alamya health

A Future Where Precision Medicine is the Standard

Carrier Screening

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Carrier Screening

Background

Reproductive genetic carrier screening is a test that looks at an individual's or a couple's genetic information, primarily to determine their likelihood of having a child with a serious childhood onset genetic condition. Such information can be helpful for family planning and pregnancy management.

Alamya Health's carrier screening tests approximately 1,300 genetic disorders, adapted from the Australian carrier screening list (https://www.nature.com/articles/s41431-020-0685-x). The table below lists the genes that are screened in this comprehensive test, along with the names of the disorders associated with pathogenic variants (aka mutations) in these genes. The OMIM number for each disorder is also listed. OMIM - Online Mendelian Inheritance in Man - is an online encyclopedia of genetic disorders. It can be accessed at OMIM. This open resource provides additional genetic and clinical information.

The Alamya Health team updates this list with additions and removals of genes based on current scientific knowledge. Relevant resources and references will be provided for positive test findings.

Typically, being identified as a carrier is not directly associated with health concerns for that individual. However, variants detected in a small subset of disorders can have health implications for individuals who are identified to be carriers. In such situations, the report will include potential health concerns and management resources.

It is important to note that this reproductive genetic carrier screening does not test for the risk of chromosomal disorders such as Down syndrome. Chromosomal disorders are caused by extra or missing copies/parts of chromosomes. You may wish to speak with your healthcare provider or a genetic counselor about your chance of having a child with a chromosome disorder.

Limitations

Reproductive genetic carrier screening will identify most people who are carriers of pathogenic or likely pathogenic variants for the conditions screened. The interpretation of the testing results relies on the current knowledge of the genetic basis of these disorders. As this knowledge is constantly changing, there is a small chance that a person who is a genetic carrier may not be identified by the testing that is used. This can be related to the ancestry of the individual who is tested as well as limitations of the sequencing technology and interpretation parameters that are used.

The term screening is used because not all the genes in a person's DNA are screened, not all genetic conditions are covered, and not all gene variants can be identified by the testing technology. A 'negative' result means the chance that a person or couple will have a child with any of the genetic conditions screened is low but does not completely rule out the possibility.

Gene Symbol	Disease	OMIM Number
AQP2	Diabetes insipidus, nephrogenic	125800
ARFGEF2	Periventricular heterotopia with microcephaly	608097
ARG1	Argininemia	207800
ARHGEF9	Epileptic encephalopathy, early infantile, 8	300607
ARL13B	Joubert syndrome 8	612291
ARL6	Bardet-Biedl syndrome 3	600151
ARMC4	Ciliary dyskinesia, primary, 23	615451
ARPC1B	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease	617718
ARSA	Metachromatic leukodystrophy	250100
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	253200
ARV1	Epileptic encephalopathy, early infantile, 38	617020
ARX	Hydranencephaly with abnormal genitalia	300215
ASAH1	Farber lipogranulomatosis	228000
ASCC1	Barrett esophagus/esophageal adenocarcinoma	614266
ASL	Argininosuccinic aciduria	207900
ASNS	Asparagine synthetase deficiency	615574
ASPA	Canavan disease	271900
ASPM	Microcephaly 5, primary, autosomal recessive	608716
ASS1	Citrullinemia	215700
ATAD1	Hyperekplexia 4	618011
ATCAY	Ataxia, cerebellar, Cayman type	601238
ATF6	Achromatopsia 7	616517
ATM	Ataxia-telangiectasia	208900
ATOH7	Persistent hyperplastic primary vitreous, autosomal recessive	221900
ATP13A2	Spastic paraplegia 78, autosomal recessive	617225
ATP6AP1	Immunodeficiency 47	300972
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA	219200
ATP6V0A4	Renal tubular acidosis, distal, autosomal recessive	602722
ATP6V1B1	Renal tubular acidosis with deafness	267300
ATP7A	Menkes disease	309400
ATP7B	Wilson disease	277900
ATP8A2	Cerebellar ataxia, intellectual developmental disorder, and dysequilibrium syndrome 4	615268
ATP8B1	Cholestasis, progressive familial intrahepatic 1	211600
ATR	Seckel syndrome 1	210600
ATRX	Intellectual developmental disorder-hypotonic facies syndrome, X-linked	309580
AUH	3-methylglutaconic aciduria, type I	250950
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	615181
B3GALT6	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	271640
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects	245600
B3GLCT	Peters-plus syndrome	261540
B4GALNT1	Spastic paraplegia 26, autosomal recessive	609195
B4GALT7	Ehlers-Danlos syndrome, progeroid type, 1	130070
BBS1	Bardet-Biedl syndrome 1	209900
BBS10	Bardet-Biedl syndrome 10	615987
BBS12	Bardet-Biedl syndrome 12	615989
BBS2	Bardet-Biedl syndrome 2	615981
BBS4	Bardet-Biedl syndrome 4	615982
BBS5	Bardet-Biedl syndrome 5	615983
BBS7	Bardet-Biedl syndrome 7	615984
BBS9	Bardet-Biedl syndrome 9	615986
BCKDHA	Maple syrup urine disease, type Ia	248600
BCKDHB	Maple syrup urine disease, type Ib	248600
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency	614923
BCS1L	GRACILE syndrome	603358
BGN	Meester-Loeys syndrome, X-linked	300989

Gene Symbol	Disease	OMIM Number
BIN1	Myopathy, centronuclear, autosomal recessive	255200
BLM	Bloom syndrome	210900
BMPER	Diaphanospondylodysostosis	608022
BMPR1B	Acromesomelic dysplasia, Demirhan type	609441
BOLA3	Multiple mitochondrial dysfunctions syndrome 2	614299
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal	614498
BRF1	Cerebellofaciodental syndrome	616202
BRWD3	Intellectual developmental disorder, X-linked 93	300659
BSCL2	Encephalopathy, progressive, with or without lipodystrophy	615924
BSND	Bartter syndrome, type 4a	602522
BTK	Agammaglobulinemia and isolated hormone deficiency	307200
BUB1B	Mosaic variegated aneuploidy syndrome 1	257300
C12orf57	Temtamy syndrome	218340
C12orf65	Combined oxidative phosphorylation deficiency 7	613559
C19orf12	Neurodegeneration with brain iron accumulation 4	614298
C1QA	C1q deficiency	613652
C1QB	C1q deficiency	613652
C1QC	C1q deficiency	613652
C2CD3	Orofaciodigital syndrome XIV	615948
C3	C3 deficiency	613779
C5	C5 deficiency	609536
C6	C6 deficiency	612446
C7	C7 deficiency	610102
C8B	C8 deficiency, type II	613789
C8orf37	Cone-rod dystrophy 16	614500
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	259730
CABP4	Cone-rod synaptic disorder, congenital nonprogressive	610427
CANT1	Desbuquois dysplasia	251450
CAPN3	Muscular dystrophy, limb-girdle, type 2A	253600
CARD11	Immunodeficiency 11	615206
CARD9	Candidiasis, familial, 2, autosomal recessive	212050
CARS2	Combined oxidative phosphorylation deficiency 27	616672
CASK	Intellectual developmental disorder, with or without nystagmus	300422
	Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia	300749
	FG syndrome 4	300422
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2	611938
CASR	Hyperparathyroidism, neonatal	239200
CAVIN1	Lipodystrophy, congenital generalized, type 4	613327
CBS	Homocystinuria, B6-responsive and nonresponsive types	236200
CC2D1A	Intellectual developmental disorder, autosomal recessive 3	608443
CC2D2A	Joubert syndrome 9	612285
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1	235510
CCDC103	Ciliary dyskinesia, primary, 17	614679
CCDC114	Ciliary dyskinesia, primary, 20	615067
CCDC115	Congenital disorder of glycosylation, type IIo	616828
CCDC39	Ciliary dyskinesia, primary, 14	613807
CCDC40	Ciliary dyskinesia, primary, 15	613808
CCDC8	3-M syndrome 3	614205
CCDC88C	Hydrocephalus, nonsyndromic, autosomal recessive	236600
CCN6	Arthropathy, progressive pseudorheumatoid, of childhood	208230
CCNO	Ciliary diskinesia, primary, 29	615872
CD27	Lymphoproliferative syndrome 2	615122
CD3D	Immunodeficiency 19	615617
CD40	Immunodeficiency with hyper-IgM, type 3	606843
CD40LG	Immunodeficiency, X-linked, with hyper-IgM	308230
CD55	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy	226300
CD81	Immunodeficiency, common variable, 6	613496
CDC45	Meier-Gorlin syndrome 7	617063

Gene Symbol	Disease	OMIM Number
CDH11	Elsahy-Waters syndrome	211380
CDH23	Usher syndrome, type 1D	601067
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy	225280
CDK10	Al Kaissi syndrome	617694
CDK5RAP2	Microcephaly 3, primary, autosomal recessive	604804
CDT1	Meier-Gorlin syndrome 4	613804
CENPJ	Microcephaly 6, primary, autosomal recessive	608393
CEP120	Short-rib thoracic dysplasia 13 with or without polydactyly	616300
CEP152	Seckel syndrome 5	613823
CEP290	Joubert syndrome 5	610188
CEP41	Joubert syndrome 15	614464
CEP78	Cone-rod dystrophy and hearing loss	617236
CERS3	Ichthyosis, congenital, autosomal recessive 9	615023
CFAP410	Retinal dystrophy with macular staphyloma	617547
CFD	Complement factor D deficiency	613912
CFH	Complement factor H deficiency	609814
CFI	Complement factor I deficiency	610984
CFL2	Nemaline myopathy 7, autosomal recessive	610687
CFP	Properdin deficiency, X-linked	312060
CFTR	Cystic fibrosis	219700
CHAT	Myasthenic syndrome, congenital, 6, presynaptic	254210
CHKB	Muscular dystrophy, congenital, megaconial type	602541
CHM	Choroideremia	303100
CHRNA1	Multiple pterygium syndrome, lethal type	253290
CHRND	Myasthenic syndrome, congenital, 3B, fast-channel	616322
CHRNE	Myasthenic syndrome, congenital, 4A, slow-channel	605809
CHRNG	Escobar syndrome	265000
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1	601776
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations	143095
CHSY1	Temtamy preaxial brachydactyly syndrome	605282
CIB2	Usher syndrome, type IJ	614869
CIITA	Bare lymphocyte syndrome, type II, complementation group A	209920
CISD2	Wolfram syndrome 2	604928
CIT	Microcephaly 17, primary, autosomal recessive	617090
CKAP2L	Filippi syndrome	272440
CLCF1	Cold-induced sweating syndrome 2	610313
CLCN2	Leukoencephalopathy with ataxia	615651
CLCN4	Intellectual developmental disorder, X-linked 49/15	300114
CLCN5	Dent disease	300009
CLCN7	Osteopetrosis, autosomal recessive 4	611490
CLCNKB	Bartter syndrome, type 4b, digenic	613090
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	607626
CLDN10	HELIX syndrome	617671
CLDN19	Hypomagnesemia 5, renal, with ocular involvement	248190
CLMP	Congenital short bowel syndrome	615237
CLN3	Ceroid lipofuscinosis, neuronal, 3	204200
CLN5	Ceroid lipofuscinosis, neuronal, 5	256731
CLN6	Ceroid lipofuscinosis, neuronal 6	601780
CLN8	Ceroid lipofuscinosis, neuronal, 8	600143
CLP1	Pontocerebellar hypoplasia, type 10	615803
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia	616271
CLPP	Perrault syndrome 3	614129
CLRN1	Usher syndrome, type 3A	276902
CNGA3	Achromatopsia-2	216900
CNGB3	Macular degeneration, juvenile	248200
CNNM4	Jalili syndrome	217080
CNTNAP1	Lethal congenital contracture syndrome 7	616286
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome	610042

Gene Symbol	Disease	OMIM Number
COA8	Mitochondrial complex IV deficiency	220110
COG6	Congenital disorder of glycosylation, type III	614576
COG7	Congenital disorder of glycosylation, type IIe	608779
COL11A1	Fibrochondrogenesis 1	228520
COL11A2	Fibrochondrogenesis 2	614524
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type	226650
COL18A1	Knobloch syndrome, type 1	267750
COL27A1	Steel Syndrome	615155
COL2A1	Otospondylomegaepiphyseal dysplasia	215150
COL4A3	Alport syndrome, autosomal recessive	203780
COL4A4	Alport syndrome, autosomal recessive	203780
COL4A5	Alport syndrome 1, X-linked	301050
COL6A1	Ullrich congenital muscular dystrophy 1	254090
COL6A2	Ullrich congenital muscular dystrophy 1	254090
COL6A3	Ullrich congenital muscular dystrophy 1	254090
COL7A1	Epidermolysis bullosa dystrophica, AR	226600
COLEC11	3MC syndrome 2	265050
COLQ	Myasthenic syndrome, congenital, 5	603034
COQ2	Coenzyme Q10 deficiency, primary, 1	607426
COQ4	Coenzyme Q10 deficiency, primary, 7	616276
COQ6	Coenzyme Q10 deficiency, primary, 6	614650
COQ8A	Coenzyme Q10 deficiency, primary, 4	612016
COQ8B	Nephrotic syndrome, type 9	615573
COX10	Leigh syndrome due to mitochondrial COX4 deficiency	256000
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2	615119
COX20	Mitochondrial complex IV deficiency	220110
CPLANE1	Joubert syndrome 17	614615
CPS1	Carbamoylphosphate synthetase I deficiency	237300
CPT1A	CPT deficiency, hepatic, type IA	255120
CPT2	CPT II deficiency, lethal neonatal	608836
CRB1	Leber congenital amaurosis 8	613835
CRB2	Ventriculomegaly with cystic kidney disease	219730
CRLF1	Cold-induced sweating syndrome 1	272430
CRPPA	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7	614643
CRTAP	Osteogenesis imperfecta, type VII	610682
CSPP1	Joubert syndrome 21	615636
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	254800
CTC1	Cerebroretinal microangiopathy with calcifications and cysts	612199
CTNS	Cystinosis, nephropathic	219800
CTPS1	Immunodeficiency 24	615897
CTSA	Galactosialidosis	256540
CTSC	Papillon-Lefevre syndrome	245000
CTSD	Ceroid lipofuscinosis, neuronal, 10	610127
CTSF	Ceroid lipofuscinosis, neuronal, 13, Kufs type	615362
CTSK	Pycnodysostosis	265800
CUL4B	Intellectual developmental disorder, X-linked, syndromic 15 (Cabezas type)	300354
CUL7	3-M syndrome 1	273750
CWC27	Retinitis pigmentosa with or without skeletal anomalies	250410
CYB5R3	Methemoglobinemia, type I	250800
CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA	233690
CYBB	Chronic granulomatous disease, X-linked	306400
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	613743
CYP11B2	Hypoaldosteronism, congenital, due to CMO I deficiency	203400
CYP17A1	17,20-lyase deficiency, isolated	202110
CYP1B1	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset	231300
CYP27A1	Cerebrotendinous xanthomatosis	213700
CYP2U1	Spastic paraplegia 56, autosomal recessive	615030
CYP4F22	Ichthyosis, congenital, autosomal recessive 5	604777

Gene Symbol	Disease	OMIM Number
CYP7B1	Bile acid synthesis defect, congenital, 3	613812
D2HGDH	D-2-hydroxyglutaric aciduria	600721
DARS1	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	615281
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	611105
DBT	Maple syrup urine disease, type II	248600
DCAF17	Woodhouse-Sakati syndrome	241080
DCDC2	Nephronophthisis 19	616217
DCHS1	Van Maldergem syndrome 1	601390
DCLRE1C	Severe combined immunodeficiency, Athabascan type	602450
DCX	Lissencephaly, X-linked	300067
DDC	Aromatic L-amino acid decarboxylase deficiency	608643
DDHD2	Spastic paraplegia 54, autosomal recessive	615033
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type	271665
DDX11	Warsaw breakage syndrome	613398
DDX59	Orofaciodigital syndrome V	174300
DENND5A	Epileptic encephalopathy, early infantile, 49	617281
DGAT1	Diarrhea 7, protein-losing enteropathy type	615863
DGKE	Nephrotic syndrome, type 7	615008
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	251880
DHCR24	Desmosterolosis	602398
DHCR7	Smith-Lemli-Opitz syndrome	270400
DHDDS	Retinitis pigmentosa 59	613861
DHODH	Miller syndrome	263750
DIS3L2	Perlman syndrome	267000
DKC1	Dyskeratosis congenita, X-linked	305000
DLD	Dihydrolipoamide dehydrogenase deficiency	246900
DLG3	Intellectual developmental disorder, X-linked 90	300850
DLL3	Spondylocostal dysostosis 1, autosomal recessive	277300
DMD	Duchenne muscular dystrophy	310200
DNAAF1	Ciliary dyskinesia, primary, 13	613193
DNAAF3	Ciliary dyskinesia, primary, 2	606763
DNAAF4	Ciliary dyskinesia, primary, 25	615482
DNAAF5	Ciliary dyskinesia, primary, 18	614874
DNAH11	Ciliary dyskinesia, primary, 7, with or without situs inversus	611884
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus	608644
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus	244400
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus	612444
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient	617384
DNAJC19	3-methylglutaconic aciduria, type V	610198
DNAJC21	Bone marrow failure syndrome 3	617052
DNAJC6	Parkinson disease 19, juvenile-onset	615528
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	242860
DOCK2	Immunodeficiency 40	616433
DOCK6	Adams-Oliver syndrome 2	614219
DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive	243700
DOK7	Myasthenic syndrome, congenital, 10	254300
DOLK	Congenital disorder of glycosylation, type Im	610768
DONSON	Microcephaly, short stature, and limb abnormalities	617604
DPAGT1	Myasthenic syndrome, congenital, 13, with tubular aggregates	614750
DPH1	Developmental delay with short stature, dysmorphic features, and sparse hair	616901
DSP	Cardiomyopathy, dilated, with woolly hair and keratoderma	605676
DSTYK	Spastic paraplegia 23	270750
DYM	Dyggve-Melchior-Clausen disease	223800
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly	613091
DYNC2LI1	Short-rib thoracic dysplasia 15 with polydactyly	617088
DYSF	Muscular dystrophy, limb-girdle, type 2B	253601
EARS2	Combined oxidative phosphorylation deficiency 12	614924
ECEL1	Arthrogryposis, distal, type 5D	615065

Gene Symbol	Disease	OMIM Number
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	616277
EDA	Ectodermal dysplasia 1, hypohidrotic, X-linked	305100
EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive	224900
EFEMP2	Cutis laxa, autosomal recessive, type IB	614437
EFNB1	Craniofrontonasal dysplasia	304110
EIF2AK3	Wolcott-Rallison syndrome	226980
EIF2AK4	Pulmonary venoocclusive disease 2	234810
EIF2B1	Leukoencephalopathy with vanishing white matter	603896
EIF2B2	Leukoencephalopathy with vanishing white matter	603896
EIF2B3	Leukoencephalopathy with vanishing white matter	603896
EIF2B4	Leukoencephaly with vanishing white matter (3)	, 603896
EIF2B5	Leukoencephalopathy with vanishing white matter	603896
EIF2S3	MEHMO syndrome	300148
ELAC2	Combined oxidative phosphorylation deficiency 17	615440
ELP1	Dysautonomia, familial	223900
ELP2	Intellectual developmental disorder, autosomal recessive 58	617270
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked	310300
EMG1	Bowen-Conradi syndrome	211180
EML1	Band heterotopia	600348
ENPP1	Hypophosphatemic rickets, autosomal recessive, 2	613312
EOGT	Adams-Oliver syndrome 4	615297
EPCAM	Diarrhea 5, with tufting enteropathy, congenital	613217
EPG5	Vici syndrome	242840
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora)	254780
ERBB3	Lethal congenital contractural syndrome 2	607598
ERCC2	Cerebrooculofacioskeletal syndrome 2	610756
ERCC4	Fanconi anemia, complementation group Q	615272
ERCC5	Xeroderma pigmentosum, group G	278780
ERCC6	Cockayne syndrome, type B	133540
ERCC6L2	Bone marrow failure syndrome 2	615715
ERCC8	Cockayne syndrome, type A	216400
ESCO2	SC phocomelia syndrome	269000
ETFA	Glutaric acidemia IIA	231680
ETFB	Glutaric acidemia IIB	231680
ETFDH	Glutaric acidemia IIC	231680
ETHE1	Ethylmalonic encephalopathy	602473
EVC	Ellis-van Creveld syndrome	225500
EVC2	Ellis-van Creveld syndrome	225500
EXOSC3	Pontocerebellar hypoplasia, type 1B	614678
EXOSC8	Pontocerebellar hypoplasia, type 1C	616081
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities	617425
F2	Dysprothrombinemia	613679
F5	Factor V deficiency	227400
F7	Factor VII deficiency	227500
F8	Hemophilia A	306700
F9	Hemophilia B	306900
FA2H	Spastic paraplegia 35, autosomal recessive	612319
FAH	Tyrosinemia, type I	276700
FAM126A	Leukodystrophy, hypomyelinating, 5	610532
FAM161A	Retinitis pigmentosa 28	606068
FAM20C	Raine syndrome	259775
FANCA	Fanconi anemia, complementation group A	227650
FANCB	Fanconi anemia, complementation group B	300514
FANCC	Fanconi anemia, complementation group C	227645
FANCD2	Fanconi anemia, complementation group D2	227646
FANCE	Fanconi anemia, complementation group E	600901
FANCF	Fanconi anemia, complementation group F	603467
FANCG	Fanconi anemia, complementation group G	614082

Gene Symbol	Disease	OMIM Number
FANCI	Fanconi anemia, complementation group I	609053
FANCL	Fanconi anemia, complementation group L	614083
FARS2	Combined oxidative phosphorylation deficiency 14	614946
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2	616006
FBLN5	Cutis laxa, autosomal recessive, type IA	219100
FBP1	Fructose-1,6-bisphosphatase deficiency	229700
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	615471
FBXO7	Parkinson disease 15, autosomal recessive	260300
FERMT1	Kindler syndrome	173650
FERMT3	Leukocyte adhesion deficiency, type III	612840
FGA	Afibrinogenemia, congenital	202400
FGB	Afibrinogenemia, congenital	202400
FGD4	Charcot-Marie-Tooth disease, type 4H	609311
FGG	Afibrinogenemia, congenital	202400
FH	Fumarase deficiency	606812
FHL1	Emery-Dreifuss muscular dystrophy 6, X-linked	300696
FIG4	Yunis-Varon syndrome	216340
FKBP10	Bruck syndrome 1	259450
FKBP14	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss	614557
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	613153
FKTN	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	253800
FLAD1	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency	255100
FLNA	FG syndrome 2	300321
FLNB	Spondylocarpotarsal synostosis syndrome	272460
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa	609033
FLVCR2	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome	225790
FMR1	Fragile X syndrome	300624
FOLR1	Neurodegeneration due to cerebral folate transport deficiency	613068
FOXE3	Aphakia, congenital primary	610256
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	601705
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	304790
FOXRED1	Mitochondrial complex I deficiency	252010
FRAS1	Fraser syndrome	219000
FREM1	Bifid nose with or without anorectal and renal anomalies	608980
FREM2	Fraser syndrome	219000
FRRS1L	Epileptic encephalopathy, early infantile, 37	616981
FTCD	Glutamate formiminotransferase deficiency	229100
FTO	Growth retardation, developmental delay, coarse facies, and early death	612938
FTSJ1	Intellectual developmental disorder, X-linked 9	309549
FUCA1	Fucosidosis	230000
FYCO1	Cataract 18, autosomal recessive	610019
G6PC	Glycogen storage disease Ia	232200
G6PC3	Dursun syndrome	612541
GAA	Glycogen storage disease II	232300
GALC	Krabbe disease	245200
GALNS	Mucopolysaccharidosis IVA	253000
GAMT	Cerebral creatine deficiency syndrome 2	612736
GAN	Giant axonal neuropathy-1	256850
GAS8	Ciliary dyskinesia, primary, 33	616726
GATM	Cerebral creatine deficiency syndrome 3	612718
GBA	Gaucher disease, perinatal lethal	608013
GBA2	Spastic paraplegia 46, autosomal recessive	614409
GBE1	Glycogen storage disease IV	232500
GCDH	Glutaricaciduria, type I	231670
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia	128230
GDAP1	Charcot-Marie-Tooth disease, recessive intermediate, A	608340
GDF1	Right atrial isomerism	208530
GDF5	Chondrodysplasia, Grebe type	200700

Gene Symbol	Disease	OMIM Number
GDI1	Intellectual developmental disorder, X-linked 41	300849
GFM1	Combined oxidative phosphorylation deficiency 1	609060
GFPT1	Myasthenia, congenital, 12, with tubular aggregates	610542
GHR	Laron dwarfism	262500
GJA1	Hypoplastic left heart syndrome 1	241550
GJB2	Autosomal recessive deafness-1A	220290
GJC2	Leukodystrophy, hypomyelinating, 2	608804
GJB2	Deafness, autosomal recessive 1A,	220290
GK	Glycerol kinase deficiency	307030
GLA	Fabry disease	301500
GLB1	Mucopolysaccharidosis type IVB (Morquio)	253010
GLDC	Glycine encephalopathy	605899
GLDN	Lethal congenital contracture syndrome 11	617194
GLE1	Arthrogryposis, lethal, with anterior horn cell disease	611890
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism	610199
GLYCTK	D-glyceric aciduria	220120
GM2A	GM2-gangliosidosis, AB variant	272750
GMPPA	Alacrima, achalasia, and intellectual developmental disorder syndrome	615510
GMPPB	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14	615352
GNAT2	Achromatopsia-4	613856
GNB5	Intellectual developmental disorder with cardiac arrhythmia	617173
GNE	Inclusion body myopathy, autosomal recessive	600737
GNPAT	Chondrodysplasia punctata, rhizomelic, type 2	222765
GNPTAB	Mucolipidosis III alpha/beta	252600
GNPTG	Mucolipidosis III gamma	252605
GNS	Mucopolysaccharidosis type IIID	252940
GORAB	Geroderma osteodysplasticum	231070
GOSR2	Epilepsy, progressive myoclonic 6	614018
GPAA1	Glycosylphosphatidylinositol biosynthesis defect 15	617810
GPC3	Simpson-Golabi-Behmel syndrome, type 1	312870
GPC6	Omodysplasia 1	258315
GPHN	Molybdenum cofactor deficiency C	615501
GPR143	Ocular albinism, type I, Nettleship-Falls type	300500
GPR179	Night blindness, congenital stationary (complete), 1E, autosomal recessive	614565
GPSM2	Chudley-McCullough syndrome	604213
GPT2	Intellectual developmental disorder, autosomal recessive 49	616281
GRM1	Spinocerebellar ataxia, autosomal recessive 13	614831
GSS	Glutathione synthetase deficiency	266130
GTF2H5	Trichothiodystrophy 3, photosensitive	616395
GTPBP3	Combined oxidative phosphorylation deficiency 23	616198
GUCY1A1	Moyamoya 6 with achalasia	615750
GUCY2C	Meconium ileus	614665
GUCY2D	Leber congenital amaurosis 1	204000
GUSB	Mucopolysaccharidosis VII	253220
GYS2	Glycogen storage disease 0, liver	240600
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures	616756
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency	231530
HADHA	Fatty liver, acute, of pregnancy	609016
HADHB	Trifunctional protein deficiency	609015
HAMP	Hemochromatosis, type 2B	613313
HAX1	Neutropenia, severe congenital 3, autosomal recessive	610738
HBB	Thalassemias, beta-, (3)	613985
HCFC1	Intellectual developmental disorder, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	309541
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A	613925
HERC2	Intellectual developmental disorder, autosomal recessive 38	615516
HES7	Spondylocostal dysostosis 4, autosomal recessive	613686
HESX1	Septooptic dysplasia	182230

Gene Symbol	Disease	OMIM Number
HEXA	Tay-Sachs disease	272800
HEXB	Sandhoff disease, infantile, juvenile, and adult forms	268800
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C)	252930
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency	250620
HINT1	Neuromyotonia and axonal neuropathy, autosomal recessive	137200
HJV	Hemochromatosis, type 2A	602390
HK1	Neuropathy, hereditary motor and sensory, Russe type	605285
HLCS	Holocarboxylase synthetase deficiency	253270
HMGCL	HMG-CoA lyase deficiency	246450
HMGCS2	HMG-CoA synthase-2 deficiency	605911
HOXA1	Athabaskan brainstem dysgenesis syndrome	601536
HPD	Tyrosinemia, type III	276710
HPGD	Cranioosteoarthropathy	259100
HPRT1	Lesch-Nyhan syndrome	300322
HPS1	Hermansky-Pudlak syndrome 1	203300
HPS3	Hermansky-Pudlak syndrome 3	614072
HPS4	Hermansky-Pudlak syndrome 4	614073
HPS5	Hermansky-Pudlak syndrome 5	614074
HPS6	Hermansky-Pudlak syndrome 6	614075
HPSE2	Urofacial syndrome 1	236730
HSD17B10	HSD10 mitochondrial disease	300438
HSD17B4	D-bifunctional protein deficiency	261515
HSD3B2	3-beta-hydroxysteroid dehydrogenase, type II, deficiency	201810
HSD3B7	Bile acid synthesis defect, congenital, 1	607765
HSPD1	Leukodystrophy, hypomyelinating, 4	612233
HSPG2	Schwartz-Jampel syndrome, type 1	255800
HTRA2	3-methylglutaconic aciduria, type VIII	617248
HUWE1	Intellectual developmental disorder, X-linked syndromic, Turner type	300706
HYDIN	Ciliary dyskinesia, primary, 5	608647
HYLS1	Hydrolethalus syndrome	236680
IARS1	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy	617093
IARS2	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	616007
IBA57	Multiple mitochondrial dysfunctions syndrome 3	615330
ICOS	Immunodeficiency, common variable, 1	607594
IDS	Mucopolysaccharidosis II	309900
IDUA	Mucopolysaccharidosis Ih	607014
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome	614231
IFNGR1	Immunodeficiency 27A, mycobacteriosis, AR	209950
IFNGR2	Immunodeficiency 28, mycobacteriosis	614889
IFT122	Cranioectodermal dysplasia 1	218330
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly	266920
IFT172	Short-rib thoracic dysplasia 10 with or without polydactyly	615630
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly	611263
IGF1R	Insulin-like growth factor I, resistance to	270450
IGFBP7	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis	614224
IGHM	Agammaglobulinemia 1	601495
IGHMBP2	Neuronopathy, distal hereditary motor, type VI	604320
IKBKB	Immunodeficiency 15	615592
IKBKG	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency	300301
IL10RA	Inflammatory bowel disease 28, early onset, autosomal recessive	613148
IL11RA	Craniosynostosis and dental anomalies	614188
IL12RB1	Immunodeficiency 30	614891
IL17RA	Immunodeficiency 51	613953
IL1RAPL1	Intellectual developmental disorder, X-linked 21/34	300143
IL1RN	Interleukin 1 receptor antagonist deficiency	612852
IL2RG	Severe combined immunodeficiency, X-linked	300400
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	608971

Gene Symbol	Disease	OMIM Number
IMPG2	Retinitis pigmentosa 56	613581
INPP5E	Joubert syndrome 1	213300
INPP5K	Muscular dystrophy, congenital, with cataracts and intellectual disability	617404
INPPL1	Opsismodysplasia	258480
INSR	Leprechaunism	246200
INVS	Nephronophthisis 2, infantile	602088
IQCB1	Senior-Loken syndrome 5	609254
IQSEC2	Intellectual developmental disorder, X-linked 1	309530
ISCA2	Multiple mitochondrial dysfunctions syndrome 4	616370
ITCH	Autoimmune disease, multisystem, with facial dysmorphism	613385
ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis	226730
ITGB2	Leukocyte adhesion deficiency	116920
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia	226730
ITK	Lymphoproliferative syndrome 1	613011
ITPR1	Gillespie syndrome	206700
IVD	Isovaleric acidemia	243500
JAGN1	Neutropenia, severe congenital, 6, autosomal recessive	616022
JAK3	SCID, autosomal recessive, T-negative/B-positive type	600802
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	613730
JUP	Naxos disease	601214
KATNB1	Lissencephaly 6, with microcephaly	616212
KCNE1	Jervell and Lange-Nielsen syndrome 2	612347
KCNJ1	Bartter syndrome, type 2	241200
KCNJ10	SESAME syndrome	612780
KCNJ11	Hyperinsulinemic hypoglycemia, familial, 2	601820
KCNQ1	Jervell and Lange-Nielsen syndrome	220400
KCNV2	Retinal cone dystrophy 3B	610356
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions	611726
KDM5C	Intellectual developmental disorder, X-linked, syndromic, Claes-Jensen type	300534
KIAA0586	Short-rib thoracic dysplasia 14 with polydactyly	616546
KIAA1109	Alkuraya-Kucinskas syndrome	617822
KIF14	Microcephaly 20, primary, autosomal recessive	617914
KIF1A	Spastic paraplegia 30, autosomal recessive	610357
KIF1C	Spastic ataxia 2, autosomal recessive	611302
KIF7	Hydrolethalus syndrome 2	614120
KIFBP	Goldberg-Shprintzen megacolon syndrome	609460
KLHL40	Nemaline myopathy 8, autosomal recessive	615348
KLHL41	Nemaline myopathy 9	615731
KLHL7	PERCHING syndrome	617055
KNL1	Microcephaly 4, primary, autosomal recessive	604321
KPTN	Intellectual developmental disorder, autosomal recessive 41	615637
KRT10	Epidermolytic hyperkeratosis	113800
KRT14	Epidermolysis bullosa simplex, recessive 1	601001
KRT5	Epidermolysis bullosa simplex, recessive 1	601001
KRT8	Cirrhosis, cryptogenic	215600
KRT85	Ectodermal dysplasia 4, hair/nail type	602032
KY	Myopathy, myofibrillar, 7	617114
L1CAM	MASA syndrome	303350
L2HGDH	L-2-hydroxyglutaric aciduria	236792
LAMA1	Poretti-Boltshauser syndrome	615960
LAMA2	Muscular dystrophy, congenital merosin-deficient	607855
LAMA3	Epidermolysis bullosa, junctional, Herlitz type	226700
LAMB1	Lissencephaly 5	615191
LAMB2	Pierson syndrome	609049
LAMB3	Epidermolysis bullosa, junctional, Herlitz type	226700
LAMC2	Epidermolysis bullosa, junctional, Herlitz type	226700
LAMC3	Cortical malformations, occipital	614115
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6	613154

Gene Symbol	Disease	OMIM Number
LARP7	Alazami syndrome	615071
LARS1	Infantile liver failure syndrome 1	615438
LARS2	Perrault syndrome 4	615300
LAT	Immunodeficiency 52	617514
LBR	Greenberg skeletal dysplasia	215140
LCA5	Leber congenital amaurosis 5	604537
LCAT	Norum disease	245900
LDHA	Glycogen storage disease XI	612933
LDHB	Lactate dehydrogenase-B deficiency	614128
LDLR	LDL cholesterol level QTL2/Hypercholesterolemia, familial	143890
LDLRAP1	Hypercholesterolemia, familial, autosomal recessive	603813
LEP	Obesity, morbid, due to leptin deficiency	614962
LGI4	Arthrogryposis multiplex congenita, neurogenic, with myelin defect	617468
LHX3	Pituitary hormone deficiency, combined, 3	221750
LIAS	Pyruvate dehydrogenase lipoic acid synthetase deficiency	614462
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	601559
LIG4	LIG4 syndrome	606593
LINS1	Intellectual developmental disorder, autosomal recessive 27	614340
LIPA	Cholesteryl ester storage disease	278000
LIPC	Hepatic lipase deficiency	614025
LIPT1	Lipoyltransferase 1 deficiency	616299
LMAN1	Combined factor V and VIII deficiency	227300
LMBR1	Acheiropody	200500
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	277380
LMNA	Restrictive dermopathy, lethal	275210
LMOD3	Nemaline myopathy 10	616165
LONP1	CODAS syndrome	600373
LPIN1	Myoglobinuria, acute recurrent, autosomal recessive	268200
LPIN2	Majeed syndrome	609628
LPL	Lipoprotein lipase deficiency	238600
LRAT	Leber congenital amaurosis 14	613341
LRBA	Immunodeficiency, common variable, 8, with autoimmunity	614700
LRIG2	Urofacial syndrome 2	615112
LRMDA	Albinism, oculocutaneous, type VII	615179
LRP2	Donnai-Barrow syndrome	222448
LRP4	Cenani-Lenz syndactyly syndrome	212780
LRP5	Osteoporosis-pseudoglioma syndrome	259770
LRPPRC	Leigh syndrome, French-Canadian type	220111
LRRC6	Ciliary dyskinesia, primary, 19	614935
LRSAM1	Charcot-Marie-Toothe disease, axonal, type 2P	614436
LTBP3	Tooth agenesis, selective, 6	613097
LTBP4	Cutis laxa, autosomal recessive, type IC	613177
LYRM7	Mitochondrial complex III deficiency, nuclear type 8	615838
LYST	Chediak-Higashi syndrome	214500
LZTFL1	Bardet-Biedl syndrome 17	615994
MALT1	Immunodeficiency 12	615468
MAN1B1	Intellectual developmental disorder, autosomal recessive 15	614202
MAN2B1	Mannosidosis, alpha-, types I and II	248500
MANBA	Mannosidosis, beta	248510
MAOA	Brunner syndrome	300615
MAPKBP1	Nephronophthisis 20	617271
MARS1	Interstitial lung and liver disease	615486
MARS2	Spastic ataxia 3, autosomal recessive	611390
MASP1	3MC syndrome 1	257920
MBOAT7	Intellectual developmental disorder, autosomal recessive 57	617188
MBTPS2	IFAP syndrome with or without BRESHECK syndrome	308205
MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness	202200
MCFD2	Factor V and factor VIII, combined deficiency of	613625

Gene Symbol	Disease	OMIM Number
MCM4	Natural killer cell and glucocorticoid deficiency with DNA repair defect	609981
MCOLN1	Mucolipidosis IV	252650
MCPH1	Microcephaly 1, primary, autosomal recessive	251200
MECP2	Encephalopathy, neonatal severe	300673
MED12	Lujan-Fryns syndrome	309520
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy	613668
MED23	Intellectual developmental disorder, autosomal recessive 18	614249
MED25	Basel-Vanagait-Smirin-Yosef syndrome	616449
MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	614399
MEGF8	Carpenter syndrome 2	614976
MERTK	Retinitis pigmentosa 38	613862
MESP2	Spondylocostal dysostosis 2, autosomal recessive	608681
METTL23	Intellectual developmental disorder, autosomal recessive 44	615942
MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2B	617087
MFSD2A	Microcephaly 15, primary, autosomal recessive	616486
MFSD8	Ceroid lipofuscinosis, neuronal, 7	610951
MGAT2	Congenital disorder of glycosylation, type IIa	212066
MGME1	Mitochondrial DNA depletion syndrome 11	615084
MGP	Keutel syndrome	245150
MICU1	Myopathy with extrapyramidal signs	615673
MID1	Opitz GBBB syndrome, type I	300000
MKKS	McKusick-Kaufman syndrome	236700
MKS1	Meckel syndrome 1	249000
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	604004
MLYCD	Malonyl-CoA decarboxylase deficiency	248360
MMAA	Methylmalonic aciduria, vitamin B12-responsive	251100
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	251110
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	277400
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	277410
MMP2	Multicentric osteolysis, nodulosis, and arthropathy	259600
MMP21	Heterotaxy, visceral, 7, autosomal	616749
MMUT	Methylmalonic aciduria, mut(0) type	251000
MOCS1	Molybdenum cofactor deficiency A	252150
MOCS2	Molybdenum cofactor deficiency B	252160
MPDZ	Hydrocephalus, nonsyndromic, autosomal recessive 2	615219
MPI	Congenital disorder of glycosylation, type Ib	602579
MPL	Thrombocytopenia, congenital amegakaryocytic	604498
MPLKIP	Trichothiodystrophy 4, nonphotosensitive	234050
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	256810
MPZ	Dejerine-Sottas disease	145900
MRAP	Glucocorticoid deficiency 2	607398
MRE11	Ataxia-telangiectasia-like disorder	604391
MTFMT	Combined oxidative phosphorylation deficiency 15	614947
MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	617780
MTHFR	Homocystinuria due to MTHFR deficiency	236250
MTM1	Myotubular myopathy, X-linked	310400
MTMR2	Charcot-Marie-Tooth disease, type 4B1	601382
MTO1	Combined oxidative phosphorylation deficiency 10	614702
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type	250940
MTRR	Homocystinuria-megaloblastic anemia, cbl E type	236270
MTTP	Abetalipoproteinemia	200100
MUSK	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency	616325
MVK	Mevalonic aciduria	610377
MYD88	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	612260
MYMK	Carey-Fineman-Ziter syndrome	254940
MYO5B	Microvillus inclusion disease	251850

Gene Symbol	Disease	OMIM Number
MYO7A	Usher syndrome, type 1B	276900
NAA10	N-terminal acetyltransferase deficiency	300855
NAGA	Schindler disease, type I	609241
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)	252920
NAGS	N-acetylglutamate synthase deficiency	237310
NALCN	Hypotonia, infantile, with psychomotor retardation and characteristic facies	615419
NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type	610442
NARS2	Combined oxidative phosphorylation deficiency 24	616239
NAXE	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy	617186
NBAS	Short stature, optic nerve atrophy, and Pelger-Huet anomaly	614800
NBN	Nijmegen breakage syndrome	251260
NCF1	Chronic granulomatous disease due to deficiency of NCF-1	233700
NCF2	Chronic granulomatous disease due to deficiency of NCF-2	233710
NDE1	Lissencephaly 4 (with microcephaly)	614019
NDP	Norrie disease	310600
NDRG1	Charcot-Marie-Tooth disease, type 4D	601455
NDUFA1	Mitochondrial complex I deficiency	252010
NDUFA10	Leigh syndrome	256000
NDUFA11	Mitochondrial complex I deficiency	252010
NDUFAF2	Leigh syndrome	256000
NDUFAF5	Mitochondrial complex 1 deficiency	252010
NDUFAF6	Leigh syndrome due to mitochondrial complex I deficiency	256000
NDUFS1	Mitochondrial complex I deficiency	252010
NDUFS2	Mitochondrial complex I deficiency	252010
NDUFS4	Leigh syndrome	256000
NDUFS6	Mitochondrial complex I deficiency	252010
NDUFS7	Leigh syndrome	256000
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency	256000
NDUFV1	Mitochondrial complex I deficiency	252010
NDUFV2	Mitochondrial complex I deficiency	252010
NEB	Nemaline myopathy 2, autosomal recessive	256030
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome	225060
NEK1	Short-rib thoracic dysplasia 6 with or without polydactyly	263520
NEK8	Renal-hepatic-pancreatic dysplasia 2	615415
NEU1	Sialidosis, type I	256550
NEXMIF	Intellectual developmental disorder, X-linked 98	300912
NFU1	Multiple mitochondrial dysfunctions syndrome 1	605711
NGF	Neuropathy, hereditary sensory and autonomic, type V	608654
NGLY1	Congenital disorder of deglycosylation	615273
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	611291
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora)	254780
NHS	Cataract 40, X-linked	302200
NIPAL4	Ichthyosis, congenital, autosomal recessive 6	612281
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia	613330
NKX6-2	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy	617560
NLGN4X	Intellectual developmental disorder, X-linked	300495
NMNAT1	Leber congenital amaurosis 9	608553
NNT	Glucocorticoid deficiency 4	614736
NPC1	Niemann-Pick disease, type C1	257220
NPC2	Niemann-pick disease, type C2	607625
NPHP1	Joubert syndrome 4	609583
NPHP3	Meckel syndrome 7	267010
NPHP4	Senior-Loken syndrome 4	606996
NPHS1	Nephrotic syndrome, type 1	256300
NPHS2	Nephrotic syndrome, type 2	600995
NPR2	Acromesomelic dysplasia, Maroteaux type	602875
NR0B1	46XY sex reversal 2, dosage-sensitive	300018

Gene Symbol	Disease	OMIM Number
NSDHL	CK syndrome	300831
NSUN2	Intellectual developmental disorder, autosomal recessive 5	611091
NT5C2	Spastic paraplegia 45	613162
NTRK1	Insensitivity to pain, congenital, with anhidrosis	256800
NUBPL	Mitochondrial complex I deficiency	252010
NUP107	Nephrotic syndrome, type 11	616730
NUP62	Striatonigral degeneration, infantile	271930
NUP93	Nephrotic syndrome, type 12	616892
NYX	Night blindness, congenital stationary (complete), 1A, X-linked	310500
OBSL1	3-M syndrome 2	612921
OCA2	Albinism, brown oculocutaneous	203200
OCLN	Band-like calcification with simplified gyration and polymicrogyria	251290
OCRL	Lowe syndrome	309000
OFD1	Joubert syndrome 10	300804
OPA1	Behr syndrome	210000
OPA3	3-methylglutaconic aciduria, type III	258501
OPHN1	Intellectual developmental disorder, X-linked, with cerebellar hypoplasia and distinctive facial appearance	300486
OPN1LW	Blue cone monochromacy	303700
ORAI1	Immunodeficiency 9	612782
ORC1	Meier-Gorlin syndrome 1	224690
ORC6	Meier-Gorlin syndrome 3	613803
OSGEP	Galloway-Mowat syndrome 3	617729
OSTM1	Osteopetrosis, autosomal recessive 5	259720
OTC	Ornithine transcarbamylase deficiency	311250
OTUD6B	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies	617452
P3H1	Osteogenesis imperfecta, type VIII	610915
PAH	Phenylketonuria	261600
PAK3	Intellectual developmental disorder, X-linked 30/47	300558
PANK2	Neurodegeneration with brain iron accumulation 1	234200
PAPSS2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes	612847
PC	Pyruvate carboxylase deficiency	266150
PCCA	Propionicacidemia	606054
PCCB	Propionicacidemia	606054
PCDH12	Microcephaly, seizures, spasticity, and brain calcification	251280
PCDH15	Usher syndrome, type 1F	602083
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	210720
PCSK1	Obesity with impaired prohormone processing	600955
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy	608940
PDE6B	Retinitis pigmentosa-40	613801
PDE6C	Cone dystrophy 4	613093
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	312170
PDHB	Pyruvate dehydrogenase E1-beta deficiency	614111
PDP1	Pyruvate dehydrogenase phosphatase deficiency	608782
PEPD	Prolidase deficiency	170100
PET100	Mitochondrial complex IV deficiency	220110
PEX1	Peroxisome biogenesis disorder 1A (Zellweger)	214100
PEX10	Peroxisome biogenesis disorder 6A (Zellweger)	614870
PEX11B	Peroxisome biogenesis disorder 14B	614920
PEX12	Peroxisome biogenesis disorder 3A (Zellweger)	614859
PEX13	Peroxisome biogenesis disorder 11A (Zellweger)	614883
PEX16	Peroxisome biogenesis disorder 8A, (Zellweger)	614876
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)	614866
PEX26	Peroxisome biogenesis disorder 7A (Zellweger)	614872
PEX3	Peroxisome biogenesis disorder 10A (Zellweger)	614882
PEX5	Peroxisome biogenesis disorder 2A (Zellweger)	214110
PEX6	Peroxisome biogenesis disorder 4A (Zellweger)	614862
PEX7	Chondrodysplasia punctata, rhizomelic, type 1	215100

Gene Symbol	Disease	OMIM Number
PFKM	Glycogen storage disease VII	232800
PGAP1	Intellectual developmental disorder, autosomal recessive 42	615802
PGAP2	Hyperphosphatasia with intellectual developmental disorder syndrome 3	614207
PGAP3	Hyperphosphatasia with intellectual developmental disorder syndrome 4	615716
PGK1	Phosphoglycerate kinase 1 deficiency	300653
PGM1	Congenital disorder of glycosylation, type It	614921
PGM3	Immunodeficiency 23	615816
PHF6	Borjeson-Forssman-Lehmann syndrome	301900
PHF8	Intellectual developmental disorder syndrome, X-linked, Siderius type	300263
PHGDH	Neu-Laxova syndrome1	256520
PHYH	Refsum disease	266500
PIEZO2	Arthrogryposis, distal, with impaired proprioception and touch	617146
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2	300868
PIGG	Intellectual developmental disorder, autosomal recessive 53	616917
PIGL	CHIME syndrome	280000
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1	614080
PIGO	Hyperphosphatasia with intellectual developmental disorder syndrome 2	614749
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3	615398
PIGV	Hyperphosphatasia with intellectual developmental disorder syndrome 1	239300
PIH1D3	Ciliary dyskinesia, primary, 36, X-linked	300991
PIP5K1C	Lethal congenital contractural syndrome 3	611369
PKHD1	Polycystic kidney and hepatic disease	263200
PKLR	Pyruvate kinase deficiency	266200
PLA2G6	Neurodegeneration with brain iron accumulation 2B	610217
PLAA	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	617527
PLCE1	Nephrotic syndrome, type 3	610725
PLEC	Epidermolysis bullosa simplex with pyloric atresia	612138
PLG	Plasminogen deficiency, type I	217090
PLOD1	Ehlers-Danlos syndrome, type VI	225400
PLOD2	Bruck syndrome 2	609220
PLP1	Pelizaeus-Merzbacher disease	312080
PLPBP	Epilepsy, early-onset, vitamin B6-dependent	617290
PMM2	Congenital disorder of glycosylation, type Ia	212065
PMPCA	Spinocerebellar ataxia, autosomal recessive 2	213200
PNKP	Microcephaly, seizures, and developmental delay	613402
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency	613179
PNPLA6	Boucher-Neuhauser syndrome	215470
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	610090
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	614813
POLA1	Pigmentary disorder, reticulate, with systemic manifestations, X-linked	301220
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type)	203700
POLR1C	Treacher Collins syndrome 3	248390
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism	607694
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	614381
POMC	Obesity, adrenal insufficiency, and red hair due to POMC deficiency	609734
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	253280
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8	614830
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12	615249
POMP	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	601952
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	236670
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	613150
POP1	Anauxetic dysplasia 2	617396
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	201750
POU1F1	Pituitary hormone deficiency, combined, 1	613038
PPA2	Sudden cardiac failure, infantile	617222
PPT1	Ceroid lipofuscinosis, neuronal, 1	256730

Gene Symbol	Disease	OMIM Number
PQBP1	Renpenning syndrome	309500
PRDM12	Neuropathy, hereditary sensory and autonomic, type VIII	616488
PRDM5	Brittle cornea syndrome 2	614170
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	603553
PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	208250
PRICKLE1	Epilepsy, progressive myoclonic 1B	612437
PROC	Thrombophilia due to protein C deficiency, autosomal recessive	612304
PROP1	Pituitary hormone deficiency, combined, 2	262600
PROS1	Thrombophilia due to protein S deficiency, autosomal recessive	614514
PRPS1	Arts syndrome	301835
PRUNE1	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies	617481
PRX	Dejerine-Sottas disease	145900
PSAP	Metachromatic leukodystrophy due to SAP-b deficiency	249900
PSAT1	Neu-Laxova syndrome 2	616038
PSMB8	Autoinflammation, lipodystrophy, and dermatosis syndrome	256040
PSPH	Phosphoserine phosphatase deficiency	614023
PTH1R	Chondrodysplasia, Blomstrand type	215045
PTS	Hyperphenylalaninemia, BH4-deficient, A	261640
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	600462
PXDN	Corneal opacification and other ocular anomalies	269400
PYCR1	Cutis laxa, autosomal recessive, type IIB	612940
PYCR2	Leukodystrophy, hypomyelinating, 10	616420
PYROXD1	Myopathy, myofibrillar, 8	617258
QARS1	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	615760
QDPR	Hyperphenylalaninemia, BH4-deficient, C	261630
RAB18	Warburg micro syndrome 3	614222
RAB23	Carpenter syndrome	201000
RAB27A	Griscelli syndrome, type 2	607624
RAB33B	Smith-McCort dysplasia 2	615222
RAB39B	Intellectual developmental disorder, X-linked 72	300271
RAB3GAP1	Warburg micro syndrome 1	600118
RAB3GAP2	Warburg micro syndrome 2	614225
RAD50	Nijmegen breakage syndrome-like disorder	613078
RAG1	Severe combined immunodeficiency, B cell-negative	601457
RAG2	Severe combined immunodeficiency, B cell-negative	601457
RAPSN	Fetal akinesia deformation sequence	208150
RARB	Microphthalmia, syndromic 12	615524
RARS1	Leukodystrophy, hypomyelinating, 9	616140
RARS2	Pontocerebellar hypoplasia, type 6	611523
RAX	Microphthalmia, isolated 3	611038
RBBP8	Seckel syndrome 2	606744
RBCK1	Polyglucosan body myopathy 1 with or without immunodeficiency	615895
RBM10	TARP syndrome	311900
RCBTB1	Retinal dystrophy with or without extraocular anomalies	617175
RD3	Leber congenital amaurosis 12	610612
RDH12	Leber congenital amaurosis 13	612712
RECQL4	Baller-Gerold syndrome	218600
REEP6	Retinitis pigmentosa 77	617304
REN	Renal tubular dysgenesis	267430
RETREG1	Neuropathy, hereditary sensory and autonomic, type IIB	613115
RFT1	Congenital disorder of glycosylation, type In	612015
RFX6	Mitchell-Riley syndrome	615710
RFXANK	MHC class II deficiency, complementation group B	209920
RFXAP	Bare lymphocyte syndrome, type II, complementation group D	209920
RIN2	Macrocephaly, alopecia, cutis laxa, and scoliosis	613075
RIPK4	Popliteal pterygium syndrome 2, lethal type	263650
RLIM	Intellectual developmental disorder, X-linked 61	300978
RMND1	Combined oxidative phosphorylation deficiency 11	614922

Gene Symbol	Disease	OMIM Number
RMRP	Cartilage-hair hypoplasia	250250
RNASEH2A	Aicardi-Goutieres syndrome 4	610333
RNASEH2B	Aicardi-Goutieres syndrome 2	610181
RNASEH2C	Aicardi-Goutieres syndrome 3	610329
RNASET2	Leukoencephalopathy, cystic, without megalencephaly	612951
RNU4ATAC	Microcephalic osteodysplastic primordial dwarfism, type I	210710
ROBO3	Gaze palsy, horizontal, with progressive scoliosis	607313
ROGDI	Kohlschutter-Tonz syndrome	226750
ROR2	Robinow syndrome, autosomal recessive	268310
RORC	Immunodeficiency 42	616622
RP2	Retinitis pigmentosa 2	312600
RPE65	Leber congenital amaurosis 2	204100
RPGR	Macular degeneration, X-linked atrophic	300834
RPGRIP1	Cone-rod dystrophy 13	608194
RPGRIP1L	Meckel syndrome 5	611561
RPL10	Intellectual developmental disorder, X-linked, syndromic, 35	300998
RPS6KA3	Coffin-Lowry syndrome	303600
	Intellectual developmental disorder, X-linked 19	300844
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	612075
RSPH1	Ciliary dyskinesia, primary, 24	615481
RSPH4A	Ciliary dyskinesia, primary, 11	612649
RSPH9	Ciliary dyskinesia, primary, 12	612650
RTEL1	Dyskeratosis congenita, autosomal recessive 5	615190
RTN4IP1	Optic atrophy 10 with or without ataxia, intellectual developmental disorder, and seizures	616732
RTTN	Polymicrogyria with seizures	614833
RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10	615041
RYR1	Minicore myopathy with external ophthalmoplegia	255320
SACS	Spastic ataxia, Charlevoix-Saguenay type	270550
SAMD9	Tumoral calcinosis, familial, normophosphatemic	610455
SAMHD1	Aicardi-Goutieres syndrome 5	612952
SAR1B	Chylomicron retention disease	246700
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	613845
SBDS	Shwachman-Diamond syndrome	260400
SBF2	Charcot-Marie-Tooth disease, type 4B2	604563
SC5D	Lathosterolosis	607330
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure	254900
SCARF2	Van den Ende-Gupta syndrome	600920
SCN9A	Insensitivity to pain, congenital	243000
SCNN1A	Pseudohypoaldosteronism, type I	264350
SCNN1B	Pseudohypoaldosteronism, type I	264350
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	604377
SCYL1	Spinocerebellar ataxia, autosomal recessive 21	616719
SDCCAG8	Bardet-Biedl syndrome 16	615993
SDHAF1	Mitochondrial complex II deficiency	252011
SEC23A	Craniolenticulosutural dysplasia	607812
SEC23B	Dyserythropoietic anemia, congenital, type II	224100
SELENON	Muscular dystrophy, rigid spine, 1	602771
SEMA4A	Cone-rod dystrophy 10	610283
SEPSECS	Pontocerebellar hypoplasia type 2D	613811
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	614739
SERPINA1	Emphysema-cirrhosis, due to AAT deficiency	613490
SERPINF1	Osteogenesis imperfecta, type VI	613982
SERPINH1	Orofaciodigital syndrome VI	277170
SETX	Spinocerebellar ataxia, autosomal recessive 1	606002
SFTPB	Surfactant metabolism dysfunction, pulmonary, 1	265120
SGCA	Muscular dystrophy, limb-girdle, type 2D	608099
SGCB	Muscular dystrophy, limb-girdle, type 2E	604286
SGCD	Muscular dystrophy, limb-girdle, type 2F	601287

Gene Symbol	Disease	OMIM Number
SGCG	Muscular dystrophy, limb-girdle, type 2C	253700
SGO1	Chronic atrial and intestinal dysrhythmia	616201
SGPL1	Nephrotic syndrome 14	617575
SGSH	Mucopolysaccharidisis type IIIA (Sanfilippo A)	252900
SH2D1A	Lymphoproliferative syndrome, X-linked, 1	308240
SH3PXD2B	Frank-ter Haar syndrome	249420
SH3TC2	Charcot-Marie-Tooth disease, type 4C	601596
SHOX	Langer mesomelic dysplasia	249700
SIL1	Marinesco-Sjogren syndrome	248800
SKIV2L	Trichohepatoenteric syndrome 2	614602
SLC12A1	Bartter syndrome, type 1	601678
SLC12A5	Epileptic encephalopathy, early infantile, 34	616645
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	218000
SLC13A5	Epileptic encephalopathy, early infantile, 25	615905
SLC16A1	Monocarboxylate transporter 1 deficiency	616095
SLC16A2	Allan-Herndon-Dudley syndrome	300523
SLC17A5	Sialic acid storage disorder, infantile	269920
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	249270
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	607483
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	616657
SLC22A5	Carnitine deficiency, systemic primary	212140
SLC24A5	Albinism, oculocutaneous, type VI	113750
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria	615182
SLC25A13	Citrullinemia, type II, neonatal-onset	605814
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	238970
SLC25A19	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), (progressive polyneuropathy type),	613710
SLC25A22	Epileptic encephalopathy, early infantile, 3	609304
SLC25A38	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive	205950
SLC25A46	Neuropathy, hereditary motor and sensory, type VIB	616505
SLC26A2	Achondrogenesis Ib	600972
SLC26A3	Diarrhea 1, secretory chloride, congenital	214700
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome	602782
SLC2A10	Arterial tortuosity syndrome	208050
SLC2A2	Fanconi-Bickel syndrome	227810
SLC30A10	Hypermanganesemia with dystonia, polycythemia, and cirrhosis	613280
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration	614482
SLC35A3	Arthrogryposis, intellectual developmental disorder, and seizures	615553
SLC35D1	Schneckenbecken dysplasia	269250
SLC37A4	Glycogen storage disease Ib	232220
SLC38A8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis	609218
SLC39A14	Hypermanganesemia with dystonia 2	617013
SLC39A4	Acrodermatitis enteropathica	201100
SLC39A8	Congenital disorder of glycosylation, type IIn	616721
SLC45A2	Albinism, oculocutaneous, type IV	606574
SLC46A1	Folate malabsorption, hereditary	229050
SLC4A1	Renal tubular acidosis, distal, AR	611590
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities	604278
SLC52A2	Brown-Vialetto-Van Laere syndrome 2	614707
SLC52A3	Brown-Vialetto-Van Laere syndrome 1	211530
SLC5A7	Myasthenic syndrome, congenital, 20, presynaptic	617143
SLC6A3	Parkinsonism-dystonia, infantile	613135
SLC6A5	Hyperekplexia 3	614618
SLC6A8	Cerebral creatine deficiency syndrome 1	300352
SLC7A7	Lysinuric protein intolerance	222700
SLC9A3	Diarrhea 8, secretory sodium, congenital	616868
SLC9A6	Intellectual developmental disorder, X-linked syndromic, Christianson type	300243

Gene Symbol	Disease	OMIM Number
SMARCAL1	Schimke immunoosseous dysplasia	242900
SMN1	Spinal muscular atrophy-1	253300
SMPD1	Niemann-Pick disease, type A	257200
SMS	Intellectual developmental disorder, X-linked, Snyder-Robinson type	309583
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	609528
SNORD118	Leukoencephalopathy, brain calcifications, and cysts	614561
SNX14	Spinocerebellar ataxia, autosomal recessive 20	616354
SOST	Sclerosteosis 1	269500
SP110	Hepatic venoocclusive disease with immunodeficiency	235550
SPAG1	Ciliary dyskinesia, primary, 28	615505
SPART	Troyer syndrome	275900
SPATA5	Epilepsy, hearing loss, and intellectual developmental disorder syndrome	616577
SPATA7	Leber congenital amaurosis 3	604232
SPG11	Spastic paraplegia 11, autosomal recessive	604360
SPINK5	Netherton syndrome	256500
SPINT2	Diarrhea 3, secretory sodium, congenital, syndromic	270420
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	612716
SQSTM1	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	617145
SRD5A3	Congenital disorder of glycosylation, type Iq	612379
SSR4	Congenital disorder of glycosylation, type Iy	300934
ST3GAL5	Salt and pepper developmental regression syndrome	609056
STAMBP	Microcephaly-capillary malformation syndrome	614261
STAR	Lipoid adrenal hyperplasia	201710
STAT1	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive	613796
STIL	Microcephaly 7, primary, autosomal recessive	612703
STIM1	Immunodeficiency 10	612783
STRA6	Microphthalmia, isolated, with coloboma 8	601186
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy	611087
STUB1	Spinocerebellar ataxia, autosomal recessive 16	615768
STX11	Hemophagocytic lymphohistiocytosis, familial, 4	603552
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5	613101
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	612073
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	245400
SUMF1	Multiple sulfatase deficiency	272200
SUOX	Sulfite oxidase deficiency	272300
SURF1	Leigh syndrome, due to COX deficiency	256000
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	300491
SYP	Intellectual developmental disorder, X-linked 96	300802
TALDO1	Transaldolase deficiency	606003
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	616878
TAP1	Bare lymphocyte syndrome, type I	604571
TAZ	Barth syndrome	302060
TBC1D23	Pontocerebellar hypoplasia, type 11	617695
TBC1D24	Epileptic encephalopathy, early infantile, 16	615338
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	617193
TBCE	Kenny-Caffey syndrome-1	244460
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	616900
TBX19	Adrenocorticotropic hormone deficiency	201400
TCAP	Muscular dystrophy, limb-girdle, type 2G	601954
TCIRG1	Osteopetrosis, autosomal recessive 1	259700
TCN2	Transcobalamin II deficiency	275350
TCTN2	Joubert syndrome 24	616654
TCTN3	Joubert syndrome 18	614815
TDRD7	Cataract 36	613887
TELO2	You-Hoover-Fong syndrome	616954

Gene Symbol	Disease	OMIM Number
TF	Atransferrinemia	209300
TGM1	Ichthyosis, congenital, autosomal recessive 1	242300
TH	Segawa syndrome, recessive	605407
THOC2	Intellectual developmental disorder, X-linked 12/35	300957
TIMM8A	Jensen syndrome	311150
TJP2	Cholestasis, progressive familial intrahepatic 4	615878
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type)	609560
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and intellectual developmental disorder syndrome	213980
TMEM107	Orofaciodigital syndrome XVI	617563
TMEM126A	Optic atrophy 7	612989
TMEM138	Joubert syndrome 16	614465
TMEM165	Congenital disorder of glycosylation, type IIk	614727
TMEM216	Joubert syndrome 2	608091
TMEM231	Joubert syndrome 20	614970
TMEM237	Joubert syndrome 14	614424
TMEM67	Joubert syndrome 6	610688
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	614052
TMTC3	Lissencephaly 8	617255
TNFRSF11A	Osteopetrosis, autosomal recessive 7	612301
TNFRSF11B	Paget disease of bone 5, juvenile-onset	239000
TNFRSF13B	Immunodeficiency, common variable, 2	240500
TNFSF11	Osteopetrosis, autosomal recessive 2	259710
TNNT1	Nemaline myopathy 5, Amish type	605355
TOE1	Pontocerebellar hypoplasia, type 7	614969
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency	615512
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	614458
TPM3	Nemaline myopathy 1, autosomal dominant or recessive	609284
TPP1	Ceroid lipofuscinosis, neuronal, 2	204500
TRAC	Immunodeficiency 7, TCR-alpha/beta deficient	615387
TRAPPC11	Muscular dystrophy, limb-girdle, type 2S	615356
TRAPPC9	Intellectual developmental disorder, autosomal recessive 13	613192
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness	615441
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive	225750
TRIM32	Muscular dystrophy, limb-girdle, type 2H	254110
TRIM37	Mulibrey nanism	253250
TRIP11	Achondrogenesis, type IA	200600
TRIT1	Combined oxidative phosphorylation deficiency 35	617873
TRMT10A	Microcephaly, short stature, and impaired glucose metabolism	616033
TRMU	Liver failure, transient infantile	613070
TRNT1	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay	616084
TRPM6	Hypomagnesemia 1, intestinal	602014
TSEN2	Pontocerebellar hypoplasia type 2B	612389
TSEN54	Pontocerebellar hypoplasia type 2A	277470
TSFM	Combined oxidative phosphorylation deficiency 3	610505
TSHB	Hypothryoidism, congenital, nongoitrous 4	275100
TSPAN7	Intellectual developmental disorder, X-linked 58	300210
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome	608800
TTC19	Mitochondrial complex III deficiency, nuclear type 2	615157
TTC21B	Short-rib thoracic dysplasia 4 with or without polydactyly	613819
TTC37	Trichohepatoenteric syndrome 1	222470
TTC7A	Gastrointestinal defects and immunodeficiency syndrome	243150
TTC8	Bardet-Biedl syndrome 8	615985
TTI2	Intellectual developmental disorder, autosomal recessive 39	615541
TTN	Myopathy, early-onset, with fatal cardiomyopathy	611705
TTPA	Ataxia with isolated vitamin E deficiency	277460
TUBA8	Polymicrogyria with optic nerve hypoplasia	613180
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, 3	616335

Gene Symbol	Disease	OMIM Number
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1	251270
TUFM	Combined oxidative phosphorylation deficiency 4	610678
TULP1	Retinitis pigmentosa 14	600132
TUSC3	Intellectual developmental disorder, autosomal recessive 7	611093
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	271245
TXNL4A	Burn-McKeown syndrome	608572
TYK2	Immunodeficiency 35	611521
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	603041
TYR	Albinism, oculocutaneous, type IA	203100
TYRP1	Albinism, oculocutaneous, type III	203290
UBA1	Spinal muscular atrophy, X-linked 2, infantile	301830
UBA5	Epileptic encephalopathy, early infantile, 44	617132
UBE2A	Intellectual developmental disorder, X-linked syndromic, Nascimento-type	300860
UBE2T	Fanconi anemia, complementation group T	616435
UBE3B	Kaufman oculocerebrofacial syndrome	244450
UBR1	Johanson-Blizzard syndrome	243800
UFM1	Leukodystrophy, hypomyelinating, 14	617899
UGT1A1	Crigler-Najjar syndrome, type I	218800
UMPS	Orotic aciduria	258900
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3	608898
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	616801
UPB1	Beta-ureidopropionase deficiency	613161
UPF3B	Intellectual developmental disorder, X-linked, syndromic 14	300676
UQCRC2	Mitochondrial complex III deficiency, nuclear type 5	615160
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4	615159
UROS	Porphyria, congenital erythropoietic	263700
USB1	Poikiloderma with neutropenia	604173
USH1C	Usher syndrome, type 1C	276904
USH1G	Usher syndrome, type 1G	606943
USH2A	Usher syndrome, type 2A	276901
USP9X	Intellectual developmental disorder, X-linked 99	300919
VARS1	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy	617802
VARS2	Combined oxidative phosphorylation deficiency 20	615917
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2	613404
VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, 2	607473
VLDLR	Cerebellar hypoplasia and intellectual developmental disorder with or without quadrupedal locomotion 1	224050
VMA21	Myopathy, X-linked, with excessive autophagy	310440
VPS11	Leukodystrophy, hypomyelinating, 12	616683
VPS13A	Choreoacanthocytosis	200150
VPS13B	Cohen syndrome	216550
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1	208085
VPS37A	Spastic paraplegia 53, autosomal recessive	614898
VPS45	Neutropenia, severe congenital, 5, autosomal recessive	615285
VPS53	Pontocerebellar hypoplasia, type 2E	615851
VRK1	Pontocerebellar hypoplasia type 1A	607596
VSX2	Microphthalmia with coloboma 3	610092
VWF	von Willebrand disease, types 2A, 2B, 2M, and 2N	613554
WARS2	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures	617710
WAS	Wiskott-Aldrich syndrome	301000
WDR19	Senior-Loken syndrome 8	616307
WDR34	Short-rib thoracic dysplasia 11 with or without polydactyly	615633
WDR35	Short-rib thoracic dysplasia 7 with or without polydactyly	614091
WDR45B	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures	617977
WDR60	Short-rib thoracic dysplasia 8 with or without polydactyly	615503
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations	604317

Gene Symbol	Disease	OMIM Number
WDR73	Galloway-Mowat syndrome	251300
WDR81	Cerebellar ataxia, intellectual developmental disorder, and disequilibrium syndrome 2	610185
WFS1	Wolfram syndrome	222300
WHRN	Usher syndrome, type 2D	611383
WNK1	Neuropathy, hereditary sensory and autonomic, type II	201300
WNT1	Osteogenesis imperfecta, type XV	615220
WNT10B	Split-hand/foot malformation 6	225300
WNT7A	Ulna and fibula, absence of, with severe limb deficiency	276820
WRAP53	Dyskeratosis congenita, autosomal recessive 3	613988
WRN	Werner syndrome	277700
WWOX	Epileptic encephalopathy, early infantile, 28	616211
XIAP	Lymphoproliferative syndrome, X-linked, 2	300635
XPA	Xeroderma pigmentosum, group A	278700
XPC	Xeroderma pigmentosum, group C	278720
XPNPEP3	Nephronophthisis-like nephropathy 1	613159
XRCC4	Short stature, microcephaly, and endocrine dysfunction	616541
XYLT1	Desbuquois dysplasia 2	615777
XYLT2	Spondyloocular syndrome	605822
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2	613561
ZAP70	Selective T-cell defect	269840
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome-2	614069
ZC4H2	Wieacker-Wolff syndrome	314580
ZDHHC9	Intellectual developmental disorder, X-linked syndromic, Raymond type	300799
ZFYVE26	Spastic paraplegia 15, autosomal recessive	270700
ZIC3	Congenital heart defects, nonsyndromic, 1, X-linked	306955
ZMPSTE24	Restrictive dermopathy, lethal	275210
ZMYND10	Ciliary dyskinesia, primary, 22	615444
ZNF335	Microcephaly 10, primary, autosomal recessive	615095
ZNF469	Brittle cornea syndrome 1	229200
ZNF711	Intellectual developmental disorder, X-linked 97	300803
ZNHIT3	PEHO syndrome	260565