VISION DISORDERS GENE PANEL DG 3.5.0 (520 genes)

Releasedate: 05-12-2022

Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCA4	100%	100%	Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Stargardt disease 1, 248200
ABCC6	100%	100%	Pseudoxanthoma elasticum, 264800 Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, forme fruste, 177850
ABHD12	100%	100%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACBD5	100%	100%	Retinal dystrophy with leukodystrophy, 618863
ACO2	100%	100%	Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ADAM9	100%	100%	Cone-rod dystrophy 9, 612775
ADAMTS18	100%	100%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTSL4	100%	100%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADGRV1	100%	100%	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
ADIPOR1	100%	100%	No OMIM disease ID
AFG3L2	100%	100%	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGBL1	100%	100%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	100%	100%	Retinitis pigmentosa 75, 617023
AGK	92%	92%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AHI1	100%	100%	Joubert syndrome 3, 608629
AHR	100%	100%	?Retinitis pigmentosa 85, 618345

AIPL1	100%	100%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
ALDH18A1	100%	100%	Spastic paraplegia 9A, autosomal dominant, 601162 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9B, autosomal recessive, 616586 Cutis laxa, autosomal dominant 3, 616603
ALDH1A3	100%	100%	Microphthalmia, isolated 8, 615113
ALDH3A2	94%	94%	Sjogren-Larsson syndrome, 270200
ALMS1	100%	100%	Alstrom syndrome, 203800
AMACR	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
ANK3	100%	100%	Intellectual developmental disorder, autosomal recessive 37, 615493
AP3B1	100%	100%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	100%	100%	?Hermansky-Pudlak syndrome 10, 617050
ARHGEF18	100%	100%	Retinitis pigmentosa 78, 617433
ARL13B	100%	100%	Joubert syndrome 8, 612291
ARL2	100%	100%	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082
ARL2BP	100%	100%	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	100%	100%	Retinitis pigmentosa 83, 618173 Joubert syndrome 35, 618161
ARL6	100%	100%	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARR3	100%	100%	Myopia 26, X-linked, female-limited, 301010
ARSG	100%	100%	Usher syndrome, type IV, 618144
ASB10	100%	100%	Glaucoma 1, open angle, F, 603383
ASPH	100%	100%	Traboulsi syndrome, 601552
ASRGL1	100%	100%	No OMIM disease ID
ATF6	100%	100%	Achromatopsia 7, 616517
ATOH7	100%	100%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
B3GLCT	100%	100%	Peters-plus syndrome, 261540
BBIP1	100%	100%	?Bardet-Biedl syndrome 18, 615995
BBS1	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	100%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	Bardet-Biedl syndrome 12, 615989

BBS2	100%	100%	Retinitis pigmentosa 74, 616562 Bardet-Biedl syndrome 2, 615981
BBS4	100%	100%	Bardet-Biedl syndrome 4, 615982
BBS5	100%	100%	Bardet-Biedl syndrome 5, 615983
BBS7	100%	100%	Bardet-Biedl syndrome 7, 615984
BBS9	96%	96%	Bardet-Biedl syndrome 9, 615986
BCOR	100%	100%	Microphthalmia, syndromic 2, 300166
BEST1	100%	100%	Macular dystrophy, vitelliform, 2, 153700 ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 Retinitis pigmentosa-50, 613194 Retinitis pigmentosa, concentric, 613194 Vitreoretinochoroidopathy, 193220 Bestrophinopathy, autosomal recessive, 611809
BFSP1	100%	100%	Cataract 33, multiple types, 611391
BFSP2	100%	100%	Cataract 12, multiple types, 611597
BLOC1S3	100%	100%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S5	100%	100%	Hermansky-Pudlak syndrome 11, 619172
BLOC1S6	100%	100%	?Hermansky-Pudlak syndrome 9, 614171
BMP4	100%	100%	Orofacial cleft 11, 600625 Microphthalmia, syndromic 6, 607932
BMPR1B	100%	100%	Acromesomelic dysplasia 3, 609441 Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849
C19orf12	100%	100%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1QTNF5	100%	100%	Retinal degeneration, late-onset, autosomal dominant, 605670
CABP4	100%	100%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1F	100%	100%	Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Aland Island eye disease, 300600
CACNA2D4	100%	100%	Retinal cone dystrophy 4, 610478
CAPN5	100%	100%	Vitreoretinopathy, neovascular inflammatory, 193235
CC2D2A	98%	98%	COACH syndrome 2, 619111 Retinitis pigmentosa 93, 619845 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285

CCT2	100%	100%	No OMIM disease ID
CDH2	100%	100%	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 ?Attention deficit-hyperactivity disorder 8, 619957 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDH23	100%	100%	Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 12, 601386
CDH3	100%	100%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDH4	100%	100%	No OMIM disease ID
CDHR1	100%	100%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK10	100%	100%	Al Kaissi syndrome, 617694
CEP120	100%	100%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 Joubert syndrome 31, 617761
CEP164	100%	100%	Nephronophthisis 15, 614845
CEP250	100%	100%	Cone-rod dystrophy and hearing loss 2, 618358
CEP290	100%	100%	Leber congenital amaurosis 10, 611755 Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991 Meckel syndrome 4, 611134
CEP41	100%	100%	Joubert syndrome 15, 614464
CEP78	100%	100%	Cone-rod dystrophy and hearing loss, 617236
CEP83	100%	100%	Nephronophthisis 18, 615862
CERKL	99%	98%	Retinitis pigmentosa 26, 608380
CFAP410	100%	100%	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C8orf37	100%	100%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500 Bardet-Biedl syndrome 21, 617406
CFH	100%	100%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CHD7	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHM	99%	98%	Choroideremia, 303100
CHMP4B	100%	100%	Cataract 31, multiple types, 605387

CHN1	97%	97%	Duane retraction syndrome 2, 604356
CHRDL1	100%	100%	Megalocornea 1, X-linked, 309300
CHST6	100%	100%	Macular corneal dystrophy, 217800
CIB2	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CISD2	100%	100%	Wolfram syndrome 2, 604928
CLCC1	100%	100%	Retinitis pigmentosa 32, 609913
CLDN19	100%	100%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC3B	100%	100%	Macular dystrophy, retinal, 4, 619977
CLN3	93%	93%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	83%	83%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	100%	100%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 Ceroid lipofuscinosis, neuronal, 8, 600143
CLRN1	100%	100%	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180
CLUAP1	100%	100%	No OMIM disease ID
CNGA1	91%	91%	Retinitis pigmentosa 49, 613756
CNGA3	100%	100%	Achromatopsia 2, 216900
CNGB1	100%	100%	Retinitis pigmentosa 45, 613767
CNGB3	100%	100%	Achromatopsia 3, 262300
CNNM4	100%	100%	Jalili syndrome, 217080
COA8	100%	100%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COL11A1	100%	100%	Fibrochondrogenesis 1, 228520 Stickler syndrome, type II, 604841 Marshall syndrome, 154780 Deafness, autosomal dominant 37, 618533
COL17A1	100%	100%	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional 4, intermediate, 619787
COL18A1	100%	100%	Knobloch syndrome, type 1, 267750 Glaucoma, primary closed-angle, 618880
COL25A1	99%	99%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL2A1	100%	100%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 Czech dysplasia, 609162 Achondrogenesis, type II or hypochondrogenesis, 200610

			Spondyloperipheral dysplasia, 271700 SMED Strudwick type, 184250 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 SED congenita, 183900 Kniest dysplasia, 156550 Stickler syndrome, type I, nonsyndromic ocular, 609508 Osteoarthritis with mild chondrodysplasia, 604864 Stickler syndrome, type I, 108300 Platyspondylic skeletal dysplasia, Torrance type, 151210 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Avascular necrosis of the femoral head, 608805 Legg-Calve-Perthes disease, 150600
COL4A1	100%	100%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 Brain small vessel disease with or without ocular anomalies, 175780
COL8A2	100%	100%	Corneal dystrophy, posterior polymorphous 2, 609140 Corneal dystrophy, Fuchs endothelial, 1, 136800
COL9A1	100%	100%	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	100%	100%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284
COL9A3	100%	100%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 Stickler syndrome, type VI, 620022
COX7B	100%	100%	Linear skin defects with multiple congenital anomalies 2, 300887
CPAMD8	100%	100%	Anterior segment dysgenesis 8, 617319
CPLANE1	100%	100%	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CPSF1	100%	100%	Myopia 27, 618827
CRB1	100%	100%	Leber congenital amaurosis 8, 613835 Retinitis pigmentosa-12, 600105 Pigmented paravenous chorioretinal atrophy, 172870
CRX	100%	100%	Leber congenital amaurosis 7, 613829 Cone-rod retinal dystrophy-2, 120970
CRYAA	100%	100%	Cataract 9, multiple types, 604219
CRYAB	100%	100%	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 Myopathy, myofibrillar, 2, 608810

			Cataract 16, multiple types, 613763 Cardiomyopathy, dilated, 1II, 615184
CRYBA1	100%	100%	Cataract 10, multiple types, 600881
CRYBA2	100%	100%	?Cataract 42, 115900
CRYBA4	100%	100%	Cataract 23, 610425
CRYBB1	100%	100%	Cataract 17, multiple types, 611544
CRYBB2	100%	100%	Cataract 3, multiple types, 601547
CRYBB3	100%	100%	Cataract 22, 609741
CRYGB	100%	100%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	100%	100%	Cataract 2, multiple types, 604307
CRYGD	100%	100%	Cataract 4, multiple types, 115700
CRYGS	100%	100%	Cataract 20, multiple types, 116100
CSPP1	100%	100%	Joubert syndrome 21, 615636
CTDP1	100%	100%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA1	100%	100%	Macular dystrophy, patterned, 2, 608970
CTNNB1	100%	100%	Exudative vitreoretinopathy 7, 617572 Pilomatricoma, somatic, 132600 Colorectal cancer, somatic, 114500 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Medulloblastoma, somatic, 155255 Ovarian cancer, somatic, 167000 Hepatocellular carcinoma, somatic, 114550
CTSD	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSH	100%	100%	No OMIM disease ID
CWC27	100%	100%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CYP1B1	100%	100%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP4V2	100%	100%	Bietti crystalline corneoretinal dystrophy, 210370
DCN	95%	95%	Corneal dystrophy, congenital stromal, 610048
DCT	100%	100%	Oculocutaneous albinism, type VIII, 619165
DDHD1	100%	100%	Spastic paraplegia 28, autosomal recessive, 609340
DHDDS	94%	94%	Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861 Retinitis pigmentosa 59, 613861
DHX38	100%	100%	Retinitis pigmentosa 84, 618220
DKC1	100%	100%	Dyskeratosis congenita, X-linked, 305000

DNAJC30	100%	100%	Leber hereditary optic neuropathy, autosomal recessive, 619382
DNM1L	100%	100%	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNMBP	100%	100%	Cataract 48, 618415
DRAM2	100%	100%	Cone-rod dystrophy 21, 616502
DTNBP1	100%	100%	Hermansky-Pudlak syndrome 7, 614076
DYNC2H1	100%	99%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
WDR34	100%	100%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
EFEMP1	100%	100%	Doyne honeycomb degeneration of retina, 126600
ELOVL1	100%	100%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100%	100%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
EMC1	100%	100%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EPG5	100%	100%	Vici syndrome, 242840
EPHA2	100%	100%	Cataract 6, multiple types, 116600
ERCC2	100%	100%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100%	100%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC6	100%	100%	UV-sensitive syndrome 1, 600630 Cerebrooculofacioskeletal syndrome 1, 214150 ?De Sanctis-Cacchione syndrome, 278800 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946
ERCC8	100%	100%	UV-sensitive syndrome 2, 614621 Cockayne syndrome, type A, 216400
EXOSC2	100%	100%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EYA1	100%	100%	Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 602588 ?Otofaciocervical syndrome, 166780
EYS	100%	100%	Retinitis pigmentosa 25, 602772
FA2H	100%	100%	Spastic paraplegia 35, autosomal recessive, 612319
FAM161A	100%	100%	Retinitis pigmentosa 28, 606068

FBN1	100%	100%	Geleophysic dysplasia 2, 614185 Weill-Marchesani syndrome 2, dominant, 608328 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Acromicric dysplasia, 102370 Marfan syndrome, 154700 Stiff skin syndrome, 184900
FBXW11	100%	100%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FDXR	100%	100%	Auditory neuropathy and optic atrophy, 617717
FLVCR1	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOXC1	100%	100%	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXE3	100%	99%	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968
FREM1	100%	100%	Manitoba oculotrichoanal syndrome, 248450 Bifid nose with or without anorectal and renal anomalies, 608980 Trigonocephaly 2, 614485
FRMD7	100%	99%	Nystagmus, infantile periodic alternating, X-linked, 310700 Nystagmus 1, congenital, X-linked, 310700
FTL	100%	100%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FYCO1	100%	100%	Cataract 18, autosomal recessive, 610019
FZD4	100%	100%	Retinopathy of prematurity, 133780 Exudative vitreoretinopathy 1, 133780
GALK1	100%	100%	Galactokinase deficiency with cataracts, 230200
GALM	100%	100%	Galactosemia IV, 618881
GALT	100%	100%	Galactosemia, 230400
GCNT2	100%	100%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700
GDF3	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia, isolated, with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF6	100%	100%	Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Leber congenital amaurosis 17, 615360

			Multiple synostoses syndrome 4, 617898 Klippel-Feil syndrome 1, autosomal dominant, 118100
GDPD1	100%	100%	No OMIM disease ID
GFER	100%	100%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GJA1	100%	100%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Oculodentodigital dysplasia, 164200 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Atrioventricular septal defect 3, 600309
GJA3	100%	100%	Cataract 14, multiple types, 601885
GJA8	100%	100%	Cataract 1, multiple types, 116200
GNAT1	100%	100%	Night blindness, congenital stationary, autosomal dominant 3, 610444 Night blindness, congenital stationary, type 1G, 616389
GNAT2	100%	100%	Achromatopsia 4, 613856
GNB3	100%	100%	Night blindness, congenital stationary, type 1H, 617024
GNPTG	100%	100%	Mucolipidosis III gamma, 252605
GPR143	100%	100%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GPR179	100%	100%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GRHL2	100%	100%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
GRK1	100%	100%	Oguchi disease-2, 613411
GRM6	100%	100%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GSN	100%	100%	Amyloidosis, Finnish type, 105120
GUCA1A	100%	100%	Cone-rod dystrophy 14, 602093 Cone dystrophy-3, 602093
GUCA1B	100%	100%	Retinitis pigmentosa 48, 613827
GUCY2D	100%	100%	Cone-rod dystrophy 6, 601777 ?Choroidal dystrophy, central areolar 1, 215500 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1I, 618555
HARS1	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504

HCCS	100%	100%	Linear skin defects with multiple congenital anomalies 1, 309801
HGSNAT	92%	92%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HK1	100%	100%	Retinitis pigmentosa 79, 617460 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Hemolytic anemia due to hexokinase deficiency, 235700
HKDC1	100%	100%	Retinitis pigmentosa 92, 619614
HMX1	100%	100%	Oculoauricular syndrome, 612109
HPS1	100%	100%	Hermansky-Pudlak syndrome 1, 203300
HPS3	100%	100%	Hermansky-Pudlak syndrome 3, 614072
HPS4	100%	100%	Hermansky-Pudlak syndrome 4, 614073
HPS5	100%	100%	Hermansky-Pudlak syndrome 5, 614074
HPS6	100%	100%	Hermansky-Pudlak syndrome 6, 614075
HRAS	100%	100%	Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Congenital myopathy with excess of muscle spindles, 218040 Nevus sebaceous or woolly hair nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HSF4	100%	100%	Cataract 5, multiple types, 116800
HSPG2	100%	100%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
IDH3A	100%	100%	Retinitis pigmentosa 90, 619007
IDH3B	100%	100%	Retinitis pigmentosa 46, 612572
IFT140	100%	100%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	100%	100%	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100%	100%	Bardet-Biedl syndrome 19, 615996
IFT43	100%	100%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	100%	100%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102

IFT74	100%	100%	Bardet-Biedl syndrome 22, 617119 Spermatogenic failure 58, 619585 Joubert syndrome 40, 619582
IFT81	95%	95%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGSF3	100%	100%	?Lacrimal duct defect, 149700
IKBKG	100%	98%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636 Autoinflammatory disease, systemic, X-linked, 301081
IMPDH1	100%	100%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
IMPG1	100%	100%	Macular dystrophy, vitelliform, 4, 616151 Retinitis pigmentosa 91, 153870
IMPG2	100%	100%	Retinitis pigmentosa 56, 613581 Macular dystrophy, vitelliform, 5, 616152
INPP5E	100%	100%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	100%	100%	Nephronophthisis 2, infantile, 602088
IQCB1	100%	100%	Senior-Loken syndrome 5, 609254
IRX1	100%	100%	No OMIM disease ID
ITM2B	100%	100%	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 Dementia, familial British, 176500 Dementia, familial Danish, 117300
ITPR1	100%	100%	Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658
JAG1	100%	100%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JAM3	100%	100%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KCNJ13	100%	100%	Snowflake vitreoretinal degeneration, 193230 Leber congenital amaurosis 16, 614186
KCNV2	100%	100%	Retinal cone dystrophy 3B, 610356
KERA	100%	100%	Cornea plana 2, autosomal recessive, 217300
KIAA0586	96%	96%	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490

KIAA0753	100%	100%	?Orofaciodigital syndrome XV, 617127 ?Joubert syndrome 38, 619476 Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIAA1549	100%	100%	Retinitis pigmentosa 86, 618613
KIF11	100%	100%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF21A	100%	100%	Fibrosis of extraocular muscles, congenital, 3B, 135700 Fibrosis of extraocular muscles, congenital, 1, 135700
KIF3B	100%	100%	Retinitis pigmentosa 89, 618955
KIF7	100%	100%	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
KIZ	100%	100%	Retinitis pigmentosa 69, 615780
KLHL7	100%	100%	Retinitis pigmentosa 42, 612943 PERCHING syndrome, 617055
KRT12	100%	100%	Meesmann corneal dystrophy 1, 122100
KRT3	100%	100%	Meesmann corneal dystrophy 2, 618767
LAMA1	100%	100%	Poretti-Boltshauser syndrome, 615960
LAMB2	100%	100%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMP2	100%	100%	Danon disease, 300257
LCA5	100%	100%	Leber congenital amaurosis 5, 604537
LEMD2	100%	100%	Marbach-Rustad progeroid syndrome, 619322 Cataract 46, juvenile-onset, 212500
LIM2	100%	100%	Cataract 19, multiple types, 615277
LMX1B	100%	100%	Focal segmental glomerulosclerosis 10, 256020 Nail-patella syndrome, 161200
LOXL3	100%	100%	Myopia 28, autosomal recessive, 619781
LRAT	100%	100%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRIT3	100%	100%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	98%	98%	Albinism, oculocutaneous, type VII, 615179
LRP2	100%	100%	Donnai-Barrow syndrome, 222448
LRP5	100%	100%	Osteopetrosis, autosomal dominant 1, 607634 Hyperostosis, endosteal, 144750

			Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Exudative vitreoretinopathy 4, 601813 van Buchem disease, type 2, 607636
LRPAP1	100%	100%	Myopia 23, autosomal recessive, 615431
LSS	100%	100%	Hypotrichosis 14, 618275 Cataract 44, 616509 Alopecia-intellectual disability syndrome 4, 618840
LTBP2	100%	100%	Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 ?Weill-Marchesani syndrome 3, recessive, 614819
LYST	100%	100%	Chediak-Higashi syndrome, 214500
LZTFL1	100%	100%	Bardet-Biedl syndrome 17, 615994
MAB21L2	100%	100%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAF	94%	90%	Cataract 21, multiple types, 610202 Ayme-Gripp syndrome, 601088
MAFB	100%	100%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAK	100%	100%	Retinitis pigmentosa 62, 614181
MAPKAPK3	100%	100%	?Macular dystrophy, patterned, 3, 617111
MCAT	100%	100%	No OMIM disease ID
MERTK	99%	99%	Retinitis pigmentosa 38, 613862
MFN2	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	100%	100%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	100%	100%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MIP	100%	100%	Cataract 15, multiple types, 615274
MIR184	NC	NC	EDICT syndrome, 614303
MIR204	NC	NC	?Retinal dystrophy and iris coloboma with or without cataract, 616722
MITF	100%	100%	Waardenburg syndrome, type 2A, 193510 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome/ocular albinism, digenic, 103470 COMMAD syndrome, 617306

MKKS	100%	100%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKS1	100%	100%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
C12orf65	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
MVK	90%	90%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYO5A	100%	100%	Griscelli syndrome, type 1, 214450
MYO7A	100%	100%	Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900 Deafness, autosomal dominant 11, 601317
MYOC	100%	100%	Glaucoma 1A, primary open angle, 137750
NAA10	100%	100%	Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NBAS	100%	100%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NDP	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDUFB11	100%	98%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFS2	100%	100%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NEK1	100%	100%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK2	96%	96%	?Retinitis pigmentosa 67, 615565
NEUROD1	100%	100%	Maturity-onset diabetes of the young 6, 606394
NHS	100%	100%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NMNAT1	100%	98%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 Leber congenital amaurosis 9, 608553
NPHP1	100%	100%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900

NPHP3	100%	100%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	100%	100%	Senior-Loken syndrome 4, 606996 Nephronophthisis 4, 606966
NR2E3	100%	100%	Retinitis pigmentosa 37, 611131 Enhanced S-cone syndrome, 268100
NR2F1	100%	100%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRL	100%	100%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type,
NYX	100%	100%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	100%	100%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCA2	100%	100%	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200
OCRL	100%	100%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	100%	100%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
OPA1	100%	100%	Optic atrophy plus syndrome, 125250 Optic atrophy 1, 165500 Behr syndrome, 210000 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPN1LW	95%	94%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	98%	95%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700
OPTN	100%	100%	Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435
OTX2	100%	100%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 Pituitary hormone deficiency, combined, 6, 613986 Microphthalmia, syndromic 5, 610125
OVOL2	100%	100%	Corneal dystrophy, posterior polymorphous, 1, 122000
P3H2	100%	100%	Myopia, high, with cataract and vitreoretinal degeneration, 614292

P4HA2	100%	100%	Myopia 25, autosomal dominant, 617238
PAK2	100%	100%	?Knobloch syndrome 2, 618458
PANK2	100%	100%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PANK4	100%	100%	?Cataract 49, 619593
PAX2	100%	100%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX6	100%	100%	Optic nerve hypoplasia, 165550 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma, ocular, 120200 ?Coloboma of optic nerve, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 ?Morning glory disc anomaly, 120430 Foveal hypoplasia 1, 136520 Keratitis, 148190
PCARE	100%	100%	Retinitis pigmentosa 54, 613428
PCDH15	100%	100%	Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1F, 602083
PCYT1A	100%	100%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE6A	100%	100%	Retinitis pigmentosa 43, 613810
PDE6B	100%	100%	Retinitis pigmentosa-40, 613801 Night blindness, congenital stationary, autosomal dominant 2, 163500
PDE6C	100%	100%	Cone dystrophy 4, 613093
PDE6D	100%	100%	Joubert syndrome 22, 615665
PDE6G	100%	100%	Retinitis pigmentosa 57, 613582
PDE6H	100%	100%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDGFRA	100%	100%	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDZD7	100%	99%	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PET100	100%	100%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	100%	100%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100

PEX2	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	100%	100%	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX6	100%	100%	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PEX7	91%	91%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PGK1	100%	100%	Phosphoglycerate kinase 1 deficiency, 300653
PHOX2A	100%	100%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	100%	100%	Refsum disease, 266500
PIKFYVE	100%	100%	Corneal fleck dystrophy, 121850
PITX2	100%	100%	Ring dermoid of cornea, 180550 Axenfeld-Rieger syndrome, type 1, 180500 Anterior segment dysgenesis 4, 137600
PITX3	100%	100%	Cataract 11, multiple types, 610623 Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, syndromic, autosomal recessive, 610623
PLA2G5	100%	100%	No OMIM disease ID
PLK4	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PNPLA6	100%	100%	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470
POC1B	100%	100%	Cone-rod dystrophy 20, 615973
POC5	100%	100%	No OMIM disease ID
POLG2	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425
POMGNT1	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
PPT1	90%	90%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	100%	100%	Retinitis pigmentosa 36, 610599

PRDM13	100%	100%	Pontocerebellar hypoplasia, type 17, 619909 Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761
PRDM5	100%	100%	Brittle cornea syndrome 2, 614170
PRDX3	100%	100%	Spinocerebellar ataxia, autosomal recessive 32, 619862 Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871
PRIMPOL	100%	100%	Myopia 22, autosomal dominant, 615420
PROM1	100%	100%	Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657
PRPF3	100%	100%	Retinitis pigmentosa 18, 601414
PRPF31	100%	100%	Retinitis pigmentosa 11, 600138
PRPF4	100%	100%	Retinitis pigmentosa 70, 615922
PRPF6	100%	100%	Retinitis pigmentosa 60, 613983
PRPF8	100%	100%	Retinitis pigmentosa 13, 600059
PRPH2	100%	100%	Macular dystrophy, patterned, 1, 169150 Choroidal dystrophy, central areolar 2, 613105 Retinitis punctata albescens, 136880 Leber congenital amaurosis 18, 608133 Macular dystrophy, vitelliform, 3, 608161 Retinitis pigmentosa 7 and digenic form, 608133
PRR11	100%	100%	No OMIM disease ID
PRR12	100%	100%	Neuroocular syndrome, 619539
PRSS56	100%	100%	Microphthalmia, isolated 6, 613517
PTCHD1	100%	100%	No OMIM disease ID
PXDN	100%	100%	Anterior segment dysgenesis 7, with sclerocornea, 269400
RAB28	100%	100%	Cone-rod dystrophy 18, 615374
RAB3GAP2	100%	100%	Martsolf syndrome 1, 212720 Warburg micro syndrome 2, 614225
RARB	100%	100%	Microphthalmia, syndromic 12, 615524
RAX	100%	100%	Microphthalmia, syndromic 16, 611038
RAX2	100%	100%	Retinitis pigmentosa 95, 620102 Cone-rod dystrophy 11, 610381 ?Macular degeneration, age-related, 6, 613757
RBP3	100%	100%	?Retinitis pigmentosa 66, 615233

RBP4	100%	100%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	100%	100%	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	100%	100%	Leber congenital amaurosis 12, 610612
RDH11	100%	100%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	100%	100%	Leber congenital amaurosis 13, 612712
RDH5	100%	100%	Fundus albipunctatus, 136880
REEP6	100%	100%	Retinitis pigmentosa 77, 617304
RGS9	100%	100%	Bradyopsia, 608415
RGS9BP	100%	100%	Bradyopsia, 608415
RHO	100%	100%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
DDX58	100%	100%	Singleton-Merten syndrome 2, 616298
RIMS1	100%	100%	Cone-rod dystrophy 7, 603649
RIMS2	100%	100%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RLBP1	100%	100%	Bothnia retinal dystrophy, 607475 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880 Fundus albipunctatus, 136880
ROM1	100%	100%	Retinitis pigmentosa 7, digenic form, 608133
RP1	100%	100%	Retinitis pigmentosa 1, 180100
RP1L1	100%	100%	Occult macular dystrophy, 613587 Retinitis pigmentosa 88, 618826
RP2	100%	100%	Retinitis pigmentosa 2, 312600
RP9	100%	100%	?Retinitis pigmentosa 9, 180104
RPE65	100%	100%	Retinitis pigmentosa 20, 613794 Retinitis pigmentosa 87 with choroidal involvement, 618697 Leber congenital amaurosis 2, 204100
RPGR	99%	95%	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Cone-rod dystrophy, X-linked, 1, 304020 Retinitis pigmentosa 3, 300029 Macular degeneration, X-linked atrophic, 300834
RPGRIP1	100%	100%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826

RPGRIP1L	100%	100%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RS1	100%	100%	Retinoschisis, 312700
RTN4IP1	100%	100%	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732
SAG	100%	100%	Retinitis pigmentosa 47, 613758 Oguchi disease-1, 258100
SAMD11	100%	100%	No OMIM disease ID
SC5D	100%	100%	Lathosterolosis, 607330
SCAPER	100%	100%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCO2	100%	100%	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SDCCAG8	100%	100%	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993
SEMA4A	100%	100%	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283
SGSH	100%	100%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3BP2	100%	99%	Cherubism, 118400
SHH	100%	100%	Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SIL1	100%	100%	Marinesco-Sjogren syndrome, 248800
SIPA1L3	100%	100%	?Cataract 45, 616851
SIX6	100%	100%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SLC16A12	100%	100%	Cataract 47, juvenile, with microcornea, 612018
SLC24A1	100%	100%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	100%	100%	Albinism, oculocutaneous, type VI, 113750
SLC25A46	100%	100%	Neuropathy, hereditary motor and sensory, type VIB, 616505 Pontocerebellar hypoplasia, type 1E, 619303
SLC33A1	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC37A3	100%	100%	No OMIM disease ID
SLC38A8	100%	100%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A12	100%	100%	No OMIM disease ID
SLC39A5	100%	100%	Myopia 24, autosomal dominant, 615946

SLC45A2	100%	100%	Albinism, oculocutaneous, type IV, 606574
SLC4A11	100%	100%	Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A7	100%	100%	No OMIM disease ID
SLC52A2	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC66A1	100%	100%	No OMIM disease ID
SLC7A14	100%	100%	Retinitis pigmentosa 68, 615725
SMG8	100%	100%	Alzahrani-Kuwahara syndrome, 619268
SMOC1	100%	100%	Microphthalmia with limb anomalies, 206920
SNRNP200	100%	100%	Retinitis pigmentosa 33, 610359
SOX2	100%	100%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900
SOX5	100%	100%	Lamb-Shaffer syndrome, 616803
SPATA7	100%	100%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232
SPG7	100%	100%	Spastic paraplegia 7, autosomal recessive, 607259
SPP2	100%	100%	No OMIM disease ID
SSBP1	100%	100%	Optic atrophy 13 with retinal and foveal abnormalities, 165510
STRA6	100%	100%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STX3	100%	100%	Retinal dystrophy and microvillus inclusion disease, 619446 Diarrhea 12, with microvillus atrophy, 619445
TACSTD2	100%	100%	Corneal dystrophy, gelatinous drop-like, 204870
TBC1D2B	100%	100%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323
TCTN1	95%	94%	Joubert syndrome 13, 614173
TCTN2	100%	100%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100%	100%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDRD7	100%	100%	Cataract 36, 613887
TEAD1	100%	100%	Sveinsson chorioretinal atrophy, 108985
TEK	100%	100%	Venous malformations, multiple cutaneous and mucosal, 600195 Glaucoma 3, primary congenital, E, 617272
TENM3	100%	100%	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145

TFPT	100%	100%	No OMIM disease ID
TGFBI	100%	100%	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471
TIMM8A	100%	100%	Mohr-Tranebjaerg syndrome, 304700
TIMP3	100%	100%	Sorsby fundus dystrophy, 136900
TLCD3B	100%	100%	Cone-rod dystrophy 22, 619531
TMCO3	100%	100%	No OMIM disease ID
TMEM126A	100%	100%	Optic atrophy 7, 612989
TMEM138	100%	100%	Joubert syndrome 16, 614465
TMEM216	100%	100%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM218	100%	100%	Joubert syndrome 39, 619562
TMEM231	100%	100%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100%	100%	Joubert syndrome 14, 614424
TMEM67	100%	98%	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TMEM98	100%	100%	Nanophthalmos 4, 615972
TOGARAM1	100%	100%	Joubert syndrome 37, 619185
TOPORS	100%	100%	Retinitis pigmentosa 31, 609923
TPP1	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAF3IP1	100%	100%	Senior-Loken syndrome 9, 616629
TREX1	100%	100%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM32	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110

TRNT1	100%	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM1	100%	100%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TSPAN12	100%	100%	Exudative vitreoretinopathy 5, 613310
TTC8	100%	100%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTLL5	100%	100%	Cone-rod dystrophy 19, 615860
TUB	100%	100%	?Retinal dystrophy and obesity, 616188
TUBA3D	100%	100%	Keratoconus 9, 617928
TUBB3	100%	100%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4B	100%	100%	Leber congenital amaurosis with early-onset deafness, 617879
TUBGCP4	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TULP1	100%	100%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TWNK	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TYR	100%	100%	Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IA, 203100
TYRP1	100%	100%	Albinism, oculocutaneous, type III, 203290
UBIAD1	100%	100%	Corneal dystrophy, Schnyder type, 121800
UNC45B	100%	100%	?Cataract 43, 616279 Myofibrillar myopathy 11, 619178
USH1C	100%	100%	Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH1G	100%	100%	Usher syndrome, type 1G, 606943
USH2A	100%	100%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809
USP45	100%	100%	?Leber congenital amaurosis 19, 618513
VAX1	100%	99%	?Microphthalmia, syndromic 11, 614402
VCAN	100%	100%	Wagner syndrome 1, 143200
VIM	100%	100%	Cataract 30, pulverulent, 116300
VPS13B	100%	99%	Cohen syndrome, 216550

VSX1	100%	100%	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300
VSX2	100%	100%	Microphthalmia, isolated 2, 610093 Microphthalmia with coloboma 3, 610092
WDPCP	98%	97%	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	100%	100%	Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378 Senior-Loken syndrome 8, 616307 Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Spermatogenic failure 72, 619867
WDR36	100%	100%	Glaucoma 1, open angle, G, 609887
WDR73	100%	100%	Galloway-Mowat syndrome 1, 251300
WFS1	100%	100%	Deafness, autosomal dominant 6/14/38, 600965 ?Cataract 41, 116400 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	100%	100%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WRN	100%	100%	Werner syndrome, 277700
YAP1	100%	100%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433
YARS1	100%	100%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418 Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YME1L1	100%	100%	?Optic atrophy 11, 617302
YPEL2	100%	100%	No OMIM disease ID
ZEB1	100%	99%	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270
ZNF408	100%	100%	Retinitis pigmentosa 72, 616469 ?Exudative vitreoretinopathy 6, 616468
ZNF423	100%	100%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF469	100%	100%	Brittle cornea syndrome 1, 229200
ZNF513	100%	100%	?Retinitis pigmentosa 58, 613617
ZNF644	100%	100%	Myopia 21, autosomal dominant, 614167

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