MVL Vision Panel v21.1 Information

The Vision Panel from Molecular Vision Laboratory is designed to analyze genes linked to inherited eye diseases. These genes have been curated based on the most up-to-date clinical research and literature, ensuring a comprehensive and reliable testing panel. The panel is periodically updated to include clinical and literature advancements, making it the most comprehensive genetic testing panel for inherited eye diseases.

Inherited eye disorders are genetically heterogenous, and many similar phenotypes can have very different causes. Testing with MVL Vision Panel provides an efficient way to evaluate multiple genes when clinical symptoms alone cannot pinpoint a diagnosis. By identifying genetic variants, MVL Vision Panel can confirm clinical diagnoses, guide genetic counseling, and assist in developing personalized care plans.

Turnaround time

The typical turnaround time for the Vision Panel is 5-8 weeks. However, we understand that some cases require expedited results. For urgent cases, testing can be completed in as little as 10 days. On average, results are delivered in approximately 5 weeks.

Gene-specific testing

MVL offers unparalleled flexibility in testing. While the full Vision Panel is always available, clients may request analysis and reporting on any combination of genes within the panel. For instance, if only variants in 10 selected genes are of interest, we can focus the report and analysis accordingly.

Parental testing

To support comprehensive analysis, MVL provides complimentary parental testing for patients undergoing Vision Panel testing. We recommend submitting samples from both biological parents at the same time. Parental testing allows for segregation analysis, particularly in cases where a heterozygous pathogenic variant is identified alongside a heterozygous variant of uncertain significance. Determining the phasing of these variants can clarify the genetic diagnosis, which improves diagnostic confidence.

Clinical features

Because of the genetic heterogeneity associated with inherited eye diseases, it is difficult to solely rely on clinical phenotype alone for an accurate diagnosis. Genetic testing plays a vital role in differentiating between cases with similar clinical presentations but distinct genetic causes.

Our Vision Panel stands out compared to whole exome sequencing (WES) and whole genome sequencing (WGS) due to its superior read depth, which enhances the accuracy of variant detection and interpretation, particularly in the case of copy number variant (CNV) detection, where read depth is critical. With a coverage of 1,099 genes associated with inherited eye diseases MVL Vision Panel is the most comprehensive inherited eye disease panel available. The extensive coverage includes genes with established OMIM phenotypes, candidate genes without a currently defined OMIM phenotype, and genes involved in genome-wide association studies (GWAS) related to inherited eye diseases.

Also, please note that the panel includes the mitochondria genome and RPGR exon 15 (RPGR ORF15).

Test Methods

Testing is performed by targeted next-generation sequencing by hybridization capture. Alignment and variant calling are performed using GRCh37, and identified pathogenic variants are confirmed by Sanger. Targeted regions cover all exons, exon-intron boundaries, and relevant deep-intronic regions.

Limitations

Only coding regions, immediately flanking intron sequences, and select designated deep intronic regions are examined. Changes in the promoter region, most deep intronic regions, or other non-coding regions of the gene would not be detected. Multiple exon deletions, multiple exon insertions, and complete deletion of one allele may not be identified using these methods.

List of genes tested

Gene	Phenotype
AARS2	Combined oxidative phosphorylation deficiency 8, Leukoencephalopathy, progressive, with ovarian failure
AASS	Hyperlysinemia, Saccharopinuria
ABAT	GABA-transaminase deficiency
ABCA1	HDL deficiency, familial, 1, Tangier disease
ABCA4	Cone-rod dystrophy 3, Fundus flavimaculatus, Retinal dystrophy, early-onset severe, Retinitis pigmentosa 19, Stargardt disease 1
ABCB6	Dyschromatosis universalis hereditaria 3, Microphthalmia, isolated, with coloboma 7, Pseudohyperkalemia, familial, 2, due to red cell leak
ABCB7	Anemia, sideroblastic, with ataxia
ABCC6	Arterial calcification, generalized, of infancy, 2, Pseudoxanthoma elasticum, Pseudoxanthoma elasticum, forme fruste
ABCD1	Adrenoleukodystrophy, Adrenomyeloneuropathy, adult
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract
ACACA	Acetyl-CoA carboxylase deficiency
ACAD9	Mitochondrial complex I deficiency, nuclear type 20

ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of
ACADVL	VLCAD deficiency
ACAT1	Alpha-methylacetoacetic aciduria
ACBD5	Retinal dystrophy with leukodystrophy
ACO2	Optic atrophy 9, Infantile cerebellar-retinal degeneration
ADAM9	Cone-rod dystrophy 9
ADAMTS10	Weill-Marchesani syndrome 1, recessive
ADAMTS17	Weill-Marchesani 4 syndrome, recessive
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type
ADAMTSL4	Ectopia lentis et pupillae, Ectopia lentis, isolated, autosomal recessive
ADGRA3	Recessive Retinitis Pigmentosa from RETNET
ADGRV1	Usher syndrome, type 2C, Usher syndrome, type 2C, GPR98/PDZD7 digenic
ADIPOR1	Retinitis Pigmentosa from RETNET
AFG3L2	Optic atrophy 12, Spastic ataxia 5, autosomal recessive, Spinocerebellar ataxia 28
AGBL5	Retinitis pigmentosa 75
AGK	Cataract 38, autosomal recessive, Sengers syndrome
AGPS	Rhizomelic chondrodysplasia punctata, type 3
AGRN	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects
AHI1	Joubert syndrome 3
AHR	Retinitis pigmentosa 85
AIFM1	Combined oxidative phosphorylation deficiency 6, Cowchock syndrome, Deafness, X-linked 5
AIPL1	Cone-rod dystrophy, Leber congenital amaurosis 4, Retinitis pigmentosa, juvenile
ALAS2	Anemia, sideroblastic, 1, Protoporphyria, erythropoietic, X-linked
ALDH18A1	Cutis laxa, autosomal dominant 3, Cutis laxa, autosomal recessive, type IIIA, Spastic paraplegia 9A, autosomal dominant, Spastic paraplegia 9B, autosomal recessive
ALDH1A3	Microphthalmia, isolated 8
ALDH3A2	Sjogren-Larsson syndrome
ALG1	Congenital disorder of glycosylation, type Ik
ALG14	Myasthenic syndrome, congenital, 15, without tubular aggregates
ALG2	Congenital disorder of glycosylation, type Ii, Myasthenic syndrome, congenital, 14, with tubular aggregates
ALMS1	Alstrom syndrome
ALPK1	ROSAH syndrome
AMACR	Alpha-methylacyl-CoA racemase deficiency, Bile acid synthesis defect, congenital, 4
ANKS6	Nephronophthisis 16
ANO10	Spinocerebellar ataxia, autosomal recessive 10
ANTXR1	GAPO syndrome
AP3B1	Hermansky-Pudlak syndrome 2
AP3D1	Hermansky-Pudlak syndrome 10
AP5Z1	Spastic paraplegia 48, autosomal recessive
APC	Pigmented ocular fundus lesions (POFLS) - for mutations in exons 10 to the proximal portion of exon I5
APOB	Hypobetalipoproteinemia
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia
ARHGEF18	Retinitis pigmentosa 78
ARID1A	Coffin-Siris syndrome 2

ARL13B	Joubert syndrome 8
ARL2	Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1
ARL2BP	Retinitis pigmentosa with or without situs inversus
ARL3	Joubert syndrome 35, Retinitis pigmentosa 83
ARL6	Retinitis pigmentosa 55, Bardet-Biedl syndrome 3
ARMC9	Joubert syndrome 30
ARMS2	Macular degeneration, age-related, 8
ARSG	Usher syndrome, type IV
ASB10	Glaucoma 1, open angle, F
ASPH	Traboulsi syndrome
ASRGL1	Retinal degeneration from RETNET
ATAD3A	Harel-Yoon syndrome
ATF6	Achromatopsia 7
ATOH1	Deafness, autosomal dominant 89
ATOH7	Persistent hyperplastic primary vitreous, autosomal recessive
ATP13A2	Kufor-Rakeb syndrome, Spastic paraplegia 78, autosomal recessive
ATP1A3	Alternating hemiplegia of childhood 2, CAPOS syndrome, Dystonia-12
ATP5F1A	Combined oxidative phosphorylation deficiency 22, Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4
ATP5F1D	Mitochondrial complex V (ATP synthase) deficiency
ATP5F1E	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3
ATP7B	Wilson disease
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1
AUH	3-methylglutaconic aciduria, type I
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11
B3GALT6	Ehlers-Danlos syndrome, spondylodysplastic type, 2, Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures
B3GLCT	Peters-plus syndrome
B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic type, 1
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13
B9D1	Meckel syndrome 9, Joubert syndrome 27
B9D2	Meckel syndrome 10, Joubert syndrome 34
BBIP1 (BBS18)	Bardet-Biedl syndrome 18
BBS1	Bardet-Biedl syndrome 1
BBS10	Bardet-Biedl syndrome 10
BBS12	Bardet-Biedl syndrome 12
BBS2	Bardet-Biedl syndrome 2, Retinitis pigmentosa 74
BBS4	Bardet-Biedl syndrome 4
BBS5	Bardet-Biedl syndrome 5
BBS7	Bardet-Biedl syndrome 7
BBS9	Bardet-Biedl syndrome 9
BCKDHA	Maple syrup urine disease, type Ia
BCKDHB	Maple syrup urine disease, type Ib
BCO1	Hypercarotenemia and vitamin A deficiency, autosomal dominant
BCOR	Microphthalmia, syndromic 2
BCS1L	Bjornstad syndrome, GRACILE syndrome, Leigh syndrome, Mitochondrial complex III deficiency, nuclear type 1

BEST1	Bestrophinopathy, autosomal recessive, Macular dystrophy, vitelliform, 2, Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, Retinitis pigmentosa, concentric, Retinitis pigmentosa-50, Vitreoretinochoroidopathy
BFSP1	Cataract 33, multiple types
BFSP2	Cataract 12, multiple types
BLOC1S3	Hermansky-Pudlak syndrome 8
BLOC1S5	Hermansky-Pudlak syndrome 11
BLOC1S6	Hermansky-pudlak syndrome 9
BMP4	Microphthalmia, syndromic 6, Orofacial cleft 11
BMP7	Developmental eye disorder
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia
BTD	Biotinidase deficiency
BUB1B	Cancer-prone syndrome of premature chromatid separation (PCS syndrome)
C12ORF57	Temtamy syndrome
C12ORF65	MTRFR; Combined oxidative phosphorylation deficiency; Spastic paraplegia 55, autosomal recessive
C19ORF12	Neurodegeneration with brain iron accumulation; Spastic paraplegia 43, autosomal recessive
C1QBP	Combined oxidative phosphorylation deficiency 33
C1QTNF5	Retinal degeneration, late-onset, autosomal dominant
C1R	Ehlers-Danlos syndrome, periodontal type, 1
C2CD3	Orofaciodigital syndrome XIV
C8A	C8 deficiency, type I
C8B	C8 deficiency, type II
C8ORF37	Bardet-Biedl syndrome 21; Cone-rod dystrophy 16; Retinitis pigmentosa 64
CA4	Retinitis pigmentosa 17
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency
CABP4	Cone-rod synaptic disorder, congenital nonprogressive
CACNA1A	Epileptic encephalopathy, early infantile, 42, Episodic ataxia, type 2, Migraine, familial hemiplegic, 1, Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, Spinocerebellar ataxia 6
CACNA1F	Aland Island eye disease, Cone-rod dystrophy, X-linked, 3, Night blindness, congenital stationary (incomplete), 2A, X-linked
CACNA2D4	Retinal cone dystrophy 4
CAPN15	Oculogastrointestinal neurodevelopmental syndrome
CAPN5	Vitreoretinopathy, neovascular inflammatory
CARS2	Combined oxidative phosphorylation deficiency 27
CASK	FG syndrome 4, Mental retardation and microcephaly with pontine and cerebellar hypoplasia, Mental retardation, with or without nystagmus
CBS	Homocystinuria, B6-responsive and nonresponsive types, Thrombosis, hyperhomocysteinemic
CC2D2A	COACH syndrome, Joubert syndrome 9, Meckel syndrome 6
CCDC103	Ciliary dyskinesia, primary, 17
CCDC114	Ciliary dyskinesia, primary, 20
CCDC28B	Bardet-Biedl syndrome 1, modifier of
CCDC39	Ciliary dyskinesia, primary, 14
CCDC40	Ciliary dyskinesia, primary, 15
CCND1	von Hippel-Lindau syndrome, modifier of
CCT2	Leber Congenital Amaurosis

CDH23	Deafness, autosomal recessive 12, Usher syndrome, type 1D, Usher syndrome, type 1D/F digenic
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, Hypotrichosis, congenital, with juvenile macular dystrophy
CDHR1	Cone-rod dystrophy 15, Retinitis pigmentosa 65
CDK5RAP2	Microcephaly 3, primary, autosomal recessive
CEP104	Joubert syndrome 25
CEP120	Joubert syndrome 31, Short-rib thoracic dysplasia 13 with or without polydactyly
CEP164	Nephronophthisis 15
CEP19	Bardet-Biedl syndrome
CEP250	Cone-rod dystrophy and hearing loss 2
CEP290	Bardet-Biedl syndrome 14, Joubert syndrome 5, Leber congenital amaurosis 10, Meckel syndrome 4, Senior-Loken syndrome 6
CEP41	Joubert syndrome 15
CEP78	Cone-rod dystrophy and hearing loss
CEP83	Nephronophthisis 18
CERKL	Retinitis pigmentosa 26
CFAP410	Retinal dystrophy with macular staphyloma, Spondylometaphyseal dysplasia, axial
CFH	Basal laminar drusen, Complement factor H deficiency, Hemolytic uremic syndrome, atypical, susceptibility to, 1, Macular degeneration, age-related, 4
CHAT	Myasthenic syndrome, congenital, 6, presynaptic
CHCHD10	Myopathy, isolated mitochondrial, autosomal dominant, Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, Spinal muscular atrophy, Jokela type
CHD7	CHARGE syndrome, Hypogonadotropic hypogonadism 5 with or without anosmia
CHD8	Intellectual developmental disorder with autism and macrocephaly
CHKB	Muscular dystrophy, congenital, megaconial type
CHM	Choroideremia
CHMP4B	Cataract 31, multiple types
CHN1	Duane retraction syndrome 2
CHRNA1	Multiple pterygium syndrome, lethal type, Myasthenic syndrome, congenital, 1A, slow-channel, Myasthenic syndrome, congenital, 1B, fast-channel
CHRNB1	Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, Myasthenic syndrome, congenital, 2A, slow-channel
CHRND	Myasthenic syndrome, congenital, 3A, slow-channel, Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, Multiple pterygium syndrome, lethal type, Myasthenic syndrome, congenital, 3B, fast-channel
CHRNE	Myasthenic syndrome, congenital, 4A, slow-channel, Myasthenic syndrome, congenital, 4B, fast-channel, Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1
CHST6	Macular corneal dystrophy
CIB2	Deafness, autosomal recessive 48, Usher syndrome, type IJ
CISD2	Wolfram syndrome 2
CLCC1	Retinitis pigmentosa 32
CLDN19	Hypomagnesemia 5, renal, with ocular involvement
CLEC3B	Macular dystrophy, retinal, 4
CLN3	Ceroid lipofuscinosis, neuronal, 3
CLN5	Ceroid lipofuscinosis, neuronal, 5
CLN6	Ceroid lipofuscinosis, neuronal, 6, Ceroid lipofuscinosis, neuronal, Kufs type, adult onset

CLN8	Ceroid lipofuscinosis, neuronal, 8, Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia
CLPP	Perrault syndrome 3
CLRN1	Retinitis pigmentosa 61, Usher syndrome, type 3A
CLUAP1	Leber congenital amaurosis
CNGA1	Retinitis pigmentosa 49
CNGA3	Achromatopsia 2
CNGB1	Retinitis pigmentosa 45
CNGB3	Achromatopsia 3, Macular degeneration, juvenile
CNNM4	Jalili syndrome
COA3	Mitochondrial complex IV deficiency, nuclear type 14
COA5	Mitochondrial complex IV, deficiency, nuclear type 9
COA6	Mitochondrial complex IV deficiency, nuclear type 13
COA7	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3
COA8	Mitochondrial complex IV deficiency
COG4	Congenital disorder of glycosylation, type IIj, Saul-Wilson syndrome
COG6	COD2; Congenital disorder of glycosylation, typeIII; Shaheen syndrome
COL11A1	Deafness, autosomal dominant 37, Fibrochondrogenesis 1, Marshall syndrome, Stickler syndrome, type II
COL11A2	Deafness, autosomal dominant 13, Deafness, autosomal recessive 53, Fibrochondrogenesis 2, Otospondylomegaepiphyseal dysplasia, autosomal dominant, Otospondylomegaepiphyseal dysplasia, autosomal recessive
COL12A1	Ullrich congenital muscular dystrophy 2, Bethlem myopathy 2
COL13A1	Myasthenic syndrome, congenital, 19
COL18A1	Glaucoma, primary closed-angle; Knobloch syndrome, type 1
COL1A1	Caffey disease, Ehlers-Danlos syndrome, arthrochalasia type, 1, Osteogenesis imperfecta, type I, Osteogenesis imperfecta, type II, Osteogenesis imperfecta, type III, Osteogenesis imperfecta, type IV, Bone mineral density variation QTL, osteoporosis
COL1A2	Ehlers-Danlos syndrome, arthrochalasia type, 2, Ehlers-Danlos syndrome, cardiac valvular type, Osteogenesis imperfecta, type II, Osteogenesis imperfecta, type III, Osteogenesis imperfecta, type IV, Osteoporosis, postmenopausal
COL2A1	Achondrogenesis, type II or hypochondrogenesis, Avascular necrosis of the femoral head, Czech dysplasia, Epiphyseal dysplasia, multiple, with myopia and deafness, Kniest dysplasia, Legg-Calve-Perthes disease, Osteoarthritis with mild chondrodysplasia, Platyspondylic skeletal dysplasia, Torrance type, SED congenita, SMED Strudwick type, Spondyloepiphyseal dysplasia, Stanescu type, Spondyloperipheral dysplasia, Stickler sydrome, type I, nonsyndromic ocular, Stickler syndrome, type I, Vitreoretinopathy with phalangeal epiphyseal dysplasia
COL3A1	Ehlers-Danlos syndrome, vascular type; Polymicrogyria with or without vascular-type EDS
COL4A1	Retinal arteries, tortuosity of, Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, Brain small vessel disease with or without ocular anomalies, Microangiopathy and leukoencephalopathy, pontine, autosomal dominant
COL4A3	Alport syndrome 2, autosomal recessive, Alport syndrome 3, autosomal dominant, Hematuria, benign familial
COL4A4	Alport syndrome 2, autosomal recessive, Hematuria, familial benign
COL4A5	Alport syndrome 1, X-linked
COL5A1	Ehlers-Danlos syndrome, classic type, 1; Fibromuscular dysplasia, multifocal
COL5A2	Ehlers-Danlos syndrome, classic type, 2

COL9A1	Epiphyseal dysplasia, multiple, 6, Stickler syndrome, type IV
COL9A2	Stickler syndrome, type V, Epiphyseal dysplasia, multiple, 2
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy
COLQ	Myasthenic syndrome, congenital, 5
COQ2	Coenzyme Q10 deficiency, primary, 1
COQ4	Coenzyme Q10 deficiency, primary, 7
COQ5	Coenzyme Q10 deficiency, primary, 9
COQ6	Coenzyme Q10 deficiency, primary, 6
COQ7	Coenzyme Q10 deficiency, primary, 8
COQ8A	Coenzyme Q10 deficiency, primary, 4
COQ8B	Nephrotic syndrome, type 9
COQ9	Coenzyme Q10 deficiency, primary, 5
COX10	Mitochondrial complex IV deficiency, nuclear type 3
COX11	Mitochondrial complex IV deficiency, nuclear type 23
COX14	Mitochondrial complex IV deficiency, nuclear type 10
COX15	Mitochondrial complex IV deficiency, nuclear type 6
COX16	Mitochondrial complex IV deficiency, nuclear type 22
COX18	Complex IV deficiency
COX20	Mitochondrial complex IV deficiency, nuclear type 11
COX4I1	Mitochondrial complex IV deficiency, nuclear type 16
COX4I2	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis
COX6A1	Charcot-Marie-Tooth disease, recessive intermediate D
COX6A2	Mitochondrial complex IV deficiency, nuclear type 18
COX6B1	Mitochondrial complex IV deficiency, nuclear type 7
COX7B	Linear skin defects with multiple congenital anomalies 2
COX8A	Mitochondrial complex IV deficiency, nuclear type 15
CPAMD8	Anterior segment dysgenesis 8
CPLANE1	Joubert syndrome 17, Orofaciodigital syndrome VI
CPT1A	CPT deficiency, hepatic, type IA
CPT2	CPT II deficiency, infantile, CPT II deficiency, lethal neonatal, CPT II deficiency, myopathic, stress-induced
CRB1	Leber congenital amaurosis 8, Pigmented paravenous chorioretinal atrophy, Retinitis pigmentosa-12
CRELD1	Jeffries-Lakhani neurodevelopmental syndrome
CRPPA	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7
CRX	Cone-rod retinal dystrophy-2, Leber congenital amaurosis 7
CRYAA	Cataract 9, multiple types
CRYAB	Cardiomyopathy, dilated, 1II, Cataract 16, multiple types, Myopathy, myofibrillar, 2, Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related
CRYBA1	Cataract 10, multiple types
CRYBA2	Cataract 42
CRYBA4	Cataract 23
CRYBB1	Cataract 17, multiple types
CRYBB2	Cataract 3, multiple types
CRYBB3	Cataract 22
CRYGB	Cataract 39, multiple types, autosomal dominant
CRYGC	Cataract 2, multiple types
CRYGD	Cataract 4, multiple types

CRYGS	Cataract 20, multiple types
CSPP1	Joubert syndrome 21
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy
CTNNA1	Macular dystrophy, patterned, 2
CTNNB1	Exudative vitreoretinopathy 7, Neurodevelopmental disorder with spastic diplegia and visual defects
CTNND1	Blepharocheilodontic syndrome 2
CTSD	Ceroid lipofuscinosis, neuronal, 10
CTSF	Ceroid lipofuscinosis, neuronal, 13, Kufs type
CWC27	Retinitis pigmentosa with or without skeletal anomalies
CYC1	Mitochondrial complex III deficiency, nuclear type 6
CYCS	Thrombocytopenia 4
CYP1B1	Anterior segment dysgenesis 6, multiple subtypes, Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset
CYP27A1	Cerebrotendinous xanthomatosis
CYP4V2	Bietti crystalline corneoretinal dystrophy
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation
DBT	Maple syrup urine disease, type II
DCC	Gaze palsy, familial horizontal, with progressive scoliosis, 2, Mirror movements 1 and/or agenesis of the corpus callosum
DCN	Corneal dystrophy, congenital stromal
DCT	Oculocutaneous albinism, type VIII
DDX58	Singleton-Merten syndrome 2
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), Portal hypertension, noncirrhotic, Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Congenital disorder of glycosylation, type 1bb, Developmental delay and seizures with or without movement abnormalities, Retinitis pigmentosa 59
DHX38	Retinitis pigmentosa 84
DLAT	Pyruvate dehydrogenase E2 deficiency
DLD	Dihydrolipoamide dehydrogenase deficiency
DMD	Becker muscular dystrophy, Cardiomyopathy, dilated, 3B, Duchenne muscular dystrophy
DNA2	Seckel syndrome 8, Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6
DNAAF1	Ciliary dyskinesia, primary, 13
DNAAF2	Ciliary dyskinesia, primary, 10
DNAAF3	Ciliary dyskinesia, primary, 2
DNAAF5	Ciliary dyskinesia, primary, 18
DNAH11	Ciliary dyskinesia, primary, 7, with or without situs inversus
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus
DNAJC19	3-methylglutaconic aciduria, type V
DNAJC30	Leber-like hereditary optic neuropathy, autosomal recessive 1
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type
DNAL1	Ciliary dyskinesia, primary, 16

DNM1L	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, Optic atrophy 5
DNM2	Centronuclear myopathy 1, Charcot-Marie-Tooth disease, axonal type 2M, Charcot-Marie-Tooth disease, dominant intermediate B, Lethal congenital contracture syndrome 5
DOCK6	Adams-Oliver syndrome 2
DOK7	Fetal akinesia deformation sequence 3, Myasthenic syndrome, congenital, 10
DPAGT1	Congenital disorder of glycosylation, type Ij, Myasthenic syndrome, congenital, 13, with tubular aggregates
DRAM2	Cone-rod dystrophy 21
DSE	Ehlers-Danlos syndrome, musculocontractural type 2
DTHD1	Leber congenital amaurosis with myopathy
DTNBP1 (BLOC1S8)	Hermansky-Pudlak syndrome 7
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly
EARS2	Combined oxidative phosphorylation deficiency 12
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency
EDN3	Central hypoventilation syndrome, congenital, Waardenburg syndrome, type 4B, Hirschsprung disease, susceptibility to, 4
EDNRB	ABCD syndrome, Waardenburg syndrome, type 4A, Hirschsprung disease, susceptibility to, 2
EFEMP1	Doyne honeycomb degeneration of retina
ELAC2	Combined oxidative phosphorylation deficiency 17
ELOVL1	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, Spinocerebellar ataxia 34, Stargardt disease 3
ELP1	Dysautonomia, familial
ELP4	Aniridia 2
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation
EPHA2	Cataract 6, multiple types
ERAL1	Perrault syndrome 6
ERCC6	Cerebrooculofacioskeletal syndrome 1, Cockayne syndrome, type B, De Sanctis-Cacchione syndrome, Premature ovarian failure 11, UV-sensitive syndrome 1
ERCC8	Cockayne syndrome, type A, UV-sensitive syndrome 2
ESCO2	Roberts syndrome, SC phocomelia syndrome
ESPN	Usher syndrome, type 1M, Deafness, autosomal recessive 36, Deafness, neurosensory, without vestibular involvement, autosomal dominant
ETFA	Glutaric acidemia IIA
ETFB	Glutaric acidemia IIB
ETFDH	Glutaric acidemia IIC
ETHE1	Ethylmalonic encephalopathy
EVC	Weyers acrofacial dysostosis, Ellis-van Creveld syndrome
EVC2	Ellis-van Creveld syndrome, Weyers acrofacial dysostosis
EXOSC2	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies
EYA1	Otofaciocervical syndrome, Anterior segment anomalies with or without cataract, Branchiootic syndrome 1, Branchiootorenal syndrome 1, with or without cataracts
EYS	Retinitis pigmentosa 25
FA2H	Spastic paraplegia 35, autosomal recessive
FAM161A	Retinitis pigmentosa 28
FARS2	Combined oxidative phosphorylation deficiency 14, Spastic paraplegia 77, autosomal recessive

FASTKD2	Combined oxidative phosphorylation deficiency 44
FBLN5	Cutis laxa, autosomal dominant 2, Cutis laxa, autosomal recessive, type IA, Macular degeneration, age-related, 3, Neuropathy, hereditary, with or without age-related macular degeneration
FBN1	Acromicric dysplasia, Ectopia lentis, familial, Geleophysic dysplasia 2, Marfan lipodystrophy syndrome, Marfan syndrome, MASS syndrome, Stiff skin syndrome, Weill-Marchesani syndrome 2, dominant
FBP1	Fructose-1,6-bisphosphatase deficiency
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)
FDFT1	Squalene synthase deficiency
FDX2	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy
FDXR	Auditory neuropathy and optic atrophy
FH	Fumarase deficiency, Leiomyomatosis and renal cell cancer
FKBP14	Ehlers-Danlos syndrome, kyphoscoliotic type, 2
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5
FKTN	Cardiomyopathy, dilated, 1X, Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4
FLAD1	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency
FLNB	Atelosteogenesis, type I, Atelosteogenesis, type III, Boomerang dysplasia, Larsen syndrome, Spondylocarpotarsal synostosis syndrome
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa
FOXC1	Anterior segment dysgenesis 3, multiple subtypes, Axenfeld-Rieger syndrome, type 3
FOXC2	Lymphedema-distichiasis syndrome, Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus
FOXE3	Anterior segment dysgenesis 2, multiple subtypes, Cataract 34, multiple types
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19
FRAS1	Fraser syndrome 1
FREM2	Cryptophthalmos, unilateral or bilateral, isolated, Fraser syndrome 2
FRMD7	Nystagmus 1, congenital, X-linked, Nystagmus, infantile periodic alternating, X-linked
FSCN2	Retinitis pigmentosa 30
FTL	Hyperferritinemia-cataract syndrome, L-ferritin deficiency, dominant and recessive, Neurodegeneration with brain iron accumulation 3
FXN	Friedreich ataxia, Friedreich ataxia with retained reflexes
FYCO1	Cataract 18, autosomal recessive
FZD4	Exudative vitreoretinopathy 1, Retinopathy of prematurity
FZD5	Microphthalmia/coloboma 11
G6PC	Glycogen storage disease Ia
GAA	Glycogen storage disease II
GALE	Galactose epimerase deficiency
GALK1	Galactokinase deficiency with cataracts
GALM	Galactosemia IV
GALT	Galactosemia
GAMT	Cerebral creatine deficiency syndrome 2
GAN	Giant axonal neuropathy-1
GARS1	Charcot-Marie-Tooth disease, type 2D, Neuropathy, distal hereditary motor, type VA
GATB	Combined oxidative phosphorylation deficiency 41

GATC	Combined oxidative phosphorylation deficiency 42
GATM	Cerebral creatine deficiency syndrome 3; Fanconi renotubular syndrome 1
GCDH	Glutaricaciduria, type I
GCNT2	Adult i phenotype without cataract, Cataract 13 with adult i phenotype
GDAP1	Charcot-Marie-Tooth disease, axonal, type 2K, Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, Charcot-Marie-Tooth disease, recessive intermediate, A, Charcot-Marie-Tooth disease, type 4A
GDF1	Congenital heart defects, multiple types, 6, Right atrial isomerism (Ivemark)
GDF3	Klippel-Feil syndrome 3, autosomal dominant, Microphthalmia with coloboma 6, Microphthalmia, isolated 7
GDF6	Klippel-Feil syndrome 1, autosomal dominant, Leber congenital amaurosis 17, Microphthalmia with coloboma 6, digenic, Microphthalmia, isolated 4, Multiple synostoses syndrome 4
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay
GFM1	Combined oxidative phosphorylation deficiency 1
GFM2	Combined oxidative phosphorylation deficiency 39
GFPT1	Myasthenia, congenital, 12, with tubular aggregates
GJA1	Atrioventricular septal defect 3, Craniometaphyseal dysplasia, autosomal recessive, Erythrokeratodermia variabilis et progressiva 3, Hypoplastic left heart syndrome 1, Oculodentodigital dysplasia, Oculodentodigital dysplasia, autosomal recessive, Palmoplantar keratoderma with congenital alopecia, Syndactyly, type III
GJA3	Cataract 14, multiple types
GJA8	Cataract 1, multiple types
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1
GJB2	Bart-Pumphrey syndrome, Deafness, autosomal dominant 3A, Deafness, autosomal recessive 1A, Hystrix-like ichthyosis with deafness, Keratitis-ichthyosis-deafness syndrome, Keratoderma, palmoplantar, with deafness, Vohwinkel syndrome
GJB6	Deafness, autosomal dominant 3B, Deafness, autosomal recessive 1B, Deafness, digenic GJB2/GJB6, Ectodermal dysplasia 2, Clouston type
GJC3	Deafness
GLIS2	Nephronophthisis 7
GLRX5	Anemia, sideroblastic, 3, pyridoxine-refractory, Spasticity, childhood-onset, with hyperglycinemia
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14
GNAT1	Night blindness, congenital stationary, autosomal dominant 3, Night blindness, congenital stationary, type 1G
GNAT2	Achromatopsia 4
GNB3	Night blindness, congenital stationary, type 1H
GNPAT	Rhizomelic chondrodysplasia punctata, type 2
GNPTG	Mucolipidosis III gamma
GNS	Mucopolysaccharidosis type IIID
GPR143	Nystagmus 6, congenital, X-linked, Ocular albinism, type I, Nettleship-Falls type
GPR179	Night blindness, congenital stationary (complete), 1E, autosomal recessive
GRIP1	Fraser syndrome 3
GRK1	Oguchi disease-2
GRM6	Night blindness, congenital stationary (complete), 1B, autosomal recessive
GRN	Aphasia, primary progressive, Ceroid lipofuscinosis, neuronal, 11, Frontotemporal lobar degeneration with ubiquitin-positive inclusions

GTPBP3	Combined oxidative phosphorylation deficiency 23
GUCA1A	Cone dystrophy-3, Cone-rod dystrophy 14
GUCA1B	Retinitis pigmentosa 48
GUCA1C	Glkaucoma
GUCY2D	Choroidal dystrophy, central areolar 1, Cone-rod dystrophy 6, Leber congenital amaurosis 1, Night blindness, congenital stationary, type 1I
GYS2	Glycogen storage disease 0, liver
GZF1	Joint laxity, short stature, and myopia
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency, Hyperinsulinemic hypoglycemia, familial, 4
HADHA	Fatty liver, acute, of pregnancy, HELLP syndrome, maternal, of pregnancy, LCHAD deficiency, Trifunctional protein deficiency; Mitochondrial trifunctional protein deficiency 1
HADHB	Mitochondrial trifunctional protein deficiency
HARS1	Charcot-Marie-Tooth disease, axonal, type 2W, Usher syndrome type 3B
HARS2	Perrault syndrome 2
HCCS	Linear skin defects with multiple congenital anomalies 1
HCN1	Epileptic encephalopathy, early infantile, 24, Generalized epilepsy with febrile seizures plus, type 10
HESX1	Growth hormone deficiency with pituitary anomalies, Pituitary hormone deficiency, combined, 5, Septooptic dysplasia
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), Retinitis pigmentosa 73
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency
HK1	Hemolytic anemia due to hexokinase deficiency, Neurodevelopmental disorder with visual defects and brain anomalies, Neuropathy, hereditary motor and sensory, Russe type, Retinitis pigmentosa 79
HKDC1	Retinitis pigmentosa 92
HLCS	Holocarboxylase synthetase deficiency
HMCN1	Macular degeneration, age-related, 1
HMGB3	Microphthalmia, syndromic 13
HMGCL	HMG-CoA lyase deficiency
HMGCS2	HMG-CoA synthase-2 deficiency
HMX1	Oculoauricular syndrome
HPS1	Hermansky-Pudlak syndrome 1
HPS3	Hermansky-Pudlak syndrome 3
HPS4	Hermansky-Pudlak syndrome 4
HPS5	Hermansky-Pudlak syndrome 5
HPS6	Hermansky-Pudlak syndrome 6
HSD17B10	HSD10 mitochondrial disease
HSF4	Cataract 5, multiple types
HSPD1	Leukodystrophy, hypomyelinating, 4, Spastic paraplegia 13, autosomal dominant
HTRA1	CARASIL syndrome, Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, Macular degeneration, age-related, 7, Macular degeneration, age-related, neovascular type
HTRA2	3-methylglutaconic aciduria, type VIII, Parkinson disease 13
HYLS1	Hydrolethalus syndrome
IARS1	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy
IARS2	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia
IBA57	Spastic paraplegia 74, autosomal recessive, Multiple mitochondrial dysfunctions syndrome 3

IDH3A	Retinitis pigmentosa 90
IDH3B	Retinitis pigmentosa 46
IDUA	Mucopolysaccharidosis Ih, Mucopolysaccharidosis Ih/s, Mucopolysaccharidosis Is
IFT122	Cranioectodermal dysplasia 1
IFT140	Retinitis pigmentosa 80, Short-rib thoracic dysplasia 9 with or without polydactyly
IFT172	Retinitis pigmentosa 71, Short-rib thoracic dysplasia 10 with or without polydactyly
IFT27	Bardet-Biedl syndrome 19
IFT43	Cranioectodermal dysplasia 3, Retinitis pigmentosa 81, Short-rib thoracic dysplasia 18 with polydactyly
IFT52	Short-rib thoracic dysplasia 16 with or without polydactyly
IFT54	Senior-Loken syndrome 9
IFT74	Bardet-Biedl syndrome 20, Joubert syndrome 40, Spermatogenic failure 58
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly
IFT81	Short-rib thoracic dysplasia 19 with or without polydactyly
IMPDH1	Leber congenital amaurosis 11, Retinitis pigmentosa 10
IMPG1	Macular dystrophy, vitelliform, 4, Retinitis pigmentosa 91
IMPG2	Macular dystrophy, vitelliform, 5, Retinitis pigmentosa 56
INPP5E	Joubert syndrome 1, Mental retardation, truncal obesity, retinal dystrophy, and micropenis
INTS1	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies
INVS	Nephronophthisis 2, infantile
IQCB1	Senior-Loken syndrome 5
ISCA1	Multiple mitochondrial dysfunctions syndrome 5
ISCA2	Multiple mitochondrial dysfunctions syndrome 4
ISCU	Myopathy with lactic acidosis, hereditary
ITM2B	Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, Dementia, familial British, Dementia, familial Danish
JAG1	Deafness, congenital heart defects, and posterior embryotoxon, Alagille syndrome 1, Tetralogy of Fallot
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts
KARS1	Charcot-Marie-Tooth disease, recessive intermediate, B, Deafness, autosomal recessive 89, Leukoencephalopathy, progressive, infantile-onset, with or without deafness, Deafness, congenital, and adult-onset progressive leukoencephalopathy
KCNJ13	Leber congenital amaurosis 16, Snowflake vitreoretinal degeneration
KCNV2	Retinal cone dystrophy 3B
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions
KERA	Cornea plana 2, autosomal recessive
KIAA0556	Joubert syndrome 26
KIAA0586	Joubert syndrome 23, Short-rib thoracic dysplasia 14 with polydactyly
KIAA1549	Retinitis pigmentosa 86
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation
KIF1A	Mental retardation, autosomal dominant 9, Neuropathy, hereditary sensory, type IIC, Spastic paraplegia 30, autosomal recessive
KIF3B	Retinitis pigmentosa 89
KIF7	Al-Gazali-Bakalinova syndrome, Hydrolethalus syndrome 2, Acrocallosal syndrome, Joubert syndrome 12
KIZ	Retinitis pigmentosa 69
KLC2	Spastic paraplegia, optic atrophy, and neuropathy
KLHL7	Cold-induced sweating syndrome 3, Retinitis pigmentosa 42
LAMA1	Poretti-Boltshauser syndrome

LAMA5	Bent bone dysplasia syndrome 2, Nephrotic syndrome, type 26
LAMB2	Nephrotic syndrome, type 5, with or without ocular abnormalities, Pierson syndrome
LAMP2	Danon disease
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6
LARS1	Infantile liver failure syndrome 1
LARS2	Hydrops, lactic acidosis, and sideroblastic anemia, Perrault syndrome 4
LCA5	Leber congenital amaurosis 5
LCT	Lactase deficiency, congenital
LDLR	Hypercholesterolemia, familial, 1, LDL cholesterol level QTL2
LIAS	Hyperglycinemia, lactic acidosis, and seizures
LIM2	Cataract 19, multiple types
LIPT1	Lipoyltransferase 1 deficiency
LIPT2	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities
LONP1	CODAS syndrome
LOXL1	Exfoliation syndrome, susceptibility to
LOXL3	Myopia 28, autosomal recessive
LRAT	Leber congenital amaurosis 14, Retinal dystrophy, early-onset severe, Retinitis pigmentosa, juvenile
LRIT3	Night blindness, congenital stationary (complete), 1F, autosomal recessive
LRMDA	Albinism, oculocutaneous, type VII
LRP2	Donnai-Barrow syndrome
LRP4	Myasthenic syndrome, congenital, 17, Cenani-Lenz syndactyly syndrome, Sclerosteosis 2
LRP5	Exudative vitreoretinopathy 4, Hyperostosis, endosteal, Osteopetrosis, autosomal dominant 1, Osteoporosis-pseudoglioma syndrome, Osteosclerosis, Polycystic liver disease 4 with or without kidney cysts, van Buchem disease, type 2, Bone mineral density variability 1, Osteoporosis
LRPPRC	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)
LRRC32	Cleft palate, proliferative retinopathy, and developmental delay
LRRC6	Ciliary dyskinesia, primary, 19
LTBP2	Weill-Marchesani syndrome 3, recessive, Glaucoma 3, primary congenital, D, Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma
LTBP3	Dental anomalies and short stature, Geleophysic dysplasia 3
LYRM4	Combined oxidative phosphorylation deficiency 19
LYRM7	Mitochondrial complex III deficiency, nuclear type 8
LYST	Chediak-Higashi syndrome
LZTFL1	Bardet-Biedl syndrome 17
MAB21L1	Cerebellar, ocular, craniofacial, and genital syndrome
MAB21L2	Microphthalmia/coloboma and skeletal dysplasia syndrome
MACF1	Lissencephaly 9 with complex brainstem malformation
MAF	Ayme-Gripp syndrome, Cataract 21, multiple types
MAK	Retinitis pigmentosa 62
MAN2B1	Mannosidosis, alpha-, types I and II
MAPKAPK3	Macular dystrophy, patterned, 3
MAPKBP1	Nephronophthisis 20
MARS2	Combined oxidative phosphorylation deficiency 25, Spastic ataxia 3, autosomal recessive

MASP1	3MC syndrome 1
MASP2	MASP2 deficiency
MCAT	Optic atrophy 15
MDH2	Developmental and epileptic encephalopathy, , 51
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities; Optic atrophy 16
MERTK	Retinitis pigmentosa 38
MFF	Encephalopathy due to defective mitochondrial and peroxisomal fission 2
MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2A, Charcot-Marie-Tooth disease, axonal, type 2A2B, Hereditary motor and sensory neuropathy VIA
MFRP	Microphthalmia, isolated 5, Nanophthalmos 2
MFSD8	Ceroid lipofuscinosis, neuronal, 7, Macular dystrophy with central cone involvement
MGME1	Mitochondrial DNA depletion syndrome 11
MICOS13	Combined oxidative phosphorylation deficiency 37
MICU1	Myopathy with extrapyramidal signs
MIEF1	Optic atrophy 14
MIP	Cataract 15, multiple types
MIPEP	Combined oxidative phosphorylation deficiency 31
MIR204	Retinal dystrophy and iris coloboma with or without cataract
MITF	COMMAD syndrome, Tietz albinism-deafness syndrome, Waardenburg syndrome, type 2A, Waardenburg syndrome/ocular albinism, digenic, Melanoma, cutaneous malignant, susceptibility to, 8
Mitochondria genome	
MKKS	Bardet-Biedl syndrome 6, McKusick-Kaufman syndrome
MKS1	Bardet-Biedl syndrome 13, Joubert syndrome 28, Meckel syndrome 1
MLPH	Griscelli syndrome, type 3
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
MORC2	Charcot-Marie-Tooth disease, axonal, type 2Z
MPC1	Mitochondrial pyruvate carrier deficiency
MPV17	Charcot-Marie-Tooth disease, axonal, type 2EE, Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)
MRM2	Mitochondrial DNA depletion syndrome 17
MRPL12	Combined oxidative phosphorylation deficiency 45
MRPL3	Combined oxidative phosphorylation deficiency 9
MRPL44	Combined oxidative phosphorylation deficiency 16
MRPS14	Combined oxidative phosphorylation deficiency 38
MRPS16	Combined oxidative phosphorylation deficiency 2
MRPS2	Combined oxidative phosphorylation deficiency 36
MRPS22	Combined oxidative phosphorylation deficiency 5, Ovarian dysgenesis 7
MRPS23	Combined oxidative phosphorylation deficiency 46
MRPS34	Combined oxidative phosphorylation deficiency 32
MRPS7	Combined oxidative phosphorylation deficiency 34
MSTO1	Myopathy, mitochondrial, and ataxia
MTFMT	Combined oxidative phosphorylation deficiency 15, Mitochondrial complex I deficiency, nuclear type 27
MTO1	Combined oxidative phosphorylation deficiency 10
MTPAP	Spastic ataxia 4, autosomal recessive
MTTP	Abetalipoproteinemia

MUSK	Fetal akinesia deformation sequence 1, Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency
MVK	Hyper-IgD syndrome, Mevalonic aciduria, Porokeratosis 3, multiple types
MYH9	Deafness, autosomal dominant 17, Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss
MYO5A	Griscelli syndrome, type 1
MYO7A	Deafness, autosomal dominant 11, Deafness, autosomal recessive 2, Usher syndrome, type 1B
MYO9A	Myasthenic syndrome, congenital, 24, presynaptic
MYOC	Glaucoma 1A, primary open angle
NAA10	Microphthalmia, syndromic 1, Ogden syndrome
NADK2	2,4-dienoyl-CoA reductase deficiency
NARS2	Deafness, autosomal recessive 94, Combined oxidative phosphorylation deficiency 24
NAXE	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy
NBAS	Infantile liver failure syndrome 2, Short stature, optic nerve atrophy, and Pelger-Huet anomaly
NDP	Exudative vitreoretinopathy 2, X-linked, Norrie disease
NDUFA1	Mitochondrial complex I deficiency, nuclear type 12
NDUFA10	Mitochondrial complex I deficiency, nuclear type 22
NDUFA11	Mitochondrial complex I deficiency, nuclear type 14
NDUFA12	Mitochondrial complex I deficiency, nuclear type 23
NDUFA13	Mitochondrial complex I deficiency, nuclear type 28, Thyroid carcinoma, Hurthle cell
NDUFA2	Mitochondrial complex I deficiency, nuclear type 13
NDUFA4	Mitochondrial complex IV deficiency, nuclear type 21
NDUFA6	Mitochondrial complex I deficiency, nuclear type 33
NDUFA8	Mitochondrial complex I deficiency, nuclear type 37
NDUFA9	Mitochondrial complex I deficiency, nuclear type 26
NDUFAF1	Mitochondrial complex I deficiency, nuclear type 11
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10
NDUFAF3	Mitochondrial complex I deficiency, nuclear type 18
NDUFAF4	Mitochondrial complex I deficiency, nuclear type 15
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16
NDUFAF6	Mitochondrial complex I deficiency, nuclear type 17
NDUFAF8	Mitochondrial complex I deficiency, nuclear type 34
NDUFB11	Mitochondrial complex I deficiency, nuclear type 30, Linear skin defects with multiple congenital anomalies 3
NDUFB3	Mitochondrial complex I deficiency, nuclear type 25
NDUFB8	Mitochondrial complex I deficiency, nuclear type 32
NDUFB9	Mitochondrial complex I deficiency, nuclear type 24
NDUFS1	Mitochondrial complex I deficiency, nuclear type 5
NDUFS2	Mitochondrial complex I deficiency, nuclear type 6
NDUFS3	Mitochondrial complex I deficiency, nuclear type 8
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1
NDUFS6	Mitochondrial complex I deficiency, nuclear type 9
NDUFS7	Mitochondrial complex I deficiency, nuclear type 3
NDUFS8	Mitochondrial complex I deficiency, nuclear type 2
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4
NDUFV2	Mitochondrial complex I deficiency, nuclear type 7

NEFH	Amyotrophic lateral sclerosis, susceptibility to, Charcot-Marie-Tooth disease, axonal, type 2CC
NEK1	Short-rib thoracic dysplasia 6 with or without polydactyly, Amyotrophic lateral sclerosis, susceptibility to, 24
NEK2	Retinitis pigmentosa 67
NEK8	Nephronophthisis 9, Renal-hepatic-pancreatic dysplasia 2
NEUROD1	Maturity-onset diabetes of the young 6, Diabetes mellitus, noninsulin-dependent
NF2	Meningioma, NF2-related, somatic, Neurofibromatosis, type 2, Schwannomatosis, somatic
NFS1	Combined oxidative phosphorylation deficiency 52
NFU1	Multiple mitochondrial dysfunctions syndrome 1
NHS	Cataract 40, X-linked, Nance-Horan syndrome
NKX2-5	Atrial septal defect 7, with or without AV conduction defects, Conotruncal heart malformations, variable, Hypoplastic left heart syndrome 2, Hypothyroidism, congenital nongoitrous, 5, Tetralogy of Fallot, Ventricular septal defect 3
NME8	Ciliary dyskinesia, primary, 6
NMNAT1	Leber congenital amaurosis 9
NOD2	Blau syndrome, Inflammatory bowel disease 1, Crohn disease, Psoriatic arthritis, susceptibility to, Yao syndrome
NODAL	Heterotaxy, visceral, 5
NOTCH2	Alagille syndrome 2, Hajdu-Cheney syndrome
NOTCH3	Myofibromatosis, infantile 2, Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, Lateral meningocele syndrome
NPHP1	Joubert syndrome 4, Nephronophthisis 1, juvenile, Senior-Loken syndrome-1
NPHP3	Meckel syndrome 7, Nephronophthisis 3, Renal-hepatic-pancreatic dysplasia 1
NPHP4	Nephronophthisis 4, Senior-Loken syndrome 4
NR2E3	Enhanced S-cone syndrome, Retinitis pigmentosa 37
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome
NRL	Retinal degeneration, autosomal recessive, clumped pigment type, Retinitis pigmentosa 27
NSUN3	Combined oxidative phosphorylation deficiency 48
NTF4	Glaucoma 1, open angle, 1O
NUBPL	Mitochondrial complex I deficiency, nuclear type 21
NYX	Night blindness, congenital stationary (complete), 1A, X-linked
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia
OCA2	Albinism, brown oculocutaneous, Albinism, oculocutaneous, type II, Skin/hair/eye pigmentation 1, blond/brown hair, Skin/hair/eye pigmentation 1, blue/nonblue eyes
OCRL	Dent disease 2, Lowe syndrome
OFD1	Retinitis pigmentosa 23, Joubert syndrome 10, Orofaciodigital syndrome I, Simpson-Golabi-Behmel syndrome, type 2
OPA1	Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), Behr syndrome, Optic atrophy 1, Optic atrophy plus syndrome, Glaucoma, normal tension, susceptibility to
OPA3	3-methylglutaconic aciduria, type III, Optic atrophy 3 with cataract
OPA5	Optic atrophy 5
OPN1LW	Blue cone monochromacy, Colorblindness, protan
OPN1MW	Blue cone monochromacy, Colorblindness, deutan
OPN1SW	Colorblindness, tritan
OPTN	Amyotrophic lateral sclerosis 12, Glaucoma 1, open angle, E, Glaucoma, normal tension, susceptibility to
OTC	Ornithine transcarbamylase deficiency

OTOGL	Deafness, autosomal recessive 84B
OTX2	Microphthalmia, syndromic 5, Pituitary hormone deficiency, combined, 6, Retinal dystrophy, early-onset, with or without pituitary dysfunction
P3H2	Myopia, high, with cataract and vitreoretinal degeneration
PANK2	HARP syndrome, Neurodegeneration with brain iron accumulation 1
PARS2	Epileptic encephalopathy, early infantile, 75
PAX2	Glomerulosclerosis, focal segmental, 7, Papillorenal syndrome
PAX3	Craniofacial-deafness-hand syndrome, Rhabdomyosarcoma 2, alveolar, Waardenburg syndrome, type 1, Waardenburg syndrome, type 3
PAX6	Coloboma of optic nerve, Coloboma, ocular, Morning glory disc anomaly, Aniridia, Anterior segment dysgenesis 5, multiple subtypes, Cataract with late-onset corneal dystrophy, Foveal hypoplasia 1, Keratitis, Optic nerve hypoplasia
PC	Pyruvate carboxylase deficiency
PCARE	Retinitis pigmentosa 54
PCCA	Propionicacidemia
PCCB	Propionicacidemia
PCDH15	Deafness, autosomal recessive 23, Usher syndrome, type 1D/F digenic, Usher syndrome, type 1F
PCK2	PEPCK deficiency, mitochondrial
PCLO	Pontocerebellar hypoplasia, type 3
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy
PDE6A	Retinitis pigmentosa 43
PDE6B	Night blindness, congenital stationary, autosomal dominant 2, Retinitis pigmentosa-40
PDE6C	Cone dystrophy 4
PDE6D	Joubert syndrome 22
PDE6G	Retinitis pigmentosa 57
PDE6H	Achromatopsia 6, Retinal cone dystrophy 3
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency
PDHB	Pyruvate dehydrogenase E1-beta deficiency
PDHX	Lacticacidemia due to PDX1 deficiency
PDP1	Pyruvate dehydrogenase phosphatase deficiency
PDSS1	Coenzyme Q10 deficiency, primary, 2
PDSS2	Coenzyme Q10 deficiency, primary, 3
PDX1	MODY, type IV, Pancreatic agenesis 1, Diabetes mellitus, type II, susceptibility to
PDXK	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy
PDZD7	Deafness, autosomal recessive 57, Usher syndrome, type IIC, GPR98/PDZD7 digenic, Retinal disease in Usher syndrome type IIA, modifier of
PET100	Mitochondrial complex IV deficiency
PET117	Mitochondrial complex IV deficiency, nuclear type 19
PEX1	Heimler syndrome 1, Peroxisome biogenesis disorder 1A (Zellweger), Peroxisome biogenesis disorder 1B (NALD/IRD)
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), Peroxisome biogenesis disorder 6B
PEX11B	Peroxisome biogenesis disorder 14B
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), Peroxisome biogenesis disorder 3B
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), Peroxisome biogenesis disorder 11B
PEX14	Peroxisome biogenesis disorder 13A (Zellweger)
PEX16	Peroxisome biogenesis disorder 8A (Zellweger), Peroxisome biogenesis disorder 8B
PEX19	Peroxisome biogenesis disorder 12A (Zellweger)
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), Peroxisome biogenesis disorder 5B

PEX26	Peroxisome biogenesis disorder 7A (Zellweger), Peroxisome biogenesis disorder 7B
PEX3	Peroxisome biogenesis disorder 10B, Peroxisome biogenesis disorder 10A (Zellweger)
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), Peroxisome biogenesis disorder 2B, Rhizomelic chondrodysplasia punctata, type 5
PEX6	Heimler syndrome 2, Peroxisome biogenesis disorder 4A (Zellweger), Peroxisome biogenesis disorder 4B
PEX7	Peroxisome biogenesis disorder 9B, Rhizomelic chondrodysplasia punctata, type 1
PGK1	Phosphoglycerate kinase 1 deficiency
PHYH	Refsum disease
PIBF1	Joubert syndrome 33
PITPNM3	Cone-rod dystrophy 5
PITRM1	Spinocerebellar ataxia, autosomal recessive 30
PITX2	Anterior segment dysgenesis 4, Axenfeld-Rieger syndrome, type 1, Ring dermoid of cornea
PITX3	Anterior segment dysgenesis 1, multiple subtypes, Cataract 11, multiple types, Cataract 11, syndromic, autosomal recessive
PLA2G5	Fleck retina, familial benign
PLA2G6	Infantile neuroaxonal dystrophy 1, Neurodegeneration with brain iron accumulation 2B, Parkinson disease 14, autosomal recessive
PLEC	Epidermolysis bullosa simplex with nail dystrophy, Epidermolysis bullosa simplex with muscular dystrophy, Epidermolysis bullosa simplex with pyloric atresia, Epidermolysis bullosa simplex, Ogna type, Muscular dystrophy, limb-girdle, autosomal recessive 17
PLK4	Microcephaly and chorioretinopathy, autosomal recessive, 2
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1
PLOD3	Lysyl hydroxylase 3 deficiency
PMP22	Neuropathy, inflammatory demyelinating, Charcot-Marie-Tooth disease, type 1A, Charcot-Marie-Tooth disease, type 1E, Dejerine-Sottas disease, Neuropathy, recurrent, with pressure palsies, Roussy-Levy syndrome
PMPCA	Spinocerebellar ataxia, autosomal recessive 2
PMPCB	Multiple mitochondrial dysfunctions syndrome 6
PNPLA2	Neutral lipid storage disease with myopathy
PNPLA6	Laurence-Moon syndrome, Boucher-Neuhauser syndrome, Oliver-McFarlane syndrome, Spastic paraplegia 39, autosomal recessive
PNPLA8	Mitochondrial myopathy with lactic acidosis
PNPT1	Combined oxidative phosphorylation deficiency 13, Deafness, autosomal recessive 70, spinocerebellar ataxia 25
POC1B	Cone-rod dystrophy 20
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), Mitochondrial DNA depletion syndrome 4B (MNGIE type), Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), Progressive external ophthalmoplegia, autosomal dominant 1, Progressive external ophthalmoplegia, autosomal recessive 1
POLG2	Mitochondrial DNA depletion syndrome 16 (hepatic type), Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4
POLRMT	Combined oxidative phosphorylation deficiency 55
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, Retinitis pigmentosa 76
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8
POMK	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12

POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2
PORCN	Focal dermal hypoplasia
PPA2	Sudden cardiac failure, alcohol-induced, Sudden cardiac failure, infantile
PPT1	Ceroid lipofuscinosis, neuronal, 1
PRCD	Retinitis pigmentosa 36
PRDM13	Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, Pontocerebellar hypoplasia, type 17
PRDM5	Brittle cornea syndrome 2
PREPL	Myasthenic syndrome, congenital, 22
PRICKLE3	Leber hereditary optic neuropathy, modifier of
PROM1	Cone-rod dystrophy 12, Macular dystrophy, retinal, 2, Retinitis pigmentosa 41, Stargardt disease 4
PROS1	Thrombophilia due to protein S deficiency, autosomal dominant, Thrombophilia due to protein S deficiency, autosomal recessive
PRPF3	Retinitis pigmentosa 18
PRPF31	Retinitis pigmentosa 11
PRPF4	Retinitis pigmentosa 70
PRPF6	Retinitis pigmentosa 60
PRPF8	Retinitis pigmentosa 13
PRPH2	Choroidal dystrophy, central areolar 2, Leber congenital amaurosis 18, Macular dystrophy, patterned, 1, Macular dystrophy, vitelliform, 3, Retinitis pigmentosa 7 and digenic form, Retinitis punctata albescens
PRPS1	Arts syndrome, Charcot-Marie-Tooth disease, X-linked recessive, 5, Deafness, X-linked 1, Gout, PRPS-related, Phosphoribosylpyrophosphate synthetase superactivity
PRR12	Neuroocular syndrome
PRSS56	Microphthalmia, isolated 6
PTCD3	Combined oxidative phosphorylation deficiency 51
PTCH1	Basal cell carcinoma, somatic, Basal cell nevus syndrome, Holoprosencephaly 7
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1
PXDN	Anterior segment dysgenesis 7, with sclerocornea
PYGM	McArdle disease
QARS1	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy
QRSL1	Combined oxidative phosphorylation deficiency 40
RAB18	Warburg micro syndrome 3
RAB27A	Griscelli syndrome, type 2
RAB28	Cone-rod dystrophy 18
RAB3GAP1	Warburg micro syndrome 1
RAB3GAP2	Martsolf syndrome, Warburg micro syndrome 2
RAPSN	Fetal akinesia deformation sequence 2, Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency
RARB	Microphthalmia, syndromic 12
RARS1	Leukodystrophy, hypomyelinating, 9
RARS2	Pontocerebellar hypoplasia, type 6
RAX	Microphthalmia, isolated 3
RAX2	Macular degeneration, age-related, 6, Cone-rod dystrophy 11, Retinitis pigmentosa 95

RB1	Bladder cancer, somatic, Osteosarcoma, somatic, Retinoblastoma, Retinoblastoma, trilateral, Small cell cancer of the lung, somatic
RBP3	Retinitis pigmentosa 66
RBP4	Microphthalmia, isolated, with coloboma 10, Retinal dystrophy, iris coloboma, and comedogenic acne syndrome
RCBTB1	Retinal dystrophy with or without extraocular anomalies
RCD1	Retinal cone dystrophy-1
RD3	Leber congenital amaurosis 12
RDH11	Retinal dystrophy, juvenile cataracts, and short stature syndrome
RDH12	Leber congenital amaurosis 13
RDH5	Fundus albipunctatus
RECQL4	Baller-Gerold syndrome, RAPADILINO syndrome, Rothmund-Thomson syndrome, type 2,
REEP1	Neuronopathy, distal hereditary motor, type VB, Spastic paraplegia 31, autosomal dominant
REEP6	Retinitis pigmentosa 77
RERE	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart
RGR	Retinitis pigmentosa 44
RGS9	Bradyopsia, 1
RGS9BP	Bradyopsia, 2
RHO	Night blindness, congenital stationary, autosomal dominant 1, Retinitis pigmentosa 4, autosomal dominant or recessive, Retinitis punctata albescens
RIMS2	Cone-rod synaptic disorder syndrome, congenital nonprogressive
RLBP1	Bothnia retinal dystrophy, Fundus albipunctatus, Newfoundland rod-cone dystrophy, Retinitis punctata albescens
RMND1	Combined oxidative phosphorylation deficiency 11
RNANC	Persistent hyperplastic primary vitreous, autosomal recessive
RNASEH1	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2
ROM1	Retinitis pigmentosa 7, digenic form
RP1	Retinitis pigmentosa 1
RP1L1	Occult macular dystrophy, Retinitis pigmentosa 88
RP2	Retinitis pigmentosa 2
RP9	Retinitis pigmentosa 9
RPE65	Leber congenital amaurosis 2, Retinitis pigmentosa 20, Retinitis pigmentosa 87 with choroidal involvement
RPGR	Cone-rod dystrophy, X-linked, 1, Macular degeneration, X-linked atrophic, Retinitis pigmentosa 3, Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness
RPGRIP1	Cone-rod dystrophy 13, Leber congenital amaurosis 6
RPGRIP1L	COACH syndrome, Joubert syndrome 7, Meckel syndrome 5
RPL10	Mental retardation, X-linked, syndromic, 35, Autism, susceptibility to, X-linked 5
RPL15	Diamond-Blackfan anemia 12
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), Mitochondrial DNA depletion syndrome 8B (MNGIE type), Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5
RS1	Retinoschisis
RSPH4A	Ciliary dyskinesia, primary, 11
RSPH9	Ciliary dyskinesia, primary, 12
RTN4IP1	Optic atrophy 10 with or without ataxia, mental retardation, and seizures
SACS	Spastic ataxia, Charlevoix-Saguenay type

SAG	Oguchi disease-1, Retinitis pigmentosa 47, autosomal recessive, Retinitis pigmentosa 96, autosomal dominant
SALL2	Coloboma, ocular, autosomal recessive
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis
SBF2	Charcot-Marie-Tooth disease, type 4B2
SC5D	Lathosterolosis
SCAPER	Intellectual developmental disorder and retinitis pigmentosa
SCARF2	Van den Ende-Gupta syndrome
SCN4A	Hyperkalemic periodic paralysis, type 2, Hypokalemic periodic paralysis, type 2, Myasthenic syndrome, congenital, 16, Myotonia congenita, atypical, acetazolamide-responsive, Paramyotonia congenita
SCO1	Mitochondrial complex IV deficiency, nuclear type IV
SCO2	Mitochondrial complex IV deficiency, nuclear type 2, Myopia 6
SDCCAG8	Bardet-Biedl syndrome 16, Senior-Loken syndrome 7
SDHA	Cardiomyopathy, dilated, 1GG, Neurodegeneration with ataxia and late-onset optic atrophy, Mitochondrial respiratory chain complex II deficiency, Paragangliomas 5
SDHAF1	Mitochondrial complex II deficiency, nuclear type 2
SDHAF2	Paragangliomas 2
SDHB	Gastrointestinal stromal tumor, Paraganglioma and gastric stromal sarcoma, Paragangliomas 4, Pheochromocytoma
SDHC	Gastrointestinal stromal tumor, Paraganglioma and gastric stromal sarcoma, Paragangliomas 3
SDHD	Mitochondrial complex II deficiency, Paraganglioma and gastric stromal sarcoma, Paragangliomas 1, with or without deafness, Pheochromocytoma
SEC23A	Craniolenticulosutural dysplasia
SEMA4A	Cone-rod dystrophy 10, Retinitis pigmentosa 35
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome
SETX	Amyotrophic lateral sclerosis 4, juvenile, Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2
SFXN4	Combined oxidative phosphorylation deficiency 18
SHH	Holoprosencephaly 3, Microphthalmia with coloboma 5, Schizencephaly, Single median maxillary central incisor
SIL1	Marinesco-Sjogren syndrome
SIX3	Holoprosencephaly 2, Schizencephaly
SIX5	Branchiootorenal syndrome 2
SIX6	Optic disc anomalies with retinal and/or macular dystrophy
SLC16A12	Cataract 47, juvenile, with microcornea
SLC18A3	Myasthenic syndrome, congenital, 21, presynaptic
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)
SLC22A5	Carnitine deficiency, systemic primary
SLC24A1	Night blindness, congenital stationary (complete), 1D, autosomal recessive
SLC24A5	Albinism, oculocutaneous, type VI, Skin/hair/eye pigmentation 4, fair/dark skin
SLC25A1	Myasthenic syndrome, congenital, 23, presynaptic, Combined D-2- and L-2-hydroxyglutaric aciduria
SLC25A12	Epileptic encephalopathy, early infantile, 39
SLC25A13	Citrullinemia, adult-onset type II, Citrullinemia, type II, neonatal-onset
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
SLC25A19	Microcephaly, Amish type, Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)

SLC25A20	Carnitine-acylcarnitine translocase deficiency
SLC25A21	Mitochondrial DNA depletion syndrome, 18
SLC25A26	Combined oxidative phosphorylation deficiency 28
SLC25A3	Mitochondrial phosphate carrier deficiency
SLC25A32	Exercise intolerance, riboflavin-responsive
SLC25A38	Anemia, sideroblastic, 2, pyridoxine-refractory
SLC25A4	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2
SLC25A42	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression
SLC25A46	Neuropathy, hereditary motor and sensory, type VIB, Pontocerebellar hypoplasia, type 1E
SLC2A1	Dystonia 9, GLUT1 deficiency syndrome 1, infantile onset, severe, GLUT1 deficiency syndrome 2, childhood onset, Stomatin-deficient cryohydrocytosis with neurologic defects, Epilepsy, idiopathic generalized, susceptibility to, 12
SLC33A1	Huppke-Brendel syndrome. Spastic paraplegia 42, autosomal dominant
SLC37A4	Glycogen storage disease Ib, Glycogen storage disease Ic, Congenital disorder of glycosylation, type Iiw
SLC38A8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis
SLC39A13	Ehlers-Danlos syndrome, spondylodysplastic type, 3
SLC44A1	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline
SLC45A2	Albinism, oculocutaneous, type IV, Skin/hair/eye pigmentation 5, black/nonblack hair, Skin/hair/eye pigmentation 5, dark/fair skin, Skin/hair/eye pigmentation 5, dark/light eyes
SLC5A7	Myasthenic syndrome, congenital, 20, presynaptic, Neuronopathy, distal hereditary motor, type VIIA
SLC6A8	Cerebral creatine deficiency syndrome 1
SLC7A14	Retinitis pigmentosa 68
SMOC1	Microphthalmia with limb anomalies
SNAI2	Piebaldism, Waardenburg syndrome, type 2D
SNAP25	Myasthenic syndrome, congenital, 18
SNRNP200	Retinitis pigmentosa 33
SNX10	Osteopetrosis, autosomal recessive 8
SOD1	Amyotrophic lateral sclerosis 1, Spastic tetraplegia and axial hypotonia, progressive
SOX10	PCWH syndrome, Waardenburg syndrome, type 2E, with or without neurologic involvement, Waardenburg syndrome, type 4C
SOX2	Microphthalmia, syndromic 3, Optic nerve hypoplasia and abnormalities of the central nervous system
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency, Panhypopituitarism, X-linked
SPAST	Spastic paraplegia 4, autosomal dominant
SPATA5	Epilepsy, hearing loss, and mental retardation syndrome
SPATA7	Leber congenital amaurosis 3, Retinitis pigmentosa, juvenile, autosomal recessive
SPG11	Amyotrophic lateral sclerosis 5, juvenile, Charcot-Marie-Tooth disease, axonal, type 2X, Spastic paraplegia 11, autosomal recessive
SPG15	Spastic paraplegia 15, autosomal recessive
SPG2	Spastic paraplegia 2, X-linked
SPG35	Spastic paraplegia 35, autosomal recessive
SPG45	Spastic paraplegia 45, autosomal recessive

SPG46	Spastic paraplegia 46, autosomal recessive
SPG54	Spastic paraplegia 54, autosomal recessive
SPG55	Spastic paraplegia 55, autosomal recessive
SPG57	Spastic paraplegia 57, autosomal recessive
SPG7	Spastic paraplegia 7, autosomal recessive
SRD5A3	Congenital disorder of glycosylation, type Iq, Kahrizi syndrome
SSBP1	Optic atrophy 13 with retinal and foveal abnormalities
STAT2	Immunodeficiency 44
STRA6	Microphthalmia, isolated, with coloboma 8, Microphthalmia, syndromic 9
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)
SUFU	Basal cell nevus syndrome, Joubert syndrome 32, Medulloblastoma, desmoplastic, Meningioma, familial, susceptibility to
SUOX	Sulfite oxidase deficiency
SURF1	Charcot-Marie-Tooth disease, type 4K, Leigh syndrome, due to COX IV deficiency
SYNE2	Emery-Dreifuss muscular dystrophy 5, autosomal dominant
SYT2	Myasthenic syndrome, congenital, 7, presynaptic
TACO1	Mitochondrial complex IV deficiency
TARS2	Combined oxidative phosphorylation deficiency 21
TAZ	Barth syndrome
TBC1D20	Warburg micro syndrome 4
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy, Hypoparathyroidism-retardation-dysmorphism syndrome, Kenny-Caffey syndrome, type 1
TBK1	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8
TCTN1	Joubert syndrome 13
TCTN2	Meckel syndrome 8, Joubert syndrome 24
TCTN3	Joubert syndrome 18, Orofaciodigital syndrome IV
TDRD7	Cataract 36
TEAD1	Sveinsson chorioretinal atrophy
TEK	Glaucoma 3, primary congenital, E, Venous malformations, multiple cutaneous and mucosal
TENM3	Microphthalmia, isolated, with coloboma 9, Microphthalmia, syndromic 15
TFAM	Mitochondrial DNA depletion syndrome 15 (hepatocerebral type)
TFAP2A	Branchiooculofacial syndrome
TGFBI	Corneal dystrophy, Avellino type, Corneal dystrophy, epithelial basement membrane, Corneal dystrophy, Groenouw type I, Corneal dystrophy, lattice type I, Corneal dystrophy, lattice type IIIA, Corneal dystrophy, Reis-Bucklers type, Corneal dystrophy, Thiel-Behnke type
TIMM22	Combined oxidative phosphorylation deficiency 43
TIMM50	3-methylglutaconic aciduria, type IX
TIMM8A	Mohr-Tranebjaerg syndrome
TIMMDC1	Mitochondrial complex I deficiency, nuclear type 31
TIMP3	Sorsby fundus dystrophy
TK2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, Mitochondrial DNA depletion syndrome 2 (myopathic type)
TKFC	Triokinase and FMN cyclase deficiency syndrome
TLCD3B	Cone-rod dystrophy 22

TLR3	HIV1 infection, resistance to, Immunodeficiency 83, susceptibility to viral infections
TMED7	
TMEM107	Joubert syndrome 29, Meckel syndrome 13, Orofaciodigital syndrome XVI
TMEM126A	Optic atrophy 7
TMEM126B	Mitochondrial complex I deficiency, nuclear type 29
TMEM138	Joubert syndrome 16
TMEM216	Joubert syndrome 2, Meckel syndrome 2
TMEM218	Joubert syndrome 39
TMEM231	Joubert syndrome 20, Meckel syndrome 11
TMEM237	Joubert syndrome 14
TMEM67	RHYNS syndrome, COACH syndrome, Joubert syndrome 6, Meckel syndrome 3, Nephronophthisis 11, Bardet-Biedl syndrome 14, modifier of
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2
TMEM98	Nanophthalmos 4
TNXB	Ehlers-Danlos syndrome, classic-like, 1, Vesicoureteral reflux 8
TOP3A	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, Microcephaly, growth restriction, and increased sister chromatid exchange 2
TOPORS	Retinitis pigmentosa 31
TOR1AIP1	Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)
TPP1	Ceroid lipofuscinosis, neuronal, 2, Spinocerebellar ataxia, autosomal recessive 7
TRAF7	Cardiac, facial, and digital anomalies with developmental delay
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, Chilblain lupus, Vasculopathy, retinal, with cerebral leukodystrophy, Systemic lupus erythematosus, susceptibility to
TRIM32	Bardet-Biedl syndrome 11, Muscular dystrophy, limb-girdle, autosomal recessive 8
TRIT1	Combined oxidative phosphorylation deficiency 35
TRMT10C	Combined oxidative phosphorylation deficiency 30
TRMT5	Combined oxidative phosphorylation deficiency 26
TRMU	Liver failure, transient infantile, Deafness, mitochondrial, modifier of
TRNT1	Retinitis pigmentosa and erythrocytic microcytosis, Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay
TRPM1	Night blindness, congenital stationary (complete), 1C, autosomal recessive
TSFM	Combined oxidative phosphorylation deficiency 3
TSPAN12	Exudative vitreoretinopathy 5
TTC19	Mitochondrial complex III deficiency, nuclear type 2
TTC21B	Nephronophthisis 12, Short-rib thoracic dysplasia 4 with or without polydactyly
TTC8 (BBS8)	Retinitis pigmentosa 51, Bardet-Biedl syndrome 8
TTLL5	Cone-rod dystrophy 19
TTPA	Ataxia with isolated vitamin E deficiency
TUB	Retinal dystrophy and obesity
TUBB4B	Leber congenital amaurosis with early-onset deafness
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, 3
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1
TUFM	Combined oxidative phosphorylation deficiency 4
TULP1	Leber congenital amaurosis 15, Retinitis pigmentosa 14
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), Perrault syndrome 5, Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3

TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
TYR	Albinism, oculocutaneous, type IA, Albinism, oculocutaneous, type IB, Waardenburg syndrome/albinism, digenic, Skin/hair/eye pigmentation 3, blue/green eyes, Skin/hair/eye pigmentation 3, light/dark/freckling skin, Melanoma, cutaneous malignant, susceptibility to, 8
TYRP1	Albinism, oculocutaneous, type III, Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)
UBIAD1	Corneal dystrophy, Schnyder type
UCHL1	Spastic paraplegia 79, autosomal recessive, Parkinson disease 5, susceptibility to
UNC119	Cone-rod dystrophy, Immunodeficiency 13
UQCC2	Mitochondrial complex III deficiency, nuclear type 7
UQCC3	Mitochondrial complex III deficiency, nuclear type 9
UQCRB	Mitochondrial complex III deficiency, nuclear type 3
UQCRC2	Mitochondrial complex III deficiency, nuclear type 5
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4
USH1C	Deafness, autosomal recessive 18A, Usher syndrome, type 1C
USH1G	Usher syndrome, type 1G
USH2A	Retinitis pigmentosa 39, Usher syndrome, type 2A
USH2C	Usher syndrome, type 2C
USH2D	Usher syndrome, type 2D
VAMP1	Myasthenic syndrome, congenital, 25, Spastic ataxia 1, autosomal dominant
VARS2	Combined oxidative phosphorylation deficiency 20
VAX1	Microphthalmia, syndromic 11
VCAN	Wagner syndrome 1
VHL	Erythrocytosis, familial, 2, Hemangioblastoma, cerebellar, somatic, Pheochromocytoma, Renal cell carcinoma, somatic, von Hippel-Lindau syndrome
VPS13B	Cohen syndrome
VSX2	Microphthalmia with coloboma 3, Microphthalmia, isolated 2
WARS2	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures
WDPCP	Bardet-Biedl syndrome 15, Congenital heart defects, hamartomas of tongue, and polysyndactyly
WDR19	Cranioectodermal dysplasia 4, Short-rib thoracic dysplasia 5 with or without polydactyly, Nephronophthisis 13, Senior-Loken syndrome 8
WDR34	Short-rib thoracic dysplasia 11 with or without polydactyly
WDR35	Cranioectodermal dysplasia 2, Short-rib thoracic dysplasia 7 with or without polydactyly
WDR36	Glaucoma 1, open angle, G
WDR73	Galloway-Mowat syndrome 1
WFS1	Cataract 41, Deafness, autosomal dominant 6/14/38, Wolfram syndrome 1, Wolfram-like syndrome, autosomal dominant, Diabetes mellitus, noninsulin-dependent, association with
WFS2	Wolfram syndrome 2
WHRN	Deafness, autosomal recessive 31, Usher syndrome, type 2D
XPNPEP3	Nephronophthisis-like nephropathy 1
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2
YME1L1	Optic atrophy 11
ZEB2	Mowat-Wilson syndrome
ZFYVE26	Spastic paraplegia 15, autosomal recessive
ZIC3	Congenital heart defects, nonsyndromic, 1, X-linked, Heterotaxy, visceral, 1, X-linked, VACTERL association, X-linked

ZNF408	Exudative vitreoretinopathy 6, Retinitis pigmentosa 72
ZNF423	Joubert syndrome 19, Nephronophthisis 14
ZNF469	Brittle cornea syndrome 1
ZNF513	Retinitis pigmentosa 58
ZNHIT3	PEHO syndrome
ANGPT1	GWAS association with Glaucoma
ATXN7	Spinocerebellar ataxia 7 - CAG expansion disorder
C2	GWAS association with AMD
C3	GWAS association with AMD
C9	GWAS association with AMD
CD36	GWAS association with AMD and Glaucoma
CDKN2A	GWAS association with AMD
CFB	Macular degeneration, age-related, 14, reduced risk of
CFHR2	GWAS association with AMD
ACACB	Candidate Modifier Genes for the Penetrance of Leber's Hereditary Optic Neuropathy
ACADL	Candidate gene for metabolic disorder
AKR1C1	Candidate gene for nonsyndromic lipedema
ARFGAP2	Candidate gene for pigmentation disorders
ASIC5	Candidate gene for Usher syndrome
ATP5MF	Candidate gene for Mitochondria disorder; Metabolic disorder
ATP5MG	Candidate gene for Mitochondria disorder; Metabolic disorder
ATP5MGL	Candidate gene for Mitochondria disorder; Metabolic disorder
ATP5PB	Candidate gene for Mitochondria disorder; Metabolic disorder
ATP5PD	Candidate gene for Mitochondria disorder; Metabolic disorder
ATPAF1	Candidate gene for Mitochondria disorder; Metabolic disorder
BCO2	Candidate gene for metobolic disorder
C7orf26	Potential Candidate Gene
CCER1	Potential Candidate Gene
CHERP	Potential Candidate Gene
CLTA	Potential Candidate Gene
COA4	Mitochondria candidate gene
COL26A1	Potential Candidate Gene
COX17	Potential Candidate Gene
COX19	Potential Candidate Gene
COX6B2	Potential Candidate Gene
COX7A1	Potential Candidate Gene
CRYGA	Potential Candidate Gene
CSMD1	Potential Candidate Gene
CSMD2	Potential Candidate Gene
DCDC1	Potential Candidate Gene
DHX32	Inherited retinal dystrophy candidate gene
DNAJC17	Inherited retinal dystrophy candidate gene
DSCAML1	Inherited retinal dystrophy candidate gene
ENSA	Potential Candidate Gene
FCN1	Potential Candidate Gene
FGF21	Potential Candidate Gene
FOXH1	Potential Candidate Gene
GPR45	Potential Candidate Gene

IFT88	Inherited retinal dystrophy candidate gene PMID: 29978320
KIF24	Potential Candidate Gene
LEFTY2	Candidate gene for exfoliation glaucoma PMID 34964803
LHX2	Candidate gene for inherited retinal dystrophy PMID 23595746
MMP1	Candidate gene for uveal melanoma
MPRIP	Potential Candidate Gene
MRRF	Potential Candidate Gene
MRS2	Potential Candidate Gene
NDRG4	Potential Candidate Gene
NDUFA7	Potential Candidate Gene
NDUFAF7	Candidate gene for degenerative myopia; PMID 28837730
NDUFB6	Potential Candidate Gene
NDUFS5	Potential Candidate Gene
NDUFV3	Potential Candidate Gene
NECTIN3	Candidate gene for hearing loss; PMID 36568980
NUTF2	Potential Candidate Gene
NXNL1	Candidate gene for rod-cone dystrophies; PMID 20139892
OR2W3	Candidate gene for retinitis pigmentosa; PMID 25783483
OXA1L	Potential Candidate Gene
PKM	Potential Candidate Gene
POC5	Candidate gene for retinitis pigmentosa: PMID 29272404
PTCD1	Potential Candidate Gene
RHEX	Potential Candidate Gene
RIMS1	Candidate gene for rod-cone dystrophy; PMID 17237123
RPH3A	Candidate gene for neurodevelopmental disorders; PMID 37403762
RTBDN	Candidate gene for rod-cone degeneration; PMID 35873559
SAMD11	Candidate gene for retinitis pigmentosa; PMID 27734943
SCLT1	Candidate gene for Bardet-Biedl syndrome; PMID 32253632
SDHAF3	Candidate gene for paragangliomas; PMID 28738844
SDHAF4	Candidate gene for paragangliomas; PMID 32948195
SLC4A7	Candidate gene for rod-cone dystrophy; PMID 32594822
SLC6A15	Potential Candidate Gene
SNX3	Potential Candidate Gene
SPG68	Potential Candidate Gene
SPP2	Candidate gene for retinitis pigmentosa; PMID 26459573
SREBF2	Candidate gene for lens function; PMID 21858719
STARD9	Potential Candidate Gene
STK38L	Potential Candidate Gene
SYTL4	Potential Candidate Gene
TBC1D32	Candidate gene for retinitis pigmentosa; PMID 37768732
TFB1M	Candidate gene for hearing loss; PMID 15542390
TLR4	Potential Candidate Gene
TMEM114	Potential Candidate Gene
TMEM65	Potential Candidate Gene
TNPO1	Potential Candidate Gene
UNC13A	Candidate gene for congenital myasthenic syndrome
UQCC1	Potential Candidate Gene
UQCR10	Potential Candidate Gene

UQCR11	Potential Candidate Gene
VAX2	Candidate gene for retinal dystrophy; PMID 26068435
WDR17	Potential Candidate Gene