dehydrogenase deficiency	*HADH*	AR
Hyperinsulinemic hypoglycemia, familial, 4	*HADH*	AR
Mitochondrial trifunctional protein deficiency 1	*HADHA*	AR
LCHAD deficiency	*HADHA*	AR
Mitochondrial trifunctional protein deficiency 2	*HADHB*	AR
Neutropenia, severe congenital 3, autosomal recessive	*HAX1*	AR
Thalassemias, alpha-	*HBA1*	.
Thalassemia, alpha-	*HBA2*	.
Thalassemia, beta	*HBB*	.
Sickle cell disease	*HBB*	AR
Methylmalonic aciduria and homocysteinemia, cblX type	*HCFC1*	XLR
GM2-gangliosidosis, several forms	*HEXA*	AR
Tay-Sachs disease	*HEXA*	AR
Sandhoff disease, infantile, juvenile, and adult forms	*HEXB*	AR
Hemochromatosis, type 1	*HFE*	AR
Alkaptonuria	*HGD*	AR
Retinitis pigmentosa 73	*HGSNAT*	AR
Mucopolysaccharidosis type IIIC (Sanfilippo C)	*HGSNAT*	AR

Disorders	Genes	Inheritance
Holocarboxylase synthetase deficiency	*HLCS*	AR
HMG-CoA lyase deficiency	*HMGCL*	AR
HMG-CoA synthase-2 deficiency	*HMGCS2*	AR
Hyperoxaluria, primary, type III	*HOGA1*	AR
Tyrosinemia, type III	*HPD*	AR
Hermansky-Pudlak syndrome 1	*HPS1*	AR
Hermansky-Pudlak syndrome 3	*HPS3*	AR
D-bifunctional protein deficiency	*HSD17B4*	AR
Perrault syndrome 1	*HSD17B4*	AR
Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	*HSD3B2*	AR
Mucopolysaccharidosis II	*IDS*	XLR
Mucopolysaccharidosis Ih/s	*IDUA*	AR
Mucopolysaccharidosis Ih	*IDUA*	AR
Mucopolysaccharidosis Is	*IDUA*	AR
Short-rib thoracic dysplasia 9 with or without polydactyly	*IFT140*	AR
Retinitis pigmentosa 80	*IFT140*	AR
Neuronopathy, distal hereditary motor, autosomal recessive 1	*IGHMBP2*	AR
Charcot-Marie-Tooth disease, axonal, type 2S	*IGHMBP2*	AR
Inflammatory bowel disease 28, early onset, autosomal recessive	*IL10RA*	AR
Severe combined immunodeficiency, X-linked	*IL2RG*	XLR
Combined immunodeficiency, X-linked, moderate	*IL2RG*	XLR
Immunodeficiency 104, severe combined	*IL7R*	AR
Glanzmann thrombasthenia 1	*ITGA2B*	AR
Leukocyte adhesion deficiency	*ITGB2*	AR
Isovaleric acidemia	*IVD*	AR
Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	*JAM3*	AR
Hyperinsulinemic hypoglycemia, familial, 2	*KCNJ11*	AD/AR
Osteogenesis imperfecta, type XXI	*KDELR2*	AR
Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type	*KDM5C*	XLR
Nemaline myopathy 8, autosomal recessive	*KLHL40*	AR
Keratosis palmoplantaris striata III	*KRT1*	.
Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive	*KRT14*	AR
Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive	*KRT5*	AR
Vertebral, cardiac, renal, and limb defects syndrome 2	*KYNU*	AR
Hydroxykynureninuria	*KYNU*	AR
MASA syndrome	*L1CAM*	XLR
Hydrocephalus, congenital, X-linked	*L1CAM*	XLR
Corpus callosum, partial agenesis of	*L1CAM*	XLR
Poretti-Boltshauser syndrome	*LAMA1*	AR
Muscular dystrophy, congenital, merosin deficient or partially deficient	*LAMA2*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 23	*LAMA2*	AR

Disorders	Genes	Inheritance
Epidermolysis bullosa, junctional 2A, intermediate	*LAMA3*	AR
Epidermolysis bullosa, junctional 2B, severe	*LAMA3*	AR
Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous	*LAMA3*	AR
Lissencephaly 5	*LAMB1*	AR
Pierson syndrome	*LAMB2*	AR
Nephrotic syndrome, type 5, with or without ocular abnormalities	*LAMB2*	AR
Epidermolysis bullosa, junctional 1B, severe	*LAMB3*	AR
Epidermolysis bullosa, junctional 1A, intermediate	*LAMB3*	AR
Epidermolysis bullosa, junctional 3A, intermediate	*LAMC2*	AR
Epidermolysis bullosa, junctional 3B, severe	*LAMC2*	AR
Leber congenital amaurosis 5	*LCA5*	AR
Pituitary hormone deficiency, combined, 3	*LHX3*	AR
Wolman disease	*LIPA*	AR
Cholesteryl ester storage disease	*LIPA*	AR
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*	AR
Lipoprotein lipase deficiency	*LPL*	AR
Albinism, oculocutaneous, type VII	*LRMDA*	AR
Donnai-Barrow syndrome	*LRP2*	AR
Cenani-Lenz syndactyly syndrome	*LRP4*	AR
Osteoporosis-pseudoglioma syndrome	*LRP5*	AR
Exudative vitreoretinopathy 4	*LRP5*	AD/AR
Myopia 23, autosomal recessive	*LRPAP1*	AR
Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)	*LRPPRC*	AR
Chediak-Higashi syndrome	*LYST*	AR
Mannosidosis, alpha-, types I and II	*MAN2B1*	AR
Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency	*MAT1A*	AD/AR
Methionine adenosyltransferase deficiency, autosomal recessive	*MAT1A*	AD/AR
IFAP syndrome with or without BRESHECK syndrome	*MBTPS2*	XLR
3-Methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*	AR
3-Methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*	AR
Methylmalonyl-CoA epimerase deficiency	*MCEE*	AR
Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development	*MCM3AP*	AR
Mucolipidosis IV	*MCOLN1*	AR
Microcephaly 1, primary, autosomal recessive	*MCPH1*	AR
Familial Mediterranean fever, AR	*MEFV*	AR
Osteogenesis imperfecta, type XX	*MESD*	AR
Macular dystrophy with central cone involvement	*MFSD8*	AR
Ceroid lipofuscinosis, neuronal, 7	*MFSD8*	AR
Opitz GBBB syndrome	*MID1*	XLR
Combined oxidative phosphorylation deficiency 31	*MIPEP*	AR
Bardet-Biedl syndrome 13	*MKS1*	AR
Joubert syndrome 28	*MKS1*	AR
Meckel syndrome 1	*MKS1*	AR
Megalencephalic leukoencephalopathy with subcortical cysts 1	*MLC1*	AR
Malonyl-CoA decarboxylase deficiency	*MLYCD*	AR

Disorders	Genes	Inheritance
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, cblD type	*MMADHC*	AR
Methylmalonic aciduria and homocystinuria, cblD type	*MMADHC*	AR
Homocystinuria-megaloblastic anemia, cblD type	*MMADHC*	AR
Methylmalonic aciduria, mut(0) type	*MMUT*	AR
Molybdenum cofactor deficiency A	*MOCS1*	AR
Congenital disorder of glycosylation, type IIb	*MOGS*	AR
Congenital disorder of glycosylation, type Ib	*MPI*	AR
Amegakaryocytic thrombocytopenia, congenital, 1	*MPL*	AR
Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	*MPV17*	AR
Charcot-Marie-Tooth disease, axonal, type 2EE	*MPV17*	AR
Homocystinuria due to MTHFR deficiency	*MTHFR*	AR
Myopathy, centronuclear, X-linked	*MTM1*	XLR
Homocystinuria-megaloblastic anemia, cblG complementation type	*MTR*	AR
Homocystinuria-megaloblastic anemia, cbl E type	*MTRR*	AR
Abetalipoproteinemia	*MTTP*	AR
Mevalonic aciduria	*MVK*	AR
Hyper-IgD syndrome	*MVK*	AR
Deafness, autosomal recessive 3	*MYO15A*	AR
Usher syndrome, type 1B	*MYO7A*	AR
Kanzaki disease	*NAGA*	AR
Schindler disease, type III	*NAGA*	AR
Schindler disease, type I	*NAGA*	AR
Mucopolysaccharidosis type IIIB (Sanfilippo B)	*NAGLU*	AR
Nijmegen breakage syndrome	*NBN*	AR
Chronic granulomatous disease 1, autosomal recessive	*NCF1*	AR
Chronic granulomatous disease 2, autosomal recessive	*NCF2*	AR
Mitochondrial complex I deficiency, nuclear type 2	*NDUFS8*	AR
Mitochondrial complex I deficiency, nuclear type 4	*NDUFV1*	AR
Nemaline myopathy 2, autosomal recessive	*NEB*	AR
Arthrogryposis multiplex congenita 6	*NEB*	AR
Short-rib thoracic dysplasia 6 with or without polydactyly	*NEK1*	AR/DR
Niemann-Pick disease, type C1	*NPC1*	AR
Niemann-Pick disease, type D	*NPC1*	AR
Niemann-pick disease, type C2	*NPC2*	AR
Meckel syndrome 7	*NPHP3*	AR
Nephronophthisis 3	*NPHP3*	AR
Renal-hepatic-pancreatic dysplasia 1	*NPHP3*	AR
Nephrotic syndrome, type 1	*NPHS1*	AR
Nephrotic syndrome, type 2	*NPHS2*	AR
46XY sex reversal 2, dosage-sensitive	*NR0B1*	XL
Adrenal hypoplasia, congenital	*NR0B1*	XLR
Insensitivity to pain, congenital, with anhidrosis	*NTRK1*	AR

Disorders	Genes	Inheritance
Gyrate atrophy of choroid and retina with or without ornithinemia	*OAT*	AR
3-M syndrome 2	*OBSL1*	AR
Albinism, oculocutaneous, type II	*OCA2*	AR
Albinism, brown oculocutaneous	*OCA2*	AR
Dent disease 2	*OCRL*	XLR
Lowe syndrome	*OCRL*	XLR
3-methylglutaconic aciduria, type III	*OPA3*	AR
Intellectual developmental disorder, X-linked syndromic, Billuart type	*OPHN1*	XLR
Ornithine transcarbamylase deficiency	*OTC*	XL
Deafness, autosomal recessive 9	*OTOF*	AR
Auditory neuropathy, autosomal recessive, 1	*OTOF*	AR
Deafness, autosomal recessive 18B	*OTOG*	AR
Deafness, autosomal recessive 84B	*OTOGL*	AR
Osteogenesis imperfecta, type VIII	*P3H1*	AR
Phenylketonuria	*PAH*	AR
Pyruvate carboxylase deficiency	*PC*	AR
Hyperphenylalaninemia, BH4-deficient, D	*PCBD1*	AR
Propionicacidemia	*PCCA*	AR
Propionicacidemia	*PCCB*	AR
Usher syndrome, type 1F	*PCDH15*	AR
Cone dystrophy 4	*PDE6C*	AR
Retinal cone dystrophy 3	*PDE6H*	AD/AR
Deafness, autosomal recessive 57	*PDZD7*	AR
Heimler syndrome 1	*PEX1*	AR
Peroxisome biogenesis disorder 1A (Zellweger)	*PEX1*	AR
Peroxisome biogenesis disorder 1B (NALD/IRD)	*PEX1*	AR
Peroxisome biogenesis disorder 5A (Zellweger)	*PEX2*	AR
Peroxisome biogenesis disorder 5B	*PEX2*	AR
Peroxisome biogenesis disorder 4A (Zellweger)	*PEX6*	AR
Peroxisome biogenesis disorder 4B	*PEX6*	AD/AR
Peroxisome biogenesis disorder 9B	*PEX7*	AR
Rhizomelic chondrodysplasia punctata, type 1	*PEX7*	AR
Hypophosphatemic rickets, X-linked dominant	*PHEX*	XLD
Intellectual developmental disorder, X-linked syndromic, Siderius type	*PHF8*	XLR
Glycogen storage disease, type IXa1	*PHKA2*	XLR
Glycogen storage disease, type IXa2	*PHKA2*	XLR
Spastic paraplegia 84, autosomal recessive	*PI4KA*	AR
Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis	*PI4KA*	AR
Gastrointestinal defects and immunodeficiency syndrome 2	*PI4KA*	AR
Lymphatic malformation 6	*PIEZO1*	AR
Multiple congenital anomalies-hypotonia-seizures syndrome 1	*PIGN*	AR
Heterotaxy, visceral, 8, autosomal	*PKD1L1*	AR
Polycystic kidney disease 4, with or without hepatic disease	*PKHD1*	AR
Anemia, congenital, nonspherocytic hemolytic, 2, pyruvate kinase deficient	*PKLR*	AR

Disorders	Genes	Inheritance
Parkinson disease 14, autosomal recessive	*PLA2G6*	AR
Neurodegeneration with brain iron accumulation 2B	*PLA2G6*	AR
Infantile neuroaxonal dystrophy 1	*PLA2G6*	AR
Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	*PLAA*	AR
Nephrotic syndrome, type 3	*PLCE1*	AR
Ehlers-Danlos syndrome, kyphoscoliotic type, 1	*PLOD1*	AR
Bruck syndrome 2	*PLOD2*	AR
BCARD syndrome (lysyl hydroxylase 3 deficiency)	*PLOD3*	AR
Pelizaeus-Merzbacher disease	*PLP1*	XLR
Spastic paraplegia 2, X-linked	*PLP1*	XLR
Congenital disorder of glycosylation, type Ia	*PMM2*	AR
Charcot-Marie-Tooth disease, type 2B2	*PNKP*	AR
Ataxia-oculomotor apraxia 4	*PNKP*	AR
Microcephaly, seizures, and developmental delay	*PNKP*	AR
Pyridoxamine 5'-phosphate oxidase deficiency	*PNPO*	AR
Progressive external ophthalmoplegia, autosomal recessive 1	*POLG*	AR
Mitochondrial DNA depletion syndrome 4A (Alpers type)	*POLG*	AR
Mitochondrial DNA depletion syndrome 4B (MNGIE type)	*POLG*	AR
Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE)	*POLG*	AR
Treacher Collins syndrome 2	*POLR1D*	AD/AR
Wiedemann-Rautenstrauch syndrome	*POLR3A*	AR
Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism	*POLR3A*	AR
Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3	*POMGNT1*	AR
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3	*POMGNT1*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	*POMGNT1*	AR
Retinitis pigmentosa 76	*POMGNT1*	AR
Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1	*POMT1*	AR
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1	*POMT1*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	*POMT1*	AR
Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2	*POMT2*	AR
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	*POMT2*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	*POMT2*	AR
Pituitary hormone deficiency, combined or isolated, 1	*POU1F1*	AD/AR
Osteogenesis imperfecta, type IX	*PPIB*	AR
Ceroid lipofuscinosis, neuronal, 1	*PPT1*	AR
Renpenning syndrome	*PQBP1*	XLR
Hemophagocytic lymphohistiocytosis, familial, 2	*PRF1*	AR
Thrombophilia 3 due to protein C deficiency, autosomal recessive	*PROC*	AR
Hyperprolinemia, type I	*PRODH*	AR

Disorders	Genes	Inheritance
Pituitary hormone deficiency, combined, 2	*PROP1*	AR
Thrombophilia 5 due to protein S deficiency, autosomal recessive	*PROS1*	AR
Deafness, autosomal recessive 84A	*PTPRQ*	AR
Hyperphenylalaninemia, BH4-deficient, A	*PTS*	AR
Glycogen storage disease VI	*PYGL*	AR
McArdle disease	*PYGM*	AR
Hyperphenylalaninemia, BH4-deficient, C	*QDPR*	AR
Martsolf syndrome 2	*RAB3GAP1*	AR
Warburg micro syndrome 1	*RAB3GAP1*	AR
Martsolf syndrome 1	*RAB3GAP2*	AR
Warburg micro syndrome 2	*RAB3GAP2*	AR
Severe combined immunodeficiency, B cell-negative	*RAG1*	AR
Severe combined immunodeficiency, B cell-negative	*RAG2*	AR
Omenn syndrome	*RAG2*	AR
Combined cellular and humoral immune defects with granulomas	*RAG2*	AR
Fetal akinesia deformation sequence 2	*RAPSN*	AR
Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	*RAPSN*	AR
Pontocerebellar hypoplasia, type 6	*RARS2*	AR
Renal tubular dysgenesis	*REN*	AR
Retinitis pigmentosa 4, autosomal dominant or recessive	*RHO*	AD/AR
Retinitis punctata albescens	*RHO*	AD/AR
Anauxetic dysplasia 1	*RMRP*	AR
Metaphyseal dysplasia without hypotrichosis	*RMRP*	AR
Cartilage-hair hypoplasia	*RMRP*	AR
Aicardi-Goutieres syndrome 2	*RNASEH2B*	AR
Retinitis pigmentosa 1	*RP1*	AD/AR
Retinitis pigmentosa 3	*RPGR*	XL
Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness	*RPGR*	XL
Cone-rod dystrophy, X-linked, 1	*RPGR*	XLR
Macular degeneration, X-linked atrophic	*RPGR*	XLR
Leber congenital amaurosis 6	*RPGRIP1*	AR
Cone-rod dystrophy 13	*RPGRIP1*	AR
Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction	*RRM2B*	AR
Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	*RRM2B*	AR
Mitochondrial DNA depletion syndrome 8B (MNGIE type)	*RRM2B*	AR
Retinoschisis	*RS1*	XLR
Ciliary dyskinesia, primary, 24	*RSPH1*	AR
Congenital myopathy 1B, autosomal recessive	*RYR1*	AR
Spastic ataxia, Charlevoix-Saguenay type	*SACS*	AR
Shwachman-Diamond syndrome 1	*SBDS*	AR
Mitochondrial complex IV deficiency, nuclear type 2	*SCO2*	AR
Dyserythropoietic anemia, congenital, type II	*SEC23B*	AR
3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	*SERAC1*	AR

Disorders	Genes	Inheritance
Hemorrhagic diathesis due to antithrombin Pittsburgh	*SERPINA1*	AR
Emphysema due to AAT deficiency	*SERPINA1*	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
Muscular dystrophy, limb-girdle, autosomal recessive 3	*SGCA*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 4	*SGCB*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 5	*SGCG*	AR
Mucopolysaccharidosis type IIIA (Sanfilippo A)	*SGSH*	AR
Lymphoproliferative syndrome, X-linked, 1	*SH2D1A*	XLR
Gitelman syndrome	*SLC12A3*	AR
Agenesis of the corpus callosum with peripheral neuropathy	*SLC12A6*	AR
Salla disease	*SLC17A5*	AR
Sialic acid storage disorder, infantile	*SLC17A5*	AR
Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type)	*SLC19A3*	AR
Carnitine deficiency, systemic primary	*SLC22A5*	AR
Albinism, oculocutaneous, type VI	*SLC24A5*	AR
Citrullinemia, type II, neonatal-onset	*SLC25A13*	AR
Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	*SLC25A15*	AR
Carnitine-acylcarnitine translocase deficiency	*SLC25A20*	AR
De la Chapelle dysplasia	*SLC26A2*	AR
Epiphyseal dysplasia, multiple, 4	*SLC26A2*	AR
Atelosteogenesis, type II	*SLC26A2*	AR
Diastrophic dysplasia	*SLC26A2*	AR
Diastrophic dysplasia, broad bone-platyspondylic variant	*SLC26A2*	AR
Achondrogenesis Ib	*SLC26A2*	AR
Diarrhea 1, secretory chloride, congenital	*SLC26A3*	AR
Pendred syndrome	*SLC26A4*	AR
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	*SLC26A4*	AR
Fanconi-Bickel syndrome	*SLC2A2*	AR
Hypophosphatemic rickets with hypercalciuria	*SLC34A3*	AR
Congenital disorder of glycosylation, type IIf	*SLC35A1*	AR
Arthrogryposis, impaired intellectual development, and seizures	*SLC35A3*	AR
Congenital disorder of glycosylation, type IIc	*SLC35C1*	AR
Glycogen storage disease Ib	*SLC37A4*	AR
Glycogen storage disease Ic	*SLC37A4*	AR
Albinism, oculocutaneous, type IV	*SLC45A2*	AR
Distal renal tubular acidosis 4 with hemolytic anemia	*SLC4A1*	AR
Lysinuric protein intolerance	*SLC7A7*	AR
Schimke immunoosseous dysplasia	*SMARCAL1*	AR
Spinal muscular atrophy-1	*SMN1*	AR
Spinal muscular atrophy-2	*SMN1*	AR
Spinal muscular atrophy-3	*SMN1*	AR
Spinal muscular atrophy-4	*SMN1*	AR

Disorders	Genes	Inheritance
Niemann-Pick disease, type A	*SMPD1*	AR
Niemann-Pick disease, type B	*SMPD1*	AR
Amyotrophic lateral sclerosis 1	*SOD1*	AD/AR
Spastic tetraplegia and axial hypotonia, progressive	*SOD1*	AR
Osteogenesis imperfecta, type XII	*SP7*	AR
Osteogenesis imperfecta, type XVII	*SPARC*	AR
Leber congenital amaurosis 3	*SPATA7*	AR
Retinitis pigmentosa 94, variable age at onset, autosomal recessive	*SPATA7*	AR
Netherton syndrome	*SPINK5*	AR
Pseudovaginal perineoscrotal hypospadias	*SRD5A2*	AR
Mullegama-Klein-Martinez syndrome	*STAG2*	XL
Holoprosencephaly 13, X-linked	*STAG2*	XLD/XLR
Lipoid adrenal hyperplasia	*STAR*	AR
Deafness, autosomal recessive 16	*STRC*	AR
Ichthyosis, X-linked	*STS*	XLR
Hemophagocytic lymphohistiocytosis, familial, 4	*STX11*	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
Charcot-Marie-Tooth disease, type 4K	*SURF1*	AR
Mitochondrial complex IV deficiency, nuclear type 1	*SURF1*	AR
Arthrogryposis multiplex congenita 3, myogenic type	*SYNE1*	AR
Barth syndrome	*TAFAZZIN*	XLR
Tyrosinemia, type II	*TAT*	AR
Osteopetrosis, autosomal recessive 1	*TCIRG1*	AR
Transcobalamin II deficiency	*TCN2*	AR
Joubert syndrome 24	*TCTN2*	AR
Meckel syndrome 8	*TCTN2*	AR
Osteogenesis imperfecta, type XVIII	*TENT5A*	AR
Atransferrinemia	*TF*	AR
Ichthyosis, congenital, autosomal recessive 1	*TGM1*	AR
Segawa syndrome, recessive	*TH*	AR
Thrombophilia 12 due to thrombomodulin defect	*THBD*	AD
Intellectual developmental disorder, X-linked syndromic, Kumar type	*THOC2*	XL
Hypercholanemia, familial 1	*TJP2*	AR
Cholestasis, progressive familial intrahepatic 4	*TJP2*	AR
Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3	*TK2*	AR
Mitochondrial DNA depletion syndrome 2 (myopathic type)	*TK2*	AR
Joubert syndrome 2	*TMEM216*	AR
Meckel syndrome 2	*TMEM216*	AR
Retinitis pigmentosa 98	*TMEM216*	AR
Joubert syndrome 20	*TMEM231*	AR
Meckel syndrome 11	*TMEM231*	AR

Disorders	Genes	Inheritance
COACH syndrome 1	*TMEM67*	AR
Joubert syndrome 6	*TMEM67*	AR
Meckel syndrome 3	*TMEM67*	AR
RHYNS syndrome	*TMEM67*	AR
Nephronophthisis 11	*TMEM67*	AR
Thyroid dyshormonogenesis 2A	*TPO*	AR
Spinocerebellar ataxia, autosomal recessive 7	*TPP1*	AR
Ceroid lipofuscinosis, neuronal, 2	*TPP1*	AR
Spondyloepiphyseal dysplasia tarda	*TRAPPC2*	XLR
Aicardi-Goutieres syndrome 1, dominant and recessive	*TREX1*	AD/AR
Deafness, autosomal recessive 28	*TRIOBP*	AR
Short-rib thoracic dysplasia 4 with or without polydactyly	*TTC21B*	AR
Congenital myopathy 5 with cardiomyopathy	*TTN*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 10	*TTN*	AR
Ataxia with isolated vitamin E deficiency	*TTPA*	AR
Albinism, oculocutaneous, type IA	*TYR*	AR
Albinism, oculocutaneous, type IB	*TYR*	AR
Albinism, oculocutaneous, type III	*TYRP1*	AR
Crigler-Najjar syndrome, type I	*UGT1A1*	AR
Hyperbilirubinemia, familial transient neonatal	*UGT1A1*	AD/AR
Crigler-Najjar syndrome, type II	*UGT1A1*	AR
Hemophagocytic lymphohistiocytosis, familial, 3	*UNC13D*	AR
Usher syndrome, type 1C	*USH1C*	AR
Deafness, autosomal recessive 18A	*USH1C*	AR
Usher syndrome, type 1G	*USH1G*	AR
Usher syndrome, type 2A	*USH2A*	AR
Retinitis pigmentosa 39	*USH2A*	AR
Cohen syndrome	*VPS13B*	AR
von Willebrand disease, type 3	*VWF*	AR
Wiskott-Aldrich syndrome	*WAS*	XLR
Senior-Loken syndrome 8	*WDR19*	AR
Short-rib thoracic dysplasia 5 with or without polydactyly	*WDR19*	AR
Spermatogenic failure 72	*WDR19*	AR
Cranioectodermal dysplasia 4	*WDR19*	AR
Nephronophthisis 13	*WDR19*	AR
Wolfram syndrome 1	*WFS1*	AR
Usher syndrome, type 2D	*WHRN*	AR
Deafness, autosomal recessive 31	*WHRN*	AR
Osteogenesis imperfecta, type XV	*WNT1*	AR
Lymphoproliferative syndrome, X-linked, 2	*XIAP*	XLR
Xeroderma pigmentosum, group A	*XPA*	AR
Xeroderma pigmentosum, group C	*XPC*	AR
Grange syndrome	*YY1AP1*	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

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