VISION DISORDERS GENE PANEL DG 2.17 (432 genes)

Releasedate: 06-12-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ABCA4	109.9	99.9%	99.1%	Retinal dystrophy, early-onset severe, 248200 Stargardt disease 1, 248200 Fundus flavimaculatus, 248200 Cone-rod dystrophy 3, 604116 Retinitis pigmentosa 19, 601718
ABCC6	116.6	93.7%	93.1%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABHD12	96.9	100.0%	99.5%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACBD5	144.7	99.8%	98.1%	No OMIM Disease ID
ACO2	125.5	95.6%	90.3%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ADAM9	140.7	99.9%	98.1%	Cone-rod dystrophy 9, 612775
ADAMTS18	134.6	100.0%	99.8%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTSL4	137.7	100.0%	99.8%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADGRV1	126.0	99.8%	98.3%	Usher syndrome, type 2C, 605472 ?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
ADIPOR1	91.6	99.5%	95.6%	No OMIM Disease ID
AGBL1	109.9	98.5%	98.3%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	111.4	100.0%	99.4%	Retinitis pigmentosa 75, 617023
AGK	109.6	99.6%	95.5%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AHI1	125.5	99.9%	97.6%	Joubert syndrome 3, 608629
AHR	181.2	100.0%	99.5%	?Retinitis pigmentosa 85, 618345
AIPL1	124.2	100.0%	100.0%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
ALDH1A3	110.3	100.0%	98.4%	Microphthalmia, isolated 8, 615113

ALMS1	178.0	100.0%	99.8%	Alstrom syndrome, 203800
AP3B1	108.2	99.4%	95.7%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	135.1	98.5%	97.9%	?Hermansky-Pudlak syndrome 10, 617050
APOPT1	80.4	82.1%	82.1%	Mitochondrial complex IV deficiency, 220110
ARHGEF18	153.5	99.9%	98.7%	Retinitis pigmentosa 78, 617433
ARL13B	98.7	100.0%	99.7%	Joubert syndrome 8, 612291
ARL2BP	66.5	92.9%	83.8%	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	76.1	99.9%	97.6%	Joubert syndrome 35, 618161 Retinitis pigmentosa 83, 618173
ARL6	91.8	99.9%	97.7%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARR3	87.8	99.9%	99.6%	Myopia 26, X-linked, female-limited, 301010
ARSG	120.4	100.0%	98.7%	Usher syndrome, type IV, 618144
ASPH	109.3	100.0%	98.9%	Traboulsi syndrome, 601552
ASRGL1	129.9	100.0%	100.0%	No OMIM Disease ID
ATF6	125.0	100.0%	99.4%	Achromatopsia 7, 616517
ATOH7	210.8	99.8%	99.2%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
B3GLCT	97.3	100.0%	99.7%	Peters-plus syndrome, 261540
BBIP1	116.1	98.5%	91.4%	?Bardet-Biedl syndrome 18, 615995
BBS1	156.1	100.0%	100.0%	Bardet-Biedl syndrome 1, 209900
BBS10	156.7	100.0%	100.0%	Bardet-Biedl syndrome 10, 615987
BBS12	193.6	100.0%	100.0%	Bardet-Biedl syndrome 12, 615989
BBS2	153.3	100.0%	99.7%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	113.2	100.0%	98.4%	Bardet-Biedl syndrome 4, 615982
BBS5	94.9	98.4%	92.3%	Bardet-Biedl syndrome 5, 615983
BBS7	136.8	99.0%	95.3%	Bardet-Biedl syndrome 7, 615984
BBS9	113.2	98.8%	94.8%	Bardet-Biedl syndrome 9, 615986
BCOR	109.0	99.2%	96.2%	Microphthalmia, syndromic 2, 300166
BEST1	137.3	99.9%	98.2%	Retinitis pigmentosa-50, 613194 Bestrophinopathy, autosomal recessive, 611809 Retinitis pigmentosa, concentric, 613194 Vitreoretinochoroidopathy, 193220 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Macular dystrophy, vitelliform, 2, 153700
BFSP1	105.8	100.0%	99.3%	Cataract 33, multiple types, 611391
BFSP2	106.9	100.0%	99.2%	Cataract 12, multiple types, 611597

BLOC1S3	79.6	100.0%	100.0%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	103.0	99.3%	92.1%	?Hermansky-pudlak syndrome 9, 614171
BMP4	192.0	100.0%	100.0%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C12orf65	112.4	100.0%	99.8%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C19orf12	117.5	100.0%	99.9%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1QTNF5	173.0	97.8%	91.6%	Retinal degeneration, late-onset, autosomal dominant, 605670
C21orf2	146.9	100.0%	99.4%	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
C2orf71	128.5	100.0%	99.3%	Retinitis pigmentosa 54, 613428
C5orf42	122.3	99.7%	97.4%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C8orf37	146.4	100.0%	99.9%	Retinitis pigmentosa 64, 614500 Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500
CA4	175.5	100.0%	100.0%	Retinitis pigmentosa 17, 600852
CABP4	168.9	100.0%	100.0%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1F	91.4	99.8%	97.7%	Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Aland Island eye disease, 300600
CACNA2D4	103.4	99.2%	97.4%	Retinal cone dystrophy 4, 610478
CAPN5	167.1	100.0%	100.0%	Vitreoretinopathy, neovascular inflammatory, 193235
CC2D2A	112.6	99.0%	97.0%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CCT2	145.5	100.0%	99.9%	No OMIM Disease ID
CDH23	186.7	100.0%	100.0%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1D, 601067
CDH3	148.2	100.0%	99.9%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	153.8	99.9%	99.2%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK10	141.9	100.0%	100.0%	Al Kaissi syndrome, 617694
CEP164	96.7	99.9%	98.4%	Nephronophthisis 15, 614845

CEP250	108.0	99.9%	99.2%	Cone-rod dystrophy and hearing loss 2, 618358
CEP290	77.6	96.9%	88.7%	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CEP41	79.1	98.7%	94.4%	Joubert syndrome 15, 614464
CEP78	123.1	99.8%	96.9%	Cone-rod dystrophy and hearing loss, 617236
CEP83	103.3	99.8%	96.2%	Nephronophthisis 18, 615862
CERKL	115.2	99.5%	96.8%	Retinitis pigmentosa 26, 608380
CFH	148.8	99.4%	97.4%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CHD7	143.6	100.0%	99.5%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHM	98.7	98.3%	91.4%	Choroideremia, 303100
CHMP4B	150.6	100.0%	98.5%	Cataract 31, multiple types, 605387
CHRDL1	89.5	100.0%	99.1%	Megalocornea 1, X-linked, 309300
CHST6	322.9	100.0%	100.0%	Macular corneal dystrophy, 217800
CIB2	218.3	100.0%	99.5%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CISD2	116.7	83.4%	83.4%	Wolfram syndrome 2, 604928
CLCC1	100.3	99.9%	98.5%	No OMIM Disease ID
CLN3	123.4	92.5%	92.2%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	139.4	100.0%	99.5%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	141.7	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	156.2	83.5%	83.5%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLRN1	140.6	100.0%	99.5%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CLUAP1	138.4	100.0%	99.7%	No OMIM Disease ID
CNGA1	110.6	93.0%	86.9%	Retinitis pigmentosa 49, 613756
CNGA3	160.9	100.0%	99.8%	Achromatopsia 2, 216900
CNGB1	116.9	99.7%	98.3%	Retinitis pigmentosa 45, 613767
CNGB3	101.6	98.4%	93.7%	Macular degeneration, juvenile, 248200 Achromatopsia 3, 262300
CNNM4	177.3	100.0%	99.9%	Jalili syndrome, 217080

COL11A1	94.6	98.0%	93.6%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 Fibrochondrogenesis 1, 228520
COL18A1	153.2	99.7%	97.9%	Knobloch syndrome, type 1, 267750
COL25A1	134.2	99.3%	98.7%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL2A1	121.1	100.0%	99.8%	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805 SED congenita, 183900 Legg-Calve-Perthes disease, 150600 SMED Strudwick type, 184250 Czech dysplasia, 609162 Stickler sydrome, type I, nonsyndromic ocular, 609508 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL8A2	140.0	100.0%	100.0%	Corneal dystrophy, posterior polymorphous 2, 609140 Corneal dystrophy, Fuchs endothelial, 1, 136800
COL9A1	132.9	100.0%	99.5%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	104.9	100.0%	99.6%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
CRB1	158.9	100.0%	100.0%	Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105 Leber congenital amaurosis 8, 613835
CRX	216.9	100.0%	100.0%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	145.4	96.7%	91.1%	Cataract 9, multiple types, 604219
CRYAB	96.3	99.9%	97.9%	Myopathy, myofibrillar, 2, 608810 Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CRYBA1	108.4	100.0%	99.8%	Cataract 10, multiple types, 600881

CRYBA2	183.6	100.0%	100.0%	?Cataract 42, 115900
CRYBA4	126.6	100.0%	100.0%	Cataract 23, 610425
CRYBB1	132.7	100.0%	99.9%	Cataract 17, multiple types, 611544
CRYBB2	148.3	100.0%	100.0%	Cataract 3, multiple types, 601547
CRYBB3	149.6	100.0%	100.0%	Cataract 22, 609741
CRYGB	111.5	100.0%	98.3%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	138.2	100.0%	99.7%	Cataract 2, multiple types, 604307
CRYGD	112.7	100.0%	99.7%	Cataract 4, multiple types, 115700
CRYGS	84.2	94.4%	85.4%	Cataract 20, multiple types, 116100
CSPP1	117.4	100.0%	99.4%	Joubert syndrome 21, 615636
CTDP1	141.7	96.2%	88.2%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA1	113.7	99.4%	97.7%	Macular dystrophy, patterned, 2, 608970
CTNNB1	129.5	100.0%	100.0%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 Medulloblastoma, somatic, 155255 Hepatocellular carcinoma, somatic, 114550 Pilomatricoma, somatic, 132600 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Exudative vitreoretinopathy 7, 617572
CTSD	187.3	100.0%	99.0%	Ceroid lipofuscinosis, neuronal, 10, 610127
CWC27	82.5	99.8%	97.3%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CYP1B1	153.6	100.0%	100.0%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP4V2	140.6	99.8%	98.4%	Bietti crystalline corneoretinal dystrophy, 210370
DCN	131.5	95.7%	95.3%	Corneal dystrophy, congenital stromal, 610048
DDHD1	166.9	100.0%	99.1%	Spastic paraplegia 28, autosomal recessive, 609340
DHDDS	84.5	97.3%	94.0%	Retinitis pigmentosa 59, 613861 ?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836
DHX38	112.7	100.0%	99.5%	Retinitis pigmentosa 84, 618220
DKC1	93.9	99.7%	98.0%	Dyskeratosis congenita, X-linked, 305000
DNM1L	120.8	99.9%	97.7%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DRAM2	127.1	100.0%	100.0%	Cone-rod dystrophy 21, 616502
DTNBP1	118.8	99.8%	97.5%	Hermansky-Pudlak syndrome 7, 614076
EFEMP1	152.0	100.0%	99.5%	Doyne honeycomb degeneration of retina, 126600
ELOVL1	90.9	99.7%	96.6%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527

ELOVL4	103.3	100.0%	99.6%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
EMC1	111.0	100.0%	99.2%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EPG5	111.4	99.5%	98.3%	Vici syndrome, 242840
EPHA2	173.3	100.0%	99.9%	Cataract 6, multiple types, 116600
EXOSC2	114.1	100.0%	99.9%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EYA1	121.6	100.0%	99.9%	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Branchiootic syndrome 1, 602588
EYS	130.0	99.8%	97.3%	Retinitis pigmentosa 25, 602772
FA2H	101.5	99.3%	95.1%	Spastic paraplegia 35, autosomal recessive, 612319
FAM161A	142.0	99.9%	99.3%	Retinitis pigmentosa 28, 606068
FLVCR1	154.8	100.0%	99.4%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOXC1	96.1	99.9%	99.2%	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXE3	111.7	93.8%	87.3%	Cataract 34, multiple types, 612968 Anterior segment dysgenesis 2, multiple subtypes, 610256
FREM1	112.5	99.8%	98.8%	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980
FRMD7	101.2	99.9%	97.9%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FTL	164.3	100.0%	98.4%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
FYCO1	135.2	100.0%	100.0%	Cataract 18, autosomal recessive, 610019
FZD4	192.1	100.0%	100.0%	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
GALK1	186.1	100.0%	99.9%	Galactokinase deficiency with cataracts, 230200
GALT	165.3	100.0%	100.0%	Galactosemia, 230400
GCNT2	158.5	99.5%	99.5%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700
GDF3	132.5	100.0%	100.0%	Microphthalmia, isolated 7, 613704 Microphthalmia with coloboma 6, 613703 Klippel-Feil syndrome 3, autosomal dominant, 613702

GDF6	180.6	100.0%	100.0%	Leber congenital amaurosis 17, 615360 Klippel-Feil syndrome 1, autosomal dominant, 118100 Multiple synostoses syndrome 4, 617898 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GFER	103.0	100.0%	99.9%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GJA1	162.4	100.0%	100.0%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJA3	185.2	100.0%	100.0%	Cataract 14, multiple types, 601885
GJA8	173.2	100.0%	100.0%	Cataract 1, multiple types, 116200
GNAT1	197.4	100.0%	100.0%	Night blindness, congenital stationary, type 1G, 616389 Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	106.3	100.0%	98.5%	Achromatopsia 4, 613856
GNB3	159.3	100.0%	100.0%	Night blindness, congenital stationary, type 1H, 617024
GNPTG	199.0	99.9%	99.4%	Mucolipidosis III gamma, 252605
GPR143	60.8	91.8%	81.6%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GPR179	163.2	100.0%	100.0%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GRHL2	119.8	100.0%	100.0%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GRK1	149.0	100.0%	100.0%	Oguchi disease-2, 613411
GRM6	152.8	99.2%	94.6%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GSN	123.5	95.6%	93.8%	Amyloidosis, Finnish type, 105120
GUCA1A	189.2	100.0%	100.0%	Cone-rod dystrophy 14, 602093 Cone dystrophy-3, 602093
GUCA1B	135.9	100.0%	100.0%	Retinitis pigmentosa 48, 613827
GUCY2D	120.6	100.0%	100.0%	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1I, 618555 ?Choroidal dystrophy, central areolar 1, 215500

HARS	142.4	100.0%	100.0%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HCCS	90.4	99.6%	96.6%	Linear skin defects with multiple congenital anomalies 1, 309801
HGSNAT	99.9	88.2%	86.3%	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HK1	123.7	100.0%	99.6%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Retinitis pigmentosa 79, 617460
HMX1	47.2	89.3%	70.6%	Oculoauricular syndrome, 612109
HPS1	125.7	100.0%	100.0%	Hermansky-Pudlak syndrome 1, 203300
HPS3	133.9	99.9%	98.2%	Hermansky-Pudlak syndrome 3, 614072
HPS4	135.2	100.0%	100.0%	Hermansky-Pudlak syndrome 4, 614073
HPS5	122.5	99.9%	98.9%	Hermansky-Pudlak syndrome 5, 614074
HPS6	183.5	100.0%	99.2%	Hermansky-Pudlak syndrome 6, 614075
HRAS	196.0	100.0%	100.0%	Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HSF4	166.2	99.9%	99.3%	Cataract 5, multiple types, 116800
HSPG2	132.7	99.5%	99.2%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
IDH3B	136.5	95.8%	95.4%	Retinitis pigmentosa 46, 612572
IFT140	127.6	100.0%	99.6%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	98.4	100.0%	99.5%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	122.9	100.0%	99.9%	?Bardet-Biedl syndrome 19, 615996
IFT43	119.5	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IFT74	81.5	99.4%	93.7%	?Bardet-Biedl syndrome 20, 617119
IFT81	90.5	93.0%	88.0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IMPDH1	57.4	97.6%	87.3%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105

IMPG1	92.5	99.8%	98.2%	Macular dystrophy, vitelliform, 4, 616151
IMPG2	127.1	99.5%	98.0%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INPP5E	131.1	100.0%	99.3%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INVS	147.7	100.0%	100.0%	Nephronophthisis 2, infantile, 602088
IQCB1	90.6	91.0%	79.0%	Senior-Loken syndrome 5, 609254
IRX1	175.4	99.1%	93.2%	No OMIM Disease ID
ITPR1	136.4	100.0%	99.8%	Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658 Gillespie syndrome, 206700
JAG1	143.4	99.4%	97.6%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
JAM3	132.0	100.0%	100.0%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KCNJ13	142.5	100.0%	100.0%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNV2	154.7	100.0%	100.0%	Retinal cone dystrophy 3B, 610356
KERA	174.8	100.0%	100.0%	Cornea plana 2, autosomal recessive, 217300
KIAA1549	125.5	98.7%	97.6%	Retinitis pigmentosa 86, 618613
KIF11	89.7	97.6%	94.9%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF21A	118.6	99.9%	98.8%	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF7	120.4	99.3%	96.6%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KIZ	151.3	99.8%	97.7%	Retinitis pigmentosa 69, 615780
KLHL7	116.8	100.0%	99.9%	Retinitis pigmentosa 42, 612943 Cold-induced sweating syndrome 3, 617055
KRT12	142.5	99.8%	97.9%	Meesmann corneal dystrophy, 122100
KRT3	125.0	100.0%	99.9%	Meesmann corneal dystrophy, 122100
LAMA1	119.9	100.0%	99.6%	Poretti-Boltshauser syndrome, 615960
LAMB2	182.1	100.0%	99.7%	Pierson syndrome, 609049 Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199
LCA5	137.1	99.9%	98.9%	Leber congenital amaurosis 5, 604537
LEMD2	117.7	100.0%	99.5%	Cataract 46, juvenile-onset, 212500

LIM2	122.5	100.0%	99.9%	Cataract 19, multiple types, 615277
LRAT	252.8	100.0%	100.0%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LRIT3	111.2	94.4%	93.5%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	119.8	99.4%	98.0%	Albinism, oculocutaneous, type VII, 615179
LRP2	140.5	100.0%	99.9%	Donnai-Barrow syndrome, 222448
LRP5	183.1	99.9%	99.4%	van Buchem disease, type 2, 607636 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Osteopetrosis, autosomal dominant 1, 607634
LRPAP1	153.8	100.0%	99.6%	Myopia 23, autosomal recessive, 615431
LSS	138.5	100.0%	99.9%	Cataract 44, 616509 Hypotrichosis 14, 618275
LTBP2	124.2	100.0%	99.7%	Glaucoma 3, primary congenital, D, 613086 ?Weill-Marchesani syndrome 3, recessive, 614819 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LYST	135.6	99.3%	97.1%	Chediak-Higashi syndrome, 214500
LZTFL1	116.5	99.9%	99.2%	Bardet-Biedl syndrome 17, 615994
MAB21L2	265.2	100.0%	100.0%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAF	103.8	89.3%	84.7%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAK	134.1	99.2%	96.5%	Retinitis pigmentosa 62, 614181
MAPKAPK3	94.7	100.0%	99.6%	?Macular dystrophy, patterned, 3, 617111
MERTK	133.5	99.5%	99.0%	Retinitis pigmentosa 38, 613862
MFN2	130.9	100.0%	99.9%	Hereditary motor and sensory neuropathy VIA, 601152 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260
MFRP	140.5	100.0%	100.0%	Nanophthalmos 2, 609549 Microphthalmia, isolated 5, 611040
MFSD8	117.4	100.0%	99.6%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MIP	126.1	99.9%	97.4%	Cataract 15, multiple types, 615274
MIR184	NC	NC	NC	EDICT syndrome, 614303

MITF	145.6	100.0%	100.0%	COMMAD syndrome, 617306 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MKKS	161.5	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	98.8	99.9%	98.5%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MVK	130.3	90.5%	90.4%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYO7A	134.7	99.9%	99.1%	Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317 Usher syndrome, type 1B, 276900
MYOC	166.0	100.0%	99.1%	Glaucoma 1A, primary open angle, 137750
NAA10	112.8	100.0%	99.4%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NBAS	138.4	99.9%	99.2%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NDP	96.2	100.0%	99.6%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDUFS2	102.9	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NEK2	86.4	98.8%	92.9%	?Retinitis pigmentosa 67, 615565
NEUROD1	172.7	100.0%	99.9%	Maturity-onset diabetes of the young 6, 606394
NHS	114.3	98.6%	96.5%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NMNAT1	118.9	99.9%	98.3%	Leber congenital amaurosis 9, 608553
NPHP1	119.7	99.8%	97.8%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	121.6	99.7%	98.3%	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	134.2	100.0%	99.8%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NR2E3	117.8	99.9%	98.9%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131

NR2F1	261.9	100.0%	100.0%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRL	129.8	100.0%	99.0%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type, 0
NYX	155.4	99.9%	99.1%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	69.1	80.2%	69.8%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCA2	123.6	99.8%	97.4%	Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200
OCRL	106.2	99.9%	98.6%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	52.3	85.5%	70.0%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OPA1	121.4	99.7%	97.5%	Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	171.9	100.0%	99.9%	Optic atrophy 3 with cataract, 165300 3-methylglutaconic aciduria, type III, 258501
OPN1LW	61.2	67.9%	61.0%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	61.9	68.7%	60.9%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700
OTX2	135.4	100.0%	99.6%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OVOL2	130.8	99.9%	98.7%	Corneal dystrophy, posterior polymorphous, 1, 122000
P3H2	102.6	100.0%	99.1%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	119.3	99.9%	98.7%	Myopia 25, autosomal dominant, 617238
PANK2	161.5	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX2	198.0	100.0%	100.0%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX6	122.8	100.0%	99.9%	Optic nerve hypoplasia, 165550 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Aniridia, 106210 Keratitis, 148190

				Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430 Anterior segment dysgenesis 5, multiple subtypes, 604229
PCDH15	139.4	99.2%	98.9%	Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533
PCYT1A	97.1	99.1%	95.7%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE6A	105.5	100.0%	99.7%	Retinitis pigmentosa 43, 613810
PDE6B	171.1	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	116.7	99.4%	97.2%	Cone dystrophy 4, 613093
PDE6D	121.7	100.0%	100.0%	?Joubert syndrome 22, 615665
PDE6G	134.0	100.0%	99.7%	Retinitis pigmentosa 57, 613582
PDE6H	60.3	96.8%	76.1%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDZD7	103.9	99.7%	98.4%	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 Deafness, autosomal recessive 57, 618003
PET100	95.2	99.7%	90.6%	Mitochondrial complex IV deficiency, 220110
PEX1	126.3	100.0%	99.1%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX2	137.4	100.0%	100.0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	105.1	100.0%	100.0%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX7	108.8	91.3%	91.0%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGK1	47.0	92.1%	78.7%	Phosphoglycerate kinase 1 deficiency, 300653
PHOX2A	63.1	99.8%	94.3%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	75.9	100.0%	97.9%	Refsum disease, 266500
PIKFYVE	136.7	99.9%	99.3%	Corneal fleck dystrophy, 121850
PITX2	186.2	100.0%	99.6%	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PITX3	103.0	100.0%	99.8%	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, syndromic, autosomal recessive, 610623 Cataract 11, multiple types, 610623

PLA2G5	111.2	100.0%	100.0%	No OMIM disease ID
PLK4	145.8	99.9%	98.1%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PNPLA6	153.1	100.0%	99.6%	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
POC1B	77.8	99.7%	97.8%	Cone-rod dystrophy 20, 615973
POC5	132.3	99.3%	96.0%	No OMIM Disease ID
POMGNT1	123.6	100.0%	99.8%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
PPT1	140.2	90.3%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	107.5	100.0%	100.0%	Retinitis pigmentosa 36, 610599
PRDM13	207.6	100.0%	99.6%	No OMIM Disease ID
PRDM5	136.5	99.9%	98.2%	Brittle cornea syndrome 2, 614170
PRIMPOL	117.5	97.4%	93.8%	Myopia 22, autosomal dominant, 615420
PROM1	106.6	97.6%	95.2%	Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051
PRPF3	74.9	98.9%	95.5%	Retinitis pigmentosa 18, 601414
PRPF31	127.5	99.8%	97.9%	Retinitis pigmentosa 11, 600138
PRPF4	128.6	100.0%	99.8%	Retinitis pigmentosa 70, 615922
PRPF6	120.0	100.0%	99.8%	Retinitis pigmentosa 60, 613983
PRPF8	109.8	100.0%	99.3%	Retinitis pigmentosa 13, 600059
PRPH2	217.4	100.0%	100.0%	Macular dystrophy, patterned, 1, 169150 Retinitis punctata albescens, 136880 Choroidal dystrophy, central areolar 2, 613105 Retinitis pigmentosa 7 and digenic form, 608133 Leber congenital amaurosis 18, 608133 Macular dystrophy, vitelliform, 3, 608161
PRSS56	111.2	99.9%	99.3%	Microphthalmia, isolated 6, 613517
PXDN	150.2	100.0%	99.8%	Anterior segment dysgenesis 7, with sclerocornea, 269400
RAB28	64.3	99.1%	91.0%	Cone-rod dystrophy 18, 615374

RAB3GAP2	89.9	99.7%	96.1%	Warburg micro syndrome 2, 614225 Martsolf syndrome, 212720
RARB	93.5	100.0%	100.0%	Microphthalmia, syndromic 12, 615524
RAX	156.7	100.0%	99.9%	Microphthalmia, isolated 3, 611038
RAX2	98.8	100.0%	100.0%	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RBP3	168.5	100.0%	100.0%	?Retinitis pigmentosa 66, 615233
RBP4	149.6	99.8%	96.8%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	98.4	99.9%	99.0%	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	190.9	100.0%	100.0%	Leber congenital amaurosis 12, 610612
RDH11	94.8	99.9%	98.8%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	86.4	99.8%	98.1%	Leber congenital amaurosis 13, 612712
RDH5	182.9	100.0%	100.0%	Fundus albipunctatus, 136880
REEP6	226.6	100.0%	99.9%	Retinitis pigmentosa 77, 617304
RGS9	119.4	98.9%	96.9%	Bradyopsia, 608415
RGS9BP	170.1	100.0%	100.0%	Bradyopsia, 608415
RHO	180.4	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731
RIMS1	129.8	99.6%	98.1%	Cone-rod dystrophy 7, 603649
RLBP1	129.0	100.0%	99.9%	Fundus albipunctatus, 136880 Bothnia retinal dystrophy, 607475 Retinitis punctata albescens, 136880 Newfoundland rod-cone dystrophy, 607476
ROM1	139.5	100.0%	99.9%	Retinitis pigmentosa 7, digenic form, 608133
RP1	112.2	91.5%	91.0%	Retinitis pigmentosa 1, 180100
RP1L1	153.1	100.0%	100.0%	Occult macular dystrophy, 613587
RP2	159.7	100.0%	99.2%	Retinitis pigmentosa 2, 312600
RP9	64.9	95.2%	81.1%	?Retinitis pigmentosa 9, 180104
RPE65	133.0	100.0%	99.8%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	80.2	83.1%	74.6%	Retinitis pigmentosa 3, 300029 Cone-rod dystrophy, X-linked, 1, 304020 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834

RPGRIP1	132.7	100.0%	99.9%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	124.2	96.8%	95.8%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RS1	54.6	99.2%	89.0%	Retinoschisis, 312700
RTN4IP1	80.2	99.6%	98.2%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
SAG	131.3	100.0%	99.9%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SAMD11	110.0	96.5%	90.5%	No OMIM Disease ID
SC5D	149.4	100.0%	99.6%	Lathosterolosis, 607330
SCAPER	137.7	97.8%	96.0%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCO2	134.9	100.0%	100.0%	Myopia 6, 608908 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377
SDCCAG8	123.5	100.0%	99.7%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEMA4A	133.6	100.0%	99.5%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SGSH	152.5	98.1%	94.9%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SHH	165.7	100.0%	100.0%	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SIPA1L3	192.1	100.0%	99.8%	?Cataract 45, 616851
SIX6	303.9	100.0%	100.0%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SLC16A12	134.4	100.0%	99.9%	Cataract 47, juvenile, with microcornea, 612018
SLC24A1	175.4	100.0%	100.0%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	102.2	100.0%	99.6%	Albinism, oculocutaneous, type VI, 113750
SLC25A46	175.2	99.8%	97.2%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC33A1	135.7	99.8%	97.0%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC38A8	77.1	99.3%	95.7%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A5	143.6	100.0%	99.7%	Myopia 24, autosomal dominant, 615946
SLC45A2	119.4	100.0%	99.8%	Albinism, oculocutaneous, type IV, 606574

SLC4A11	173.6	100.0%	100.0%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC52A2	213.2	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC7A14	155.0	100.0%	100.0%	Retinitis pigmentosa 68, 615725
SMOC1	121.2	99.9%	98.4%	Microphthalmia with limb anomalies, 206920
SNRNP200	124.8	99.9%	98.7%	Retinitis pigmentosa 33, 610359
SOX2	261.8	100.0%	100.0%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX5	92.0	99.8%	96.6%	Lamb-Shaffer syndrome, 616803
SPATA7	120.5	99.7%	97.1%	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
SPP2	113.1	100.0%	100.0%	No OMIM Disease ID
STRA6	125.5	100.0%	99.9%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
TACSTD2	314.2	100.0%	100.0%	Corneal dystrophy, gelatinous drop-like, 204870
TCTN1	96.0	95.7%	92.6%	Joubert syndrome 13, 614173
TCTN3	121.0	100.0%	100.0%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TDRD7	139.3	99.9%	99.0%	Cataract 36, 613887
TEAD1	136.1	100.0%	99.7%	Sveinsson chorioretinal atrophy, 108985
TENM3	155.3	99.8%	99.4%	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TGFBI	118.3	99.9%	98.8%	Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Avellino type, 607541
TIMM8A	50.3	95.4%	80.0%	Mohr-Tranebjaerg syndrome, 304700
TIMP3	148.2	100.0%	100.0%	Sorsby fundus dystrophy, 136900
TMCO3	126.0	100.0%	99.6%	No OMIM Disease ID
TMEM126A	100.7	95.6%	79.5%	Optic atrophy 7, 612989
TMEM138	87.8	100.0%	99.0%	Joubert syndrome 16, 614465
TMEM216	92.0	99.9%	96.9%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091

TMEM231	112.1	100.0%	99.7%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	114.5	99.9%	98.8%	Joubert syndrome 14, 614424
TMEM67	80.6	99.3%	93.5%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TOPORS	181.9	100.0%	100.0%	Retinitis pigmentosa 31, 609923
TPP1	130.2	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TRAF3IP1	87.5	99.1%	96.7%	Senior-Loken syndrome 9, 616629
TREX1	261.9	100.0%	100.0%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM32	132.8	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRNT1	100.7	99.2%	95.3%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM1	134.7	100.0%	99.3%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TSPAN12	132.9	100.0%	99.8%	Exudative vitreoretinopathy 5, 613310
TTC8	116.8	99.7%	97.8%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTLL5	138.8	99.9%	98.9%	Cone-rod dystrophy 19, 615860
TUB	112.3	100.0%	99.4%	?Retinal dystrophy and obesity, 616188
TUBA3D	109.2	100.0%	97.4%	Keratoconus 9, 617928
TUBB3	135.6	99.9%	99.1%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4B	96.6	100.0%	100.0%	Leber congenital amaurosis with early-onset deafness, 617879
TUBGCP4	108.4	97.8%	95.1%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TULP1	128.8	100.0%	99.7%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
TYR	153.5	100.0%	100.0%	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 Albinism, oculocutaneous, type IA, 203100
TYRP1	155.1	100.0%	99.9%	Albinism, oculocutaneous, type III, 203290

UBIAD1	202.0	99.9%	98.2%	Corneal dystrophy, Schnyder type, 121800
UNC119	126.4	100.0%	99.9%	?Immunodeficiency 13, 615518 ?Cone-rod dystrophy, 0
UNC45B	121.5	100.0%	99.3%	?Cataract 43, 616279
USH1C	99.1	100.0%	99.3%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	221.0	99.9%	99.3%	Usher syndrome, type 1G, 606943
USH2A	130.8	100.0%	99.8%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP45	93.8	99.7%	98.1%	?Leber congenital amaurosis 19, 618513
VAX1	108.2	100.0%	99.4%	?Microphthalmia, syndromic 11, 614402
VCAN	155.3	100.0%	100.0%	Wagner syndrome 1, 143200
VIM	138.3	99.7%	98.0%	Cataract 30, pulverulent, 116300
VPS13B	135.9	99.4%	97.8%	Cohen syndrome, 216550
VSX1	73.0	99.0%	92.6%	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
VSX2	134.2	100.0%	100.0%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
WDPCP	105.7	97.1%	93.6%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	125.3	100.0%	99.4%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WFS1	210.0	100.0%	99.8%	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	145.9	100.0%	99.3%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WRN	120.3	100.0%	98.7%	Werner syndrome, 277700
YAP1	97.9	98.5%	94.0%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YME1L1	102.3	98.1%	92.4%	?Optic atrophy 11, 617302
ZEB1	155.3	100.0%	99.8%	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270

ZNF408	162.5	100.0%	100.0%	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469
ZNF423	215.2	100.0%	100.0%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF469	180.5	100.0%	100.0%	Brittle cornea syndrome 1, 229200
ZNF513	153.8	100.0%	100.0%	?Retinitis pigmentosa 58, 613617
ZNF644	153.0	100.0%	99.9%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors