VISION DISORDERS GENE PANEL DGD09072015

Gene name	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM ID*
ABCA4	88.5	99%	96%	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	51.5	72%	67%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABHD12	61	98%	83%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACBD5	105.5	100%	99%	No OMIM disease ID
ADAM9	117	100%	98%	Cone-rod dystrophy 9, 612775
ADAMTS18	99.3	100%	98%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
AGBL1	96.1	100%	99%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGK	107.5	99%	99%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AHI1	115.2	100%	99%	Joubert syndrome-3, 608629
AIPL1	83.9	100%	100%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
ALMS1	195.2	98%	98%	Alstrom syndrome, 203800
AP3B1	109.5	100%	99%	Hermansky-Pudlak syndrome 2, 608233
APOPT1	88.4	87%	87%	Mitochondrial complex IV deficiency, 220110
ARL13B	121.2	99%	95%	Joubert syndrome 8, 612291
ARL2BP	85.2	100%	96%	Retinitis pigmentosa with or without situs inversus, 615434
ARL6	161.4	100%	100%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ASPH	110.2	99%	97%	Traboulsi syndrome, 601552

ATF6	105.3	100%	100%	Achromatopsia 7, 616517
BBIP1	103.1	82%	72%	?Bardet-Biedl syndrome 18, 615995
BBS1	113.4	99%	98%	Bardet-Biedl syndrome 1, 209900
BBS10	126.9	100%	100%	Bardet-Biedl syndrome 10, 209900
BBS12	144.2	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	115	100%	99%	Bardet-Biedl syndrome 2, 209900
BBS4	96.6	97%	95%	Bardet-Biedl syndrome 4, 209900
BBS5	136.5	100%	100%	Bardet-Biedl syndrome 5, 209900
BBS7	129.7	100%	99%	Bardet-Biedl syndrome 7, 209900
BBS9	121.9	99%	98%	Bardet-Biedl syndrome 9, 209900
BCOR	61.3	99%	95%	Microphthalmia, syndromic 2, 300166
BEST1	105	100%	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	124.9	100%	100%	Cataract 33, 611391
BFSP2	62.7	94%	92%	Cataract 12, multiple types, 611597
BLOC1S3	31	79%	68%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	121.5	92%	86%	Hermansky-Pudlak syndrome 9, 614171
BMP4	114.2	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625 -3
C10orf11	90.7	99%	99%	Ablinism, oculocutaneous, type VII, 615179
C12orf65	174.3	100%	100%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	72.8	100%	95%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
C1QTNF5	119.3	98%	89%	Retinal degeneration, late-onset, autosomal dominant, 605670
C21orf2	97.9	99%	96%	No OMIM disease ID Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
C2orf71	105.5	99%	96%	Retinitis pigmentosa 54, 613428
C5orf42	129.4	100%	99%	Joubert syndrome 17, 614615

C8orf37	95.2	100%	100%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500
CA4	83.8	98%	92%	Retinitis pigmentosa 17, 600852
CABP4	64.1	100%	100%	Night blindness, congenital stationary (incomplete), 2B, autosomal recessive, 610427
CACNA1F	42.4	94%	85%	Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Cone-rod dystropy, X-linked, 3, 300476 Aland Island eye disease, 300600
CACNA2D4	75.6	97%	91%	Retinal cone dystrophy 4, 610478
CAPN5	77.3	100%	95%	Vitreoretinopathy, neovascular inflammatory, 193235
CC2D2A	92.9	98%	97%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CDH23	93.5	98%	97%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CDH3	95.2	97%	93%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDHR1	110.9	98%	98%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CEP164	73.6	98%	91%	Nephronophthisis 15, 614845
CEP250	82	99%	93%	No OMIM disease ID Usher syndrome, atypical (Khateb (2014) J Med Genet 51,460)
CEP290	101.5	100%	98%	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900
CEP41	85.8	100%	99%	Joubert syndrome 15, 614464
CERKL	137.4	100%	100%	Retinitis pigmentosa 26, 608380
CFH	107.3	95%	92%	{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 Complement factor H deficiency, 609814 {Macular degeneration, age-related, 4}, 610698 Basal laminar drusen, 126700
CHM	53.3	98%	91%	Choroideremia, 303100

CHMP4B	114.9	100%	100%	Cataract 31, multiple types, 605387
CHST6	123.3	100%	100%	Macular corneal dystrophy, 217800
CIB2	102.9	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLN3	83.5	97%	96%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	139.7	100%	90%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	65	98%	82%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	133.8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLRN1	157.1	100%	100%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180
CNGA1	119.8	91%	90%	Retinitis pigmentosa 49, 613756
CNGA3	131.4	100%	99%	Achromatopsia-2, 216900
CNGB1	86.3	96%	91%	Retinitis pigmentosa 45, 613767
CNGB3	108.7	100%	98%	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200
CNNM4	171.3	99%	96%	Jalili syndrome, 217080
COL11A1	98	98%	97%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	13.9	55%	19%	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	84.6	99%	95%	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
COL8A2	62.8	97%	94%	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140
COL9A1	108	100%	96%	Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	72.9	98%	93%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
CRB1	163.2	100%	100%	Retinitis pigmentosa-12, autosomal recessive, 600105 Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870
CRX	152.8	100%	100%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	96.2	93%	91%	Cataract 9, multiple types, 604219
CRYAB	139.7	100%	100%	Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869 Cardiomyopathy, dilated, 1II, 615184
CRYBA1	91	100%	100%	Cataract 10, multiple types, 600881
CRYBA2	85.1	100%	93%	?Cataract 42,115900
CRYBA4	79	100%	100%	Cataract 23, 610425
CRYBB1	56.9	100%	94%	Cataract 17, multiple types, 611544
CRYBB2	111.3	100%	100%	Cataract 3, multiple types, 601547
CRYBB3	110.5	100%	100%	Cataract 22, autosomal recessive, 609741
CRYGB	69.4	100%	93%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	84.1	100%	96%	Cataract 2, multiple types, 604307
CRYGD	76.2	90%	77%	Cataract 4, multiple types, 115700
CRYGS	92.6	98%	90%	Cataract 20, multiple types, 116100
CSPP1	122.7	100%	99%	Joubert syndrome 21, 615636
CTDP1	70.3	89%	87%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168

CTSD	96.2	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CYP1B1	103.3	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile or adult onset, 231300 Peters anomaly, 604229
CYP4V2	124.7	100%	99%	Bietti crystalline corneoretinal dystrophy, 210370
DCN	91	89%	88%	Corneal dystrophy, congenital stromal, 610048
DFNB31	96.1	100%	98%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DHDDS	82.7	95%	91%	Retinitis pigmentosa 59, 613861
DHX38	81.7	97%	93%	No OMIM disease ID Retinitis pigmentosa, early-onset with macular coloboma (Ajmal (2014) J Med Genet 51,444)
DKC1	50	99%	91%	Dyskeratosis congenita, X-linked, 305000
DRAM2	108.4	100%	100%	Cone-rod dystrophy 21, 616502
DTNBP1	108.1	100%	100%	Hermansky-Pudlak syndrome 7, 614076
EFEMP1	124.3	100%	94%	Doyne honeycomb degeneration of retina, 126600
ELOVL4	102.9	100%	100%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
EPG5	89.4	100%	99%	Vici syndrome, 242840
EPHA2	89.5	97%	94%	Cataract 6, multiple types, 116600
EYA1	113.1	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	125.5	100%	99%	Retinitis pigmentosa 25, 602772
FA2H	64.6	87%	76%	Spastic paraplegia 35, autosomal recessive, 612319
FAM161A	149.9	100%	100%	Retinitis pigmentosa 28, 606068
FLVCR1	95.8	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOXE3	14.2	70%	38%	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256
FTL	95.3	100%	95%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
FYCO1	86.1	99%	98%	Cataract 18, autosomal recessive, 610019
FZD4	142.2	100%	100%	Exudative vitreoretinopathy, 133780 Retinopathy of prematurity, 133780

GALK1	91.1	99%	98%	Galactokinase deficiency with cataracts, 230200
GALT	104.8	100%	98%	Galactokinase deficiency with cataracts, 230200
GCNT2	162.9	100%	100%	[Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 110800 Adult i phenotype without cataract, 110800
GDF3	127.8	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF6	135.5	100%	100%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Microphthalmia, isolated 4, 613094 Microphthalmia with coloboma 6, digenic, 613703 Leber congenital amaurosis 17, 615360
GFER	65.6	99%	94%	Myopathy, mitochondrial, with cataract, hearing loss, and developmental delay, 613076
GJA1	63.1	91%	79%	Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Hypoplastic left heart syndrome 1, 241550 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, autosomal recessive, 257850
GJA3	100	100%	99%	Cataract 14, multiple types, 601885
GJA8	135.9	100%	99%	Cataract 1, multiple types, 116200
GNAT1	81.7	100%	95%	Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	125.7	100%	100%	Achromatopsia-4, 613856
GNPTG	89.9	86%	80%	Mucolipidosis III gamma, 252605
GPR143	27.2	84%	64%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR179	144.9	100%	99%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR98	115.5	99%	98%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
GRK1	95.1	100%	99%	Oguchi disease-2, 613411
GRM6	96.1	95%	92%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	115.7	100%	99%	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706

GSN	75	96%	89%	Amyloidosis, Finnish type, 105120
GUCA1A	67.7	65%	52%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	118.6	100%	100%	Retinitis pigmentosa 48, 613827
GUCY2D	82.5	99%	96%	Leber congenital amaurosis 1, 204000 Cone-rod dystrophy 6, 601777
HARS	123.3	100%	100%	Usher syndrome type 3B, 614504
HCCS	63.9	100%	96%	Microphthalmia, syndromic 7, 309801
HGSNAT	91.5	81%	81%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HK1	111.5	100%	99%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285
HMX1	32.1	84%	67%	Oculoauricular syndrome, 612109
HPS1	74.3	96%	91%	Hermansky-Pudlak syndrome 1, 203300
HPS4	108.4	98%	97%	Hermansky-Pudlak syndrome 4, 614073
HPS5	99.2	96%	96%	Hermansky-Pudlak syndrome 5, 614074
HPS6	85.2	100%	93%	Hermansky-Pudlak syndrome 6, 614075
HSF4	100	100%	98%	Cataract 5, multiple types, 116800
IDH3B	122.4	100%	100%	Retinitis pigmentosa 46, 612572
IFT140	83.3	99%	95%	Mainzer-Saldino syndrome, 266920
IFT172	97.6	100%	97%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 Retinitis pigmentosa 71, 616394
IFT27	101.6	92%	82%	?Bardet-Biedl syndrome 19, 615996
IMPDH1	37.9	87%	65%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
IMPG1	97.1	99%	95%	Macular dystrophy, vitelliform, 4, 616151
IMPG2	125.8	99%	97%	Retinitis pigmentosa 56, 613581 Maculopathy, IMPG2-related, 613581
INPP5E	74.2	100%	99%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INVS	117.6	100%	99%	Nephronophtisis 2, infantile, 602088
IQCB1	95.1	99%	93%	Senior-Loken syndrome 5, 609254
JAG1	110.2	99%	96%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 Deafness, congenital heart defects, and posterior embryotoxon

JAM3	68.5	95%	91%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KCNJ13	216.1	100%	100%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNV2	84.3	100%	99%	Retinal cone dystrophy 3B, 610356
KERA	145.8	100%	100%	Cornea plana congenita, recessive, 217300
KIF11	98.6	99%	96%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF7	67	93%	86%	Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KLHL7	115.7	100%	100%	Retinitis pigmentosa 42, 612943
KRT12	108.2	98%	95%	Meesmann corneal dystrophy, 122100
KRT3	70	100%	100%	Meesmann corneal dystrophy, 122100
LAMA1	97.2	99%	95%	Poretti-Boltshauser syndrome, 615960
LCA5	137.1	100%	98%	Leber congenital amaurosis 5, 604537
LIM2	62.6	78%	76%	Cataract 19, 615277
LRAT	196.5	100%	100%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LRIT3	139.3	94%	94%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP5	89.4	98%	94%	Osteoporosis-pseudoglioma syndrome, 259770 [Bone mineral density variability 1], 601884 Hyperostosis, endosteal, 144750 van Buchem disease, type 2, 607636 Osteosclerosis, 144750 Exudative vitreoretinopathy 4, 60181
LSS	65.4	94%	92%	Cataract 44,616509
LYST	120.5	99%	96%	Chediak-Higashi syndrome, 214500
LZTFL1	90.2	100%	100%	Bardet-Biedl syndrome 17, 615994
MAB21L2	134.3	100%	100%	Microphthalmia, syndromic 14, 615877
MAF	80.2	80%	74%	Cataract, pulverulent or cerulean, with or without microcornea, 610202
MAK	86.6	95%	93%	Retinitis pigmentosa 62, 614181
MERTK	114.9	100%	99%	Retinitis pigmentosa 38, 613862
MFN2	103.5	100%	97%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VI, 601152

MFRP	87.8	100%	99%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	120	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951
MIP	76.1	100%	97%	Cataract 15, multiple types, 615274
MITF	128.9	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	133.8	89%	89%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 209900
MKS1	112.4	100%	99%	Meckel syndrome 1, 249000
MVK	88.3	100%	97%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
MYO7A	77	95%	91%	Usher syndrome, type 1B, 276900 Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317
MYOC	182.9	100%	100%	Glaucoma 1A, primary open angle, 137750
NAA10	50.9	97%	86%	?Microphthalmia, syndromic 1, 309800 N-terminal acetyltransferase deficiency, 300855
NDP	52.2	86%	78%	Norrie disease, 310600 Exudative vitreoretinopathy, X-linked, 305390
NEK2	27.6	47%	42%	?Retinitis pigmentosa 67, 615565
NEUROD1	130.4	100%	100%	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853
NHS	65.7	92%	88%	Nance-Horan syndrome, 302350
NMNAT1	93.1	100%	100%	Leber congenital amaurosis 9, 608553
NPHP1	174.4	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	108.9	100%	99%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	89.5	98%	93%	Nephronophthisis 4, 606966

				Senior-Loken syndrome 4, 606996
NR2F1	164.8	100%	100%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRL	47.6	100%	98%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type
NYX	47.7	97%	94%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	44.9	83%	71%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCA2	94.5	100%	99%	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	37.5	88%	78%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804
OPA1	131.1	99%	99%	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250
OPA3	108	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPN1LW	0.4	0%	0%	Colorblindness, protan, 303900 Blue cone monochromacy, 303700
OPN1MW	0.5	0%	0%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700
OR2W3	176.1	100%	100%	No OMIM disease ID
OTX2	158.2	100%	100%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, and pituitary dysfunction, 610125
P3H2	70.5	99%	88%	Myopia, high, with cataract and vitreoretinal degeneration,614292
PANK2	112.1	90%	86%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PAX2	105.5	96%	94%	Papillorenal syndrome, 120330 Renal hypoplasia, isolated, 191830

PAX6	83.9	100%	100%	Aniridia, 106210 Peters anomaly, 604229 Cataract with late-onset corneal dystrophy, 106210 Keratitis, 148190 Foveal hyperplasia, 136520 Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550 Coloboma, ocular, 120200
PCDH15	134.8	99%	99%	Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067
PCYT1A	90	100%	98%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE6A	100.7	99%	95%	Retinitis pigmentosa 43, 613810
PDE6B	100.3	100%	99%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	109.6	100%	99%	Cone dystrophy 4, 613093
PDE6D	110.2	100%	100%	?Joubert syndrome 22, 615665
PDE6G	71.8	100%	95%	Retinitis pigmentosa 57, 613582
PDE6H	40	87%	75%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDZD7	69.3	95%	88%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PET100	76.3	100%	99%	Mitochondrial complex IV deficiency, 220110
PEX1	124.5	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX2	159.3	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX7	93.8	90%	85%	Chondrodysplasia punctata, rhizomelic, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PGK1	42.7	75%	67%	Phosphoglycerate kinase 1 deficiency, 300653
PHYH	87.7	100%	99%	Refsum disease, 266500
PIKFYVE	137.9	100%	100%	Corneal fleck dystrophy, 121850

PITX2	120.6	96%	87%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PITX3	37.2	99%	84%	Anterior segment mesenchymal dysgenesis, 107250
PLA2G5	108.2	100%	100%	Fleck retina, familial benign, 228980
PLK4	142.6	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PNPLA6	82.5	100%	97%	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
POC1B	101.2	99%	99%	Cone-rod dystrophy 20, 615973
PPT1	72	100%	96%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	87	100%	100%	Retinitis pigmentosa 36, 610599
PROM1	85.6	99%	93%	Retinitis pigmentosa 41, 612095 Cone-rod dystrophy 12, 612657 Stargardt disease 4, 603786 Macular dystrophy, retinal, 2, 608051
PRPF3	96.8	100%	100%	Retinitis pigmentosa 18, 601414
PRPF31	82.2	86%	83%	Retinitis pigmentosa 11, 600138
PRPF4	114.4	100%	97%	Retinitis pigmentosa 70, 615922
PRPF6	91	100%	100%	Retinitis pigmentosa 60, 613983
PRPF8	120.7	99%	98%	Retinitis pigmentosa 13, 600059
PRPH2	147.4	100%	100%	Retinitis pigmentosa 7, 608133 Retinitis punctata albescens, 136880 Macular dystrophy, patterned, 169150 Macular dystrophy, vitelliform, 608161 Foveomacular dystrophy, adult-onset, with choroidal neovascularization, 608161
PRSS56	60.4	97%	85%	Microphthalmia, isolated 6, 613517
PXDN	101.2	99%	93%	Corneal opacification and other ocular anomalies, 269400
RAB28	78.5	96%	94%	Cone-rod dystrophy 18, 615374
RARB	148	100%	100%	Microphthalmia, syndromic 12, 615524
RAX2	57.2	100%	99%	Cone-rod dystrophy 11, 610381 Macular degeneration, age-related, 6,613757

RBP3	103.7	100%	99%	?Retinitis pigmentosa 66, 615233
RBP4	76.7	95%	90%	Retinol dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RD3	56.9	100%	100%	Leber congenital amaurosis 12, 610612
RDH11	97.6	100%	95%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	63.6	91%	80%	Leber congenital amaurosis 13, 612712
RDH5	107.3	100%	100%	Fundus albipunctatus, 136880
RGR	82.6	95%	86%	Retinitis pigmentosa 44, 613769
RGS9	107.3	98%	94%	Bradyopsia, 608415
RGS9BP	37.4	100%	94%	Bradyopsia, 608415
RHO	125.6	100%	98%	Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880
RIMS1	103.4	99%	99%	Cone-rod dystrophy 7, 603649
RLBP1	101.2	100%	98%	Fundus albipunctatus, 136880 Retinitis punctata albescens, 136880 Newfoundland rod-cone dystrophy, 607476 Bothnia retinal dystrophy, 607475
ROM1	92.5	100%	100%	Retinitis pigmentosa 7, digenic, 608133
RP1	179.8	100%	100%	Retinitis pigmentosa 1, 180100 {Hypertriglyceridemia, susceptibility to}, 145750
RP1L1	128.3	100%	100%	Occult macular dystrophy, 613587
RP2	60.2	100%	98%	Retinitis pigmentosa 2, 312600
RP9	25	50%	43%	Retinitis pigmentosa 9, 180104
RPE65	114.7	100%	98%	Leber congenital amaurosis 2, 204100
RPGR	70	85%	78%	Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834 Cone-rod dystrophy, X-linked, 1, 304020
RPGRIP1	118	100%	98%	Leber congenital amaurosis 6, 613826 Cone-rod dystrophy 13, 608194
RPGRIP1L	101.7	98%	96%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RS1	39.8	92%	82%	Retinoschisis, 312700

SAG	110.8	99%	99%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SDCCAG8	104.3	100%	99%	Senior-Loken syndrome 7, 613615
SEMA4A	100	99%	95%	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283 -3
SHH	95.9	99%	85%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SIPA1L3	107.1	98%	96%	No OMIM disease ID
SIX6	128.1	100%	97%	Microphthalmia with cataract 2, 212550
SLC24A1	143.4	100%	99%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	117.4	100%	96%	Albinism, oculocutaneous, type VI, 113750
SLC33A1	100.8	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC38A8	65.5	98%	86%	Foveal hypoplasia 2, optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC45A2	106.9	99%	95%	Albinism, oculocutaneous, type IV, 606574
SLC4A11	110	99%	98%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy 2, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC52A2	116.3	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC7A14	142.2	100%	99%	Retinitis pigmentosa 68, 615725
SNRNP200	112.4	100%	98%	Retinitis pigmentosa 33, 610359
SOX2	129.8	100%	100%	Retinitis pigmentosa 33, 610359
SPATA7	126.6	100%	100%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPP2	106.9	100%	100%	No OMIM disease ID
STRA6	73.8	100%	96%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
TACSTD2	182.1	100%	97%	Corneal dystrophy, gelatinous drop-like, 204870
TCTN1	105.4	95%	95%	Joubert syndrome 13, 614173
TCTN3	99.1	100%	99%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TDRD7	116.4	100%	100%	Cataract 36, 613887

TEAD1	89.6	100%	99%	Sveinsson choreoretinal atrophy, 108985
TENM3	131.3	100%	99%	Microphthalmia, isolated, with coloboma 9, 61545
TGFBI	106.1	100%	100%	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	26.2	73%	66%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TIMP3	123	100%	100%	Sorsby fundus dystrophy, 136900
TMEM126A	85.8	100%	99%	Optic atrophy-7, 612989
TMEM138	96.9	100%	100%	Joubert syndrome 16, 614465
TMEM231	68.2	96%	88%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397
TMEM237	101.7	100%	94%	Joubert syndrome 14, 614424
TMEM67	116.7	100%	99%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TOPORS	159.4	100%	100%	Retinitis pigmentosa 31, 609923
TPP1	135.4	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500
TREX1	120.7	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	106.2	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 Bardet-Biedl syndrome 11, 209900
TRNT1	113.1	100%	99%	Sideroblastic anemia with B-cell immunodeficiency,periodic fevers and developmental delay,616084
TRPM1	129.2	98%	98%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TSPAN12	117.9	100%	100%	Exudative vitreoretinopathy 5, 613310

TTC8	107.7	100%	100%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
TTLL5	120.9	100%	100%	Cone-rod dystrophy 19,615860
TUB	80.9	100%	94%	?Retinal dystrophy and obesity, 616188
TUBGCP4	115.1	99%	96%	Microcephaly and chorioretinopathy,autosomal recessive,616335
TULP1	84.8	100%	93%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
TYR	132.9	74%	74%	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}
TYRP1	125.9	100%	100%	Albinism, oculocutaneous, type III, 203290
UBIAD1	95	100%	100%	Corneal dystrophy, Schnyder type, 121800
UNC119	113	100%	100%	?Cone-rod dystrophy ?Immunodeficiency 13, 615518
UNC45B	92.4	99%	97%	?Cataract 43,616279
USH1C	77.9	99%	92%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904
USH1G	98.7	95%	88%	Usher syndrome, type 1G, 606943
USH2A	116.2	100%	99%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809
VAX1	75.1	100%	91%	Microphthalmia, syndromic 11, 614402
VCAN	147.3	100%	100%	Wagner syndrome 1, 143200
VIM	97.7	100%	100%	Cataract 30, pulverulent, 116300
VSX1	65.7	99%	91%	Corneal dystrophy, posterior polymorphous, 1, 122000 Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300
VSX2	62.5	100%	94%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
WDPCP	92.7	98%	97%	?Bardet-Biedl syndrome 15, 615992
WDR19	123.9	100%	100%	Asphyxiating thoracic dystrophy 5, 614376 Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378

WFS1	145.6	100%	98%	?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	140.5	100%	99%	Werner syndrome, 277700
YAP1	76.6	96%	88%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation
ZEB1	152.1	98%	97%	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZNF408	87.3	97%	92%	?Exudative vitreoretinopathy, 616468 Retinitis pigmentosa 72, 616469
ZNF423	127.9	100%	99%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF513	99.1	100%	96%	Retinitis pigmentosa 58, 613617
ZNF644	169	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
OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015
This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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