VISION DISORDERS GENE PANEL DG 2.5/2.6

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ABCA4	112.8	100%	98%	Stargardt disease 1, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 Macular degeneration, age-related, 2, 153800 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200
ABCC6	94.8	93%	91%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABHD12	93.7	89%	78%	Polyneuropathy,hearing loss, ataxia,retinitis pigmentosa and cataract,612674
ACBD5	150.6	96%	95%	No OMIM disease Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
ADAM9	149.9	98%	93%	Cone-rod dystrophy 9, 612775
ADAMTS18	147.5	98%	97%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADIPOR1	88.4	100%	94%	No OMIM disease syndromic retinitis pigmentosa (Xy (2016) Hum Mutat 37(3):246-249
AGBL1	126.5	100%	100%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGK	115.1	99%	94%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AHI1	133	99%	93%	Joubert syndrome-3, 608629
AIPL1	111.6	100%	100%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
ALMS1	167.8	99%	99%	Alstrom syndrome, 203800
AP3B1	93.9	99%	92%	Hermansky-Pudlak syndrome 2, 608233
APOPT1	62.9	87%	82%	Mitochondrial complex IV deficiency, 220110
ARL13B	78.8	99%	81%	Joubert syndrome 8, 612291

ARL2BP	72.8	78%	71%	Retinitis pigmentosa with or without situs inversus, 615434
ARL6	94.9	99%	96%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ASPH	110.5	99%	94%	Traboulsi syndrome, 601552
ATF6	134.8	100%	98%	Achromatopsia 7, 616517
BBIP1	121	99%	87%	?Bardet-Biedl syndrome 18, 615995
BBS1	133.8	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	160.2	100%	99%	Bardet-Biedl syndrome 10, 209900
BBS12	196.3	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	173	100%	98%	Bardet-Biedl syndrome 2, 209900
BBS4	136.1	99%	95%	Bardet-Biedl syndrome 4, 209900
BBS5	103.3	97%	91%	Bardet-Biedl syndrome 5, 209900
BBS7	121.3	96%	90%	Bardet-Biedl syndrome 7, 209900
BBS9	105.1	94%	93%	Bardet-Biedl syndrome 9, 209900
BCOR	71	98%	95%	Microphthalmia, syndromic 2, 300166
BEST1	111.7	99%	96%	Best macular dystrophy, 153700 Maculopathy, bull's-eye Vitelliform macular dystrophy, adult-onset, 608161 Bestrophinopathy, 611809 Vitreoretinochoroidopathy, 193220 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220
BFSP1	90.6	94%	89%	Cataract 33, 611391
BFSP2	80.3	98%	94%	Cataract 12, multiple types, 611597
BLOC1S3	38.5	99%	84%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	89.9	99%	84%	Hermansky-Pudlak syndrome 9, 614171
BMP4	120.8	100%	98%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625 -3
C10orf11	135.8	99%	99%	Ablinism, oculocutaneous, type VII, 615179
C12orf65	74.3	99%	97%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	78.4	100%	96%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
C1QTNF5	141.5	78%	60%	Retinal degeneration, late-onset, autosomal dominant, 605670

C21orf2	81.5	99%	96%	No OMIM disease Retinal dystrophy,early-onset with macular staphyloma (Khan (2015) Br J Ophtalmol 99,1725) Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
C2orf71	116.9	100%	99%	Retinitis pigmentosa 54, 613428
C5orf42	119.9	95%	91%	Joubert syndrome 17, 614615
C8orf37	116.7	100%	99%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500
CA4	133.6	100%	97%	Retinitis pigmentosa 17, 600852
CABP4	90.2	96%	92%	Night blindness, congenital stationary (incomplete), 2B, autosomal recessive, 610427
CACNA1F	62.7	98%	91%	Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Cone-rod dystropy, X-linked, 3, 300476 Aland Island eye disease, 300600
CACNA2D4	100.4	98%	97%	Retinal cone dystrophy 4, 610478
CAPN5	142.6	100%	99%	Vitreoretinopathy, neovascular inflammatory, 193235
CC2D2A	121.8	99%	94%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CDH23	183	100%	99%	Deafness,autosomal recessive 12,601386 Usher syndrome,type 1D,601067 Usher syndrome type 1D/1F digenic,601067
CDH3	132.2	100%	97%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	141.1	100%	99%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CEP164	75.2	99%	94%	Nephronophthisis 15, 614845
CEP250	84.5	100%	97%	No OMIM disease Usher syndrome,atypical (Khateb (2014) J Med Genet 51,460) Miscarriage, recurrent (Filges (2014) Mol Hum Reprod epub,epub)
CEP290	69.5	89%	76%	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900

CEP41	83.4	97%	91%	Joubert syndrome 15, 614464
CERKL	108.2	99%	95%	Retinitis pigmentosa 26, 608380
CFH	176.8	98%	95%	{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 Complement factor H deficiency, 609814 {Macular degeneration, age-related, 4}, 610698 Basal laminar drusen, 126700
CHM	66.7	90%	77%	Choroideremia, 303100
CHMP4B	125.3	99%	96%	Cataract 31, multiple types, 605387
CHST6	280	100%	100%	Macular corneal dystrophy, 217800
CIB2	188.6	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLN3	107.2	99%	97%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	144.8	97%	91%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	119.2	100%	92%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	206.6	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLRN1	141.9	100%	100%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180
CNGA1	128.2	84%	84%	Retinitis pigmentosa 49, 613756
CNGA3	149.8	99%	98%	Achromatopsia-2, 216900
CNGB1	91.5	95%	92%	Retinitis pigmentosa 45, 613767
CNGB3	104.7	91%	86%	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200
CNNM4	174.3	98%	97%	Jalili syndrome, 217080
COL11A1	85.5	92%	83%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520

COL11A2	11	46%	13%	Stickler syndrome, type III, 184840 Otospondylomegaepiphyseal dysplasia, 215150 Weissenbacher-Zweymuller syndrome, 277610 Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524
COL2A1	93.5	99%	96%	Stickler syndrome, type I, 108300 Kniest dysplasia, 156550 Achondrogenesis, type II or hypochondrogenesis, 200610 SED congenita, 183900 SMED Strudwick type, 184250 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Spondylo
COL8A2	21.8	76%	53%	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140
COL9A1	107.6	99%	95%	Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	55.5	98%	86%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
CRB1	199.9	100%	99%	Retinitis pigmentosa-12, autosomal recessive, 600105 Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870
CRX	91.4	100%	98%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	121.6	84%	82%	Cataract 9, multiple types, 604219
CRYAB	120.2	97%	95%	Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869 Cardiomyopathy, dilated, 1II, 615184
CRYBA1	130.8	100%	100%	Cataract 10, multiple types, 600881
CRYBA2	126	100%	100%	?Cataract 42,115900
CRYBA4	98.4	100%	97%	Cataract 23, 610425
CRYBB1	108.3	100%	99%	Cataract 17, multiple types, 611544
CRYBB2	152	100%	100%	Cataract 3, multiple types, 601547

CRYBB3	134.1	100%	98%	Cataract 22, autosomal recessive, 609741
CRYGB	95	100%	95%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	114.1	100%	100%	Cataract 2, multiple types, 604307
CRYGD	89	100%	100%	Cataract 4, multiple types, 115700
CRYGS	130.9	95%	91%	Cataract 20, multiple types, 116100
CSPP1	99.3	99%	92%	Joubert syndrome 21, 615636
CTDP1	93.2	90%	85%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA1	127.1	100%	98%	Macular dystrophy,patterned,2,608970
CTSD	152.8	98%	94%	Ceroid lipofuscinosis, neuronal, 10, 610127
CYP1B1	106.3	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile or adult onset, 231300 Peters anomaly, 604229
CYP4V2	157	100%	98%	Bietti crystalline corneoretinal dystrophy, 210370
DCN	141.1	95%	94%	Corneal dystrophy, congenital stromal, 610048
DFNB31	102.8	100%	97%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DHDDS	84.7	95%	93%	Retinitis pigmentosa 59, 613861
DHX38	122.8	100%	98%	No OMIM disease Retinitis pigmentosa,early-onset with macular coloboma (Ajmal (2014) J Med Genet 51,444)
DKC1	77.4	99%	92%	Dyskeratosis congenita, X-linked, 305000
DRAM2	144.9	100%	100%	Cone-rod dystrophy 21, 616502
DTNBP1	100.5	100%	95%	Hermansky-Pudlak syndrome 7, 614076
EFEMP1	176.8	100%	98%	Doyne honeycomb degeneration of retina, 126600
ELOVL4	77.5	100%	98%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
EPG5	123.6	99%	94%	Vici syndrome, 242840
EPHA2	170.6	97%	97%	Cataract 6, multiple types, 116600
EYA1	139.1	100%	99%	?Otofaciacervical syndrome, 166780 Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
EYS	143.4	98%	94%	Retinitis pigmentosa 25, 602772

FA2H	87.6	93%	81%	Spastic paraplegia 35, autosomal recessive, 612319
FAM161A	110.4	99%	93%	Retinitis pigmentosa 28, 606068
FLVCR1	121.4	99%	97%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOXE3	9.3	49%	33%	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256
FRMD7	79.4	99%	93%	Nystagmus 1,congenital,X-linked,310700
FTL	104.5	99%	82%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
FYCO1	114.3	100%	99%	Cataract 18, autosomal recessive, 610019
FZD4	207.1	100%	98%	Exudative vitreoretinopathy, 133780 Retinopathy of prematurity, 133780
GALK1	88.7	96%	91%	Galactokinase deficiency with cataracts, 230200
GALT	141.5	100%	99%	Galactokinase deficiency with cataracts, 230200
GCNT2	164.9	100%	100%	[Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 110800 Adult i phenotype without cataract, 110800
GDF3	119.6	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF6	56.8	93%	82%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Microphthalmia, isolated 4, 613094 Microphthalmia with coloboma 6, digenic, 613703 Leber congenital amaurosis 17, 615360
GFER	69.7	98%	71%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GJA1	205.3	100%	100%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive,218400 Erythrokeratodermia variabilis et progressiva,133200 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia,104100 Syndactyly, type III, 186100
GJA3	152.3	100%	100%	Cataract 14, multiple types, 601885

GJA8	136.5	100%	100%	Cataract 1, multiple types, 116200
GNAT1	153.1	100%	100%	Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	142.9	100%	98%	Achromatopsia-4, 613856
GNPTG	125.2	90%	87%	Mucolipidosis III gamma, 252605
GPR143	39.9	90%	67%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR179	119.3	100%	97%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR98	145.8	99%	94%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
GRK1	100.1	100%	100%	Oguchi disease-2, 613411
GRM6	139.7	93%	83%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	171.5	100%	100%	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GSN	109	91%	87%	Amyloidosis, Finnish type, 105120
GUCA1A	147.4	100%	100%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	130.1	100%	98%	Retinitis pigmentosa 48, 613827
GUCY2D	79.9	96%	84%	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
HARS	143.6	100%	100%	Usher syndrome type 3B, 614504
HCCS	63.8	99%	92%	Microphthalmia, syndromic 7, 309801
HGSNAT	95	81%	81%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HK1	134.8	100%	99%	Hemolytic anemia due to hexokinase deficiency,235700 Neuropathy,hereditary motor and sensory,Russe type,605285 Retinitis pigmentosa (Wang (2014) Invest Ophthalmol Vis Sci 55,7159)
HMX1	19.6	63%	39%	Oculoauricular syndrome, 612109
HPS1	102.9	100%	98%	Hermansky-Pudlak syndrome 1, 203300
HPS4	130.4	100%	98%	Hermansky-Pudlak syndrome 4, 614073
HPS5	137.7	99%	98%	Hermansky-Pudlak syndrome 5, 614074

HPS6	100.4	100%	92%	Hermansky-Pudlak syndrome 6, 614075
HSF4	89.4	96%	87%	Cataract 5, multiple types, 116800
IDH3B	166.6	100%	100%	Retinitis pigmentosa 46, 612572
IFT140	105.2	99%	96%	Mainzer-Saldino syndrome, 266920
IFT172	108.6	99%	97%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	106.4	100%	99%	?Bardet-Biedl syndrome 19, 615996
IMPDH1	47.6	91%	74%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	98.9	100%	95%	Macular dystrophy, vitelliform, 4, 616151
IMPG2	166.8	100%	97%	Macular dystrophy,vitelliform,5,616152 Retinitis pigmentosa 56,613581
INPP5E	83.9	96%	88%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	153.4	100%	99%	Nephronophthisis 2,infantile,602088
IQCB1	96.9	78%	74%	Senior-Loken syndrome 5, 609254
JAG1	139.8	100%	97%	?Deafness, congenital heart defects, and posterior embryotoxon Alagille syndrome, 118450 Tetralogy of Fallot, 187500
JAM3	136.4	100%	99%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KCNJ13	194	100%	98%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNV2	120	100%	100%	Retinal cone dystrophy 3B, 610356
KERA	185.9	100%	100%	Cornea plana congenita, recessive, 217300
KIF11	76.6	97%	95%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF7	70	91%	84%	?Al-Gazali-Bakalinova syndrome,607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KIZ	150.4	98%	94%	Retinitis pigmentosa 69,615780
KLHL7	139.1	99%	98%	Retinitis pigmentosa 42, 612943
KRT12	88	95%	92%	Meesmann corneal dystrophy, 122100

KRT3	90.9	100%	96%	Meesmann corneal dystrophy, 122100
LAMA1	134.2	99%	98%	Poretti-Boltshauser syndrome, 615960
LCA5	130	95%	94%	Leber congenital amaurosis 5, 604537
LEPREL1	87.8	93%	87%	Myopia,high,with cataract and vitreoretinal degeneration,614292
LIM2	88.4	100%	99%	Cataract 19, 615277
LRAT	285.3	100%	100%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRIT3	148.4	94%	93%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP5	165.5	98%	96%	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis,autosomal dominant 1,607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LSS	108.7	100%	98%	Cataract 44,616509
LYST	129.6	97%	92%	Chediak-Higashi syndrome, 214500
LZTFL1	121.2	98%	90%	Bardet-Biedl syndrome 17, 615994
MAB21L2	210	100%	100%	Microphthalmia, syndromic 14, 615877
MAF	51.8	75%	72%	Cataract, pulverulent or cerulean, with or without microcornea, 610202
MAK	146.6	94%	93%	Retinitis pigmentosa 62, 614181
MAPKAPK3	76.9	98%	95%	No OMIM disease Martinique crinkled retinal pigment epitheliopathy (Meunier (2016) Hum Mol Gene 25,916)
MERTK	162.2	100%	99%	Retinitis pigmentosa 38, 613862
MFN2	138	100%	100%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VI, 601152
MFRP	117.5	100%	100%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	120.9	100%	98%	Ceroid lipofuscinosis, neuronal, 7, 610951
MIP	104.3	94%	87%	Cataract 15, multiple types, 615274

MITF	134.3	100%	100%	Tietz albinism-deafness syndrome,103500 Waardenburg syndrome,type 2A,193510 Waardenburg syndrome/ocular albinism, digenic,103470 {Melanoma,cutaneous malignant,susceptibility to,8},614456
MKKS	222.1	89%	89%	Bardet-Biedl syndrome 6, 209900 McKusick-Kaufman syndrome, 236700
MKS1	95.9	99%	96%	Meckel syndrome 1, 249000
MVK	126.3	100%	100%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, disseminated superficial actinic, 175900
MYO7A	127	98%	95%	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYOC	171.9	100%	100%	Glaucoma 1A, primary open angle, 137750
NAA10	58	100%	91%	?Microphthalmia, syndromic 1, 309800 N-terminal acetyltransferase deficiency, 300855
NDP	80.3	100%	100%	Exudative vitreoretinopathy, X-linked, 305390 Norrie disease, 310600
NEK2	81.6	95%	86%	?Retinitis pigmentosa 67, 615565
NEUROD1	153.6	100%	100%	Maturity-onset diabetes of the young 6,606394 {Diabetes mellitus,noninsulin dependent},125853 Retinitis pigmentosa,autosomal recessive (Wang (2015) Invest Opthalmol Vis Sci 56,150)
NHS	83.7	93%	88%	Nance-Horan syndrome, 302350
NMNAT1	128.7	100%	99%	Leber congenital amaurosis 9, 608553
NPHP1	122	99%	94%	Nephronophthisis 1, juvenile, 256100 Joubert syndrome 4, 609583 Senior-Loken syndrome-1, 266900
NPHP3	111.4	98%	91%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	127.5	99%	97%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NR2E3	95.9	100%	100%	Enhanced S-cone syndrome,268100 Retinitis pigmentosa 37,61131

NR2F1	182.2	99%	92%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRL	61.1	98%	86%	Retinal degeneration, autosomal recessive, clumped pigment type Retinitis pigmentosa 27, 613750
NYX	50.7	96%	95%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	73.6	68%	61%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCA2	126.1	97%	95%	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
OFD1	29.6	74%	53%	?Retinitis pigmentosa 23,300424 Joubert syndrome 10, 300804 Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OPA1	115.4	98%	89%	?Mitochondrial DNA depletion syndrome 14 (encephacardiomyopathic type),616896 Behr syndrome,210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 {Glaucoma, normal tension, susceptibility to}, 606657
OPA3	89.2	99%	93%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPN1LW	48.9	66%	44%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	64.3	65%	50%	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800
OR2W3	194.8	100%	100%	No OMIM disease Retinitis pigmentosa (Ma (2015) Sci Rep 5,9236)
OTX2	116.2	100%	97%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, and pituitary dysfunction, 610125
PANK2	142.9	95%	86%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX2	148.6	100%	99%	Papillorenal syndrome, 120330 Renal hypoplasia, isolated, 191830

PAX6	129.3	100%	100%	?Morning glory disc anomaly, 120430 Aniridia, 106210 Cataract with late-onset corneal dystrophy, 106210 Coloboma of optic nerve,120430 Coloboma,ocular,120200 Foveal hyperplasia, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550 Peters anomaly, 604229
PCDH15	164.2	99%	98%	Deafness, autosomal recessive 23,609533 Usher syndrome, type 1D/1F digenic,601067 Usher syndrome,type 1F,602083
PCYT1A	123.1	97%	94%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE6A	120.3	100%	99%	Retinitis pigmentosa 43, 613810
PDE6B	136.8	100%	99%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	131.5	96%	94%	Cone dystrophy 4, 613093
PDE6D	93.9	100%	100%	?Joubert syndrome 22, 615665
PDE6G	85.3	99%	94%	Retinitis pigmentosa 57, 613582
PDE6H	58.1	51%	48%	Achromatopsia 6,610024 Retinal cone dystrophy 3,610024
PDZD7	77.7	100%	89%	Usher syndrome,type IIC,FPR98/PDZD7 digenic,605472 {Retinal disease in Usher syndrome type IIA,modifier of},276901
PET100	110.5	89%	72%	Mitochondrial complex IV deficiency, 220110
PEX1	106.9	97%	97%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX2	147.6	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX7	121.4	91%	82%	Chondrodysplasia punctata, rhizomelic, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PGK1	35.1	80%	65%	Phosphoglycerate kinase 1 deficiency, 300653
PHYH	87.5	97%	92%	Refsum disease, 266500
PIKFYVE	137.7	99%	98%	Corneal fleck dystrophy, 121850

PITX2	128.1	96%	93%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PITX3	56	100%	92%	Anterior segment mesenchymal dysgenesis, 107250
PLA2G5	117.5	100%	100%	Fleck retina, familial benign, 228980
PLK4	135.2	99%	86%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PNPLA6	111.2	99%	97%	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
POC1B	93.5	92%	90%	Cone-rod dystrophy 20, 615973
PPT1	150.6	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	89.1	100%	100%	Retinitis pigmentosa 36, 610599
PROM1	112.9	96%	93%	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PRPF3	73.3	98%	91%	Retinitis pigmentosa 18, 601414
PRPF31	99.1	100%	93%	Retinitis pigmentosa 11, 600138
PRPF4	137.2	99%	98%	Retinitis pigmentosa 70, 615922
PRPF6	114.1	100%	99%	Retinitis pigmentosa 60, 613983
PRPF8	132.4	100%	99%	Retinitis pigmentosa 13, 600059
PRPH2	209.9	100%	98%	Choroidal dystrophy,central areolar 2,613105 Leber congenital amaurosis 18,608133 Macular dystrophy,patterned,1,169150 Macular dystrophy,vitelliform,3,608161 Retinitis pigmentosa 7 and digenic,608133 Retinitis punctata albescens,136880
PRSS56	38.9	93%	73%	Microphthalmia, isolated 6, 613517
PXDN	153.8	99%	97%	Corneal opacification and other ocular anomalies, 269400
RAB28	50.6	93%	77%	Cone-rod dystrophy 18, 615374
RARB	124.5	100%	100%	Microphthalmia, syndromic 12, 615524

RAX2	45.5	90%	69%	Cone-rod dystrophy 11, 610381 Macular degeneration, age-related, 6,613757
RBP3	128	100%	99%	?Retinitis pigmentosa 66, 615233
RBP4	87	93%	91%	Retinol dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RD3	136.9	100%	100%	Leber congenital amaurosis 12, 610612
RDH11	125.6	100%	97%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	87.6	97%	83%	Leber congenital amaurosis 13, 612712
RDH5	144	100%	100%	Fundus albipunctatus, 136880
RGR	123.5	100%	100%	Retinitis pigmentosa 44, 613769
RGS9	94.3	100%	99%	Bradyopsia, 608415
RGS9BP	59	100%	97%	Bradyopsia, 608415
RHO	224.6	100%	100%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIMS1	113.5	96%	93%	Cone-rod dystrophy 7, 603649
RLBP1	127.9	100%	100%	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
ROM1	97.7	100%	98%	Retinitis pigmentosa 7,digenic,608133
RP1	123.7	99%	96%	Retinitis pigmentosa 1, 180100 {Hypertriglyceridemia, susceptibility to}, 145750
RP1L1	81.7	100%	98%	Occult macular dystrophy, 613587
RP2	117.2	100%	98%	Retinitis pigmentosa 2, 312600
RP9	46	77%	77%	Retinitis pigmentosa 9, 180104
RPE65	138.6	100%	99%	Leber congenital amaurosis 2, 204100
RPGR	59.2	81%	66%	Cone-rod dystrophy, X-linked, 1, 304020 Macular degeneration, X-linked atrophic, 300834 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455
RPGRIP1	145.2	100%	99%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826

RPGRIP1L	133.2	95%	93%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RS1	32.2	83%	72%	Retinoschisis, 312700
SAG	126.2	100%	100%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SDCCAG8	116.7	100%	96%	Senior-Loken syndrome 7, 613615
SEMA4A	118.9	100%	97%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SHH	84.8	95%	91%	Holoprosencephaly-3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SIPA1L3	121.9	98%	96%	?Cataract 45,616851
SIX6	180.4	100%	100%	Microphthalmia with cataract 2, 212550
SLC24A1	199.4	100%	99%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	119.9	95%	93%	Albinism, oculocutaneous, type VI, 113750
SLC33A1	133.8	98%	89%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC38A8	74	100%	93%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC45A2	128.1	100%	98%	Albinism, oculocutaneous, type IV, 606574
SLC4A11	133.3	100%	99%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy 2, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC52A2	168	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC7A14	179.5	100%	100%	Retinitis pigmentosa 68, 615725
SNRNP200	144.9	100%	98%	Retinitis pigmentosa 33, 610359
SOX2	80.8	99%	96%	Retinitis pigmentosa 33, 610359
SPATA7	128	96%	86%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPP2	155.5	100%	100%	No OMIM disease Autism (Neale (2012) Nature 485,242) Retinitis pigmentosa (Li (2015) Sci Rep 5,14867)

STRA6	105.6	100%	100%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
TACSTD2	176.2	99%	97%	Corneal dystrophy, gelatinous drop-like, 204870
TCTN1	98.8	94%	92%	Joubert syndrome 13, 614173
TCTN3	116.3	99%	98%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDRD7	170.6	98%	97%	Cataract 36, 613887
TEAD1	161.4	100%	96%	Sveinsson choreoretinal atrophy, 108985
TENM3	185.5	99%	98%	Microphthalmia, isolated, with coloboma 9, 61545
TGFBI	121.8	100%	92%	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Reis
TIMM8A	17.5	64%	30%	Mohr-Tranebjaerg syndrome, 304700
TIMP3	160.3	100%	100%	Sorsby fundus dystrophy, 136900
TMEM126A	108.6	99%	91%	Optic atrophy-7, 612989
TMEM138	102.7	99%	97%	Joubert syndrome 16, 614465
TMEM231	86.1	99%	95%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397
TMEM237	96.1	99%	97%	Joubert syndrome 14, 614424
TMEM67	70.3	91%	84%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 209900
TOPORS	234.8	100%	100%	Retinitis pigmentosa 31, 609923
TPP1	124	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500
TREX1	214.3	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRIM32	134.2	100%	100%	Bardet-Biedl syndrome 11, 209900

				Muscular dystrophy, limb-girdle, type 2H, 254110
TRNT1	93.1	95%	91%	Retinitis pigmentosa and erythrocytic microcytosis,616959 Sideroblastic anemia with B-cell immunodeficiency,periodic fevers and developmental delay,616084
TRPM1	161.4	100%	98%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TSPAN12	135.9	100%	98%	Exudative vitreoretinopathy 5, 613310
TTC8	88.4	98%	88%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
TTLL5	149.6	100%	96%	Cone-rod dystrophy 19,615860
TUB	91.6	99%	94%	?Retinal dystrophy and obesity, 616188
TUBGCP4	135.5	98%	95%	Microcephaly and chorioretinopathy,autosomal recessive,616335
TULP1	88.6	98%	93%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TYR	189.7	100%	100%	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}
TYRP1	190.1	100%	100%	Albinism, oculocutaneous, type III, 203290
UBIAD1	225.9	98%	93%	Corneal dystrophy, Schnyder type, 121800
UNC119	91.1	98%	72%	?Cone-Rod dystrophy ?Immunodeficiency 13,615518
UNC45B	116.5	100%	98%	?Cataract 43,616279
USH1C	99.1	98%	95%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904
USH1G	153.6	95%	93%	Usher syndrome, type 1G, 606943
USH2A	156.8	99%	98%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
VAX1	44.7	87%	70%	Microphthalmia, syndromic 11, 614402
VCAN	185.9	100%	100%	Wagner syndrome 1, 143200
VIM	117	100%	93%	Cataract 30, pulverulent, 116300
VSX1	54.9	91%	73%	Corneal dystrophy, posterior polymorphous, 1, 122000 Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300

VSX2	67.6	98%	88%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
WDPCP	107.1	94%	93%	?Bardet-Biedl syndrome 15, 615992
WDR19	133.5	100%	98%	?Cranioectodermal dysplasia 4,614378 ?Short-rib thoracic dysplasia 5 w/wo polydactyly,614376 Nephronophthisis 13,614377 Senior-Loken syndrome 8,616307
WFS1	218.6	98%	97%	?Cataract 41,116400 Deafness,autosomal dominant 6/14/38, 600965 Wolfram syndrome, 222300 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent,association with}
WRN	128.3	97%	94%	Werner syndrome, 277700
YAP1	95.5	85%	79%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation
ZEB1	188	100%	99%	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZNF408	112	100%	100%	?Exudative vitreoretinopathy, 616468 Retinitis pigmentosa 72, 616469
ZNF423	226.6	100%	100%	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF513	90.9	100%	97%	Retinitis pigmentosa 58, 613617
ZNF644	163.1	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.
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 : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors