VISION DISORDERS GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ABCA4	96,1	99%	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
ABCB6	132,1	100%	100%	Microphthalmia, isolated, with coloboma 7, 614497 [Blood group, Langereis system], 111600 Dyschromatosis universalis hereditaria 3, 615402
ABCC6	53,7	72%	69%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABHD12	62,1	99%	94%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ADAM9	116,9	100%	99%	Cone-rod dystrophy 9, 612775
ADAMTS18	105,2	100%	98%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
AGK	115	100%	99%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AHI1	119,8	100%	99%	Joubert syndrome-3, 608629
AIPL1	95,3	100%	99%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
ALMS1	206,7	98%	98%	Alstrom syndrome, 203800

ARL13B	132,1	100%	98%	Joubert syndrome 8, 612291
ARL2BP	96,4	100%	96%	Retinitis pigmentosa with or without situs inversus, 615434
ARL6	155,8	100%	100%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
BBS1	127,4	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	135,2	100%	100%	Bardet-Biedl syndrome 10, 209900
BBS12	160	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	118,5	100%	100%	Bardet-Biedl syndrome 2, 209900
BBS4	99,8	98%	92%	Bardet-Biedl syndrome 4, 209900
BBS5	142,1	100%	100%	Bardet-Biedl syndrome 5, 209900
BBS7	130,3	100%	99%	Bardet-Biedl syndrome 7, 209900
BBS9	134,9	100%	100%	Bardet-Biedl syndrome 9, 209900
BCOR	69,2	98%	94%	Microphthalmia, syndromic 2, 300166
BEST1	122,1	100%	97%	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 Retinitis pigmentosa-50, 613194 Retinitis pigmentosa, concentric, 613194

BFSP1	131,1	100%	100%	Cataract 33, 611391
BFSP2	68,9	96%	89%	Cataract 12, multiple types, 611597
BMP4	124,6	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625 -3
C1QTNF5	131,1	97%	80%	Retinal degeneration, late-onset, autosomal dominant, 605670
C21orf2	80,8	99%	95%	No OMIM phenotype Cone-Rod dystrophy (Abu-Safieh (2013) Genome Res 23. 236)
C2ORF71	129,3	99%	94%	Retinitis pigmentosa 54, 613428
C5ORF42	133,7	100%	100%	Joubert syndrome 17, 614615
C8ORF37	91,8	100%	100%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500 -3
CA4	104,1	100%	98%	Retinitis pigmentosa 17, 600852
CABP4	73,9	100%	99%	Night blindness, congenital stationary (incomplete), 2B, autosomal recessive, 610427
CACNA1F	48,9	94%	87%	Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Cone-rod dystropy, X-linked, 3, 300476 Aland Island eye disease, 300600
CACNA2D4	85,3	98%	95%	Retinal cone dystrophy 4, 610478
CAPN5	87,2	100%	97%	Vitreoretinopathy, neovascular inflammatory, 193235
CC2D2A	103,3	99%	99%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CDH23	103,3	99%	98%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CDH3	105,3	99%	95%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280

CDHR1	117,8	98%	97%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660 -3
CEP164	89,8	99%	94%	Nephronophthisis 15, 614845
CEP250	86,4	97%	94%	No OMIM phenotype Usher Syndrome, atypical (Khateb (2014) J Med Genet 51,460)
CEP290	104,1	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	89,9	100%	100%	Joubert syndrome 15, 614464
CERKL	141,9	100%	100%	Retinitis pigmentosa 26, 608380
CFH	108,8	96%	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	54,1	99%	92%	Choroideremia, 303100
CHMP4B	120,4	100%	100%	Cataract 31, multiple types, 605387
CIB2	125,1	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLN3	94,6	100%	100%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	135,9	98%	95%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	73,2	99%	92%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	139,7	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLRN1	174,6	100%	100%	?digenic interaction with MYO7A Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180

CNGA1	125,9	91%	90%	Retinitis pigmentosa 49, 613756
CNGA3	149	98%	97%	Achromatopsia-2, 216900
CNGB1	93	99%	93%	Retinitis pigmentosa 45, 613767
CNGB3	112,1	99%	94%	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200
CNNM4	172	99%	97%	Jalili syndrome, 217080
COL11A1	98,8	98%	98%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	13,2	56%	22%	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	93,2	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 Spondyloperipheral dysplasia, 271700 SED, Namaqualand type Osteoarthritis with mild chondrodysplasia, 604864 Vitreoretinopathy with phalangeal epiphyseal dysplasia Platyspondylic skeletal dysplasia, Torrance type, 151210 Otospondylomegaepiphyseal dysplasia, 215150 Avascular necrosis of the femoral head, 608805 Legg-Calve-Perthes disease, 150600 Stickler sydrome, type I, nonsyndromic ocular, 609508 Czech dysplasia, 609162

COL9A1	112,6	98%	96%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	77,8	98%	94%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284

CRB1	170,9	100%	100%	Retinitis pigmentosa-12, autosomal recessive, 600105 Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870
CRX	161,5	100%	100%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	124,8	100%	100%	Cataract 9, multiple types, 604219
CRYAB	147	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	112,1	100%	100%	Cataract 10, multiple types, 600881
CRYBA4	77,3	100%	99%	Cataract 23, 610425
CRYBB1	59,7	100%	93%	Cataract 17, multiple types, 611544
CRYBB2	111,4	100%	100%	Cataract 3, multiple types, 601547
CRYBB3	104,2	100%	100%	Cataract 22, autosomal recessive, 609741
CRYGB	72,1	100%	94%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	81,3	100%	97%	Cataract 2, multiple types, 604307
CRYGD	81,4	85%	79%	Cataract 4, multiple types, 115700
CRYGS	104,6	95%	89%	Cataract 20, multiple types, 116100

CSPP1	126,7	100%	100%	Joubert syndrome 21, 615636
CTDP1	82,2	89%	86%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTSD	98,3	99%	94%	Ceroid lipofuscinosis, neuronal, 10, 610127
CYP4V2	121,3	100%	100%	Bietti crystalline corneoretinal dystrophy, 210370
DFNB31	89,2	100%	98%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DHDDS	91,2	100%	98%	Retinitis pigmentosa 59, 613861
DHX38	89,9	99%	93%	No OMIM phenotype Retinitis pigmentosa, early-onset with macular coloboma (Ajmal (2014) J Med Genet 51,444)
EFEMP1	123	100%	100%	Doyne honeycomb degeneration of retina, 126600
ELOVL4	108,5	100%	100%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
EMC1	104,2	100%	97%	No OMIM phenotype Retinitis pigmentosa (Abu-Safieh (2013) Genome Res 23, 236) Schizophrenia (Fromer (2014) Nature 506,179)
EPG5	95,1	100%	99%	Vici syndrome, 242840
EPHA2	101,5	98%	95%	Cataract 6, multiple types, 116600
EYA1	116,9	100%	100%	Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Otofaciocervical syndrome, 166780
EYS	138,9	100%	99%	Retinitis pigmentosa 25, 602772
FAM161A	151,4	100%	100%	Retinitis pigmentosa 28, 606068
FLVCR1	105,2	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033

FOXE3	22,9	94%	53%	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256
FSCN2	88,1	100%	100%	Retinitis pigmentosa 30, 607921
FTL	103,6	100%	99%	Epilepsy, familial temporal lobe, 2 (2)
FTL	103,6	100%	99%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
FYCO1	103,4	100%	99%	Cataract 18, autosomal recessive, 610019
FZD4	144,2	100%	100%	Exudative vitreoretinopathy, 133780 Retinopathy of prematurity, 133780
GALK1	108,3	100%	100%	Galactokinase deficiency with cataracts, 230200
GALT	126,9	100%	100%	Galactosemia, 230400
GCNT2	173	100%	100%	[Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 110800 Adult i phenotype without cataract, 110800
GDF6	148,2	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	65,4	99%	93%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GJA1	60,1	92%	83%	Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Hypoplastic left heart syndrome 1, 241550 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, autosomal recessive, 257850 Craniometaphyseal dysplasia, autosomal recessive, 218400

GJA3	94,1	100%	99%	Cataract 14, multiple types, 601885
GJA8	64,3	100%	82%	Cataract 1, multiple types, 116200
GNAT1	96,3	100%	98%	Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	123,6	100%	100%	Achromatopsia-4, 613856
GPR125	96,3	89%	86%	No OMIM phenotype Retinitis pigmentosa (Abu-afieh (2013) Genome Res 23,236)
GPR179	158,1	100%	99%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR98	118,4	99%	98%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
GRK1	109,4	100%	100%	Oguchi disease-2, 613411
GRM6	102,4	95%	93%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	127,5	100%	100%	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
GUCA1A	68	65%	58%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	123,7	100%	100%	Retinitis pigmentosa 48, 613827
GUCY2D	89,1	99%	98%	Leber congenital amaurosis 1, 204000 Cone-rod dystrophy 6, 601777
HARS	130,8	100%	95%	Usher syndrome type 3B, 614504
HCCS	64,3	99%	92%	Microphthalmia, syndromic 7, 309801
HMX1	35,7	92%	83%	Oculoauricular syndrome, 612109
HSF4	110,2	100%	98%	Cataract 5, multiple types, 116800
IDH3B	144,1	100%	100%	Retinitis pigmentosa 46, 612572

IFT140	93,6	99%	91%	Mainzer-Saldino syndrome, 266920
IFT172	101,4	100%	96%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 Mainzer-Saldino syndrome (Halbritter (2013) Am J Hum Genet 93, 915) Asphyxiating thoracic dystrophy with or without Joubert Syndrome (Halbritter (2013) Am J Hum Genet 93, 915)
IMPDH1	42,6	79%	66%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
IMPG2	130,2	99%	97%	Retinitis pigmentosa 56, 613581 Maculopathy, IMPG2-related, 613581
INPP5E	81,6	99%	97%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
IQCB1	93,6	97%	88%	Senior-Loken syndrome 5, 609254
JAG1	110,5	97%	96%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 Deafness, congenital heart defects, and posterior embryotoxon
JAM3	82,4	98%	89%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KCNJ13	244,6	100%	100%	Snowflake vitreoretinal degeneration, 193230 Leber congenital amaurosis 16, 614186
KCNV2	98,2	100%	100%	Retinal cone dystrophy 3B, 610356
KIAA1549	118,3	97%	94%	No OMIM phenotype Autism (Neale (2012) Nature 485,242) Retinitis pigmentosa (Abu-Safieh (2013) Genome Res 23,236)
KIF11	103,6	100%	99%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF7	80,4	94%	89%	Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KLHL7	123	100%	100%	Retinitis pigmentosa 42, 612943
LCA5	148,7	100%	98%	Leber congenital amaurosis 5, 604537

LIM2	61,2	77%	76%	Cataract 19, 615277

LRAT	190,6	100%	100%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LRIT3	157,4	94%	93%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP5	100,7	97%	95%	Osteoporosis-pseudoglioma syndrome, 259770 [Bone mineral density variability 1], 601884 Hyperostosis, endosteal, 144750 van Buchem disease, type 2, 607636 Osteosclerosis, 144750 {Osteoporosis}, 166710 Exudative vitreoretinopathy 4, 601813 Osteopetrosis, autosomal dominant 1, 607634
LZTFL1	90,7	100%	100%	Bardet-Biedl syndrome 17, 615994
MAF	79,9	83%	74%	Cataract, pulverulent or cerulean, with or without microcornea, 610202
MAK	83,1	96%	92%	Retinitis pigmentosa 62, 614181
MERTK	128,7	100%	99%	Retinitis pigmentosa 38, 613862
MFN2	111,2	100%	99%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VI, 601152
MFRP	91,8	100%	99%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	129,3	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951
MIP	95,6	100%	99%	Cataract 15, multiple types, 615274
MKKS	153,8	100%	100%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 209900
MKS1	111,2	100%	97%	Meckel syndrome 1, 249000 Bardet-Biedl syndrome 13, 209900

MVK	94	100%	99%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
MYO7A	86,9	97%	92%	Usher syndrome, type 1B, 276900 Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317
NDP	46,8	97%	72%	Norrie disease, 310600 Exudative vitreoretinopathy, X-linked, 305390
NEK2	27,4	50%	38%	?Retinitis pigmentosa 67, 615565
NHS	71,5	94%	91%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NMNAT1	100,1	100%	100%	Leber congenital amaurosis 9, 608553
NPHP1	117,9	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	118,8	100%	100%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	91,5	99%	96%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NR2E3	89,1	98%	92%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	162,9	100%	99%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRL	57,9	100%	100%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type
NYX	54,7	96%	94%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	47,3	86%	66%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OFD1	38,9	87%	77%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804

OPA1	135,7	100%	98%	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250
OPA3	106,3	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPN1LW	1,2	1%	0%	Colorblindness, protan, 303900 Blue cone monochromacy, 303700 -3
OPN1MW	0,4	0%	0%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700 -3
OTX2	181,6	100%	100%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, and pituitary dysfunction, 610125
PANK2	126,3	100%	99%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PAX2	117,7	98%	96%	Papillorenal syndrome, 120330 Renal hypoplasia, isolated, 191830 -3
PAX6	98,6	100%	99%	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 Coloboma of optic nerve, 120430 Gillespie syndrome, 206700
PCDH15	138,4	100%	99%	Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067
PCYT1A	90,4	100%	94%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE6A	106,9	97%	96%	Retinitis pigmentosa 43, 613810
PDE6B	104,2	100%	96%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801

PDE6C	111,8	100%	99%	Cone dystrophy 4, 613093
PDE6D	98	100%	100%	?Joubert syndrome 22, 615665
PDE6G	94,5	100%	100%	Retinitis pigmentosa 57, 613582
PDE6H	28,9	99%	71%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDZD7	80,9	95%	84%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PITPNM3	84	98%	94%	Cone-rod dystrophy 5, 600977
PITX3	42,2	100%	94%	Anterior segment mesenchymal dysgenesis, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623
PLA2G5	106,9	100%	100%	Fleck retina, familial benign, 228980
PPT1	77,4	100%	99%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	109,6	100%	100%	Retinitis pigmentosa 36, 610599
PROM1	91	100%	98%	Retinitis pigmentosa 41, 612095 Cone-rod dystrophy 12, 612657 Stargardt disease 4, 603786 Macular dystrophy, retinal, 2, 608051
PRPF3	105,6	100%	100%	Retinitis pigmentosa 18, 601414
PRPF31	94	86%	83%	Retinitis pigmentosa 11, 600138
PRPF4	123,7	100%	99%	Retinitis pigmentosa 70, 615922
PRPF6	92,5	100%	99%	Retinitis pigmentosa 60, 613983
PRPF8	128,8	99%	98%	Retinitis pigmentosa 13, 600059

PRPH2	155,8	100%	99%	Retinitis pigmentosa 7, 608133 Retinitis punctata albescens, 136880 Macular dystrophy, patterned, 169150 Macular dystrophy, vitelliform, 608161 Foveomacular dystrophy, adult-onset, with choroidal neovascularization, 608161 Macular dystrophy Retinitis pigmentosa, digenic, 608133 Choriodal dystrophy, central areolar 2, 613105
PRSS56	78,5	98%	92%	Microphthalmia, isolated 6, 613517
RAB28	84,4	100%	93%	Cone-rod dystrophy 18, 615374
RARB	156,2	100%	100%	Microphthalmia, syndromic 12, 615524
RBP3	111,2	100%	100%	?Retinitis pigmentosa 66, 615233
RBP4	88,3	95%	88%	Retinol dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RD3	74,7	100%	99%	Leber congenital amaurosis 12, 610612
RDH12	61,1	98%	90%	Leber congenital amaurosis 13, 612712
RDH5	109,3	100%	100%	Fundus albipunctatus, 136880
RGR	82,7	93%	76%	Retinitis pigmentosa 44, 613769
RGS9	111,5	98%	94%	Bradyopsia, 608415
RGS9BP	54,8	100%	94%	Bradyopsia, 608415
RHO	139,9	100%	100%	Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880
RIMS1	112,4	100%	100%	Cone-rod dystrophy 7, 603649

RLBP1	111,7	100%	99%	Fundus albipunctatus, 136880 Retinitis punctata albescens, 136880 Newfoundland rod-cone dystrophy, 607476 Bothnia retinal dystrophy, 607475
RLBP1	111,7	100%	99%	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
ROM1	96,8	100%	100%	Retinitis pigmentosa 7, digenic, 608133
RP1	189,1	100%	100%	Retinitis pigmentosa 1, 180100 {Hypertriglyceridemia, susceptibility to}, 145750
RP1L1	134,2	100%	100%	Occult macular dystrophy, 613587
RP2	63	100%	100%	Retinitis pigmentosa 2, 312600
RP9	25,8	59%	43%	Retinitis pigmentosa 9, 180104
RPE65	120	100%	100%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	55,9	82%	76%	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	130,1	100%	99%	Leber congenital amaurosis 6, 613826 Cone-rod dystrophy 13, 608194
RPGRIP1L	108,6	98%	96%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RS1	49	97%	88%	Retinoschisis, 312700
SAG	116,3	100%	100%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SDCCAG8	105,2	100%	100%	Senior-Loken syndrome 7, 613615

SEMA4A	104,8	99%	96%	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283 -3
SHH	113,1	99%	91%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SIX6	153,3	100%	98%	Microphthalmia with cataract 2, 212550
SLC24A1	159,6	100%	100%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC33A1	100,8	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SNRNP200	119,3	99%	98%	Retinitis pigmentosa 33, 610359
SOX2	160	100%	100%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SPATA7	136,5	100%	97%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
STRA6	73,6	99%	97%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
TCTN3	107,8	100%	98%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TDRD7	131,3	100%	100%	Cataract 36, 613887
TEAD1	91	100%	100%	Sveinsson choreoretinal atrophy, 108985
TIMM8A	35,2	99%	72%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TIMP3	139,1	100%	100%	Sorsby fundus dystrophy, 136900
TMEM126A	90,9	100%	99%	Optic atrophy-7, 612989
TMEM138	99,9	100%	100%	Joubert syndrome 16, 614465

TMEM231	71,1	95%	85%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397 -3
TMEM237	98,6	100%	97%	Joubert syndrome 14, 614424
TMEM67	125,3	100%	98%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TOPORS	165,3	100%	100%	Retinitis pigmentosa 31, 609923
TPP1	145,4	100%	97%	Ceroid lipofuscinosis, neuronal, 2, 204500
TREX1	147,2	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	125,9	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 Bardet-Biedl syndrome 11, 209900
TRPM1	139,1	100%	99%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TSPAN12	128,7	100%	100%	Exudative vitreoretinopathy 5, 613310
TTC8	114,3	100%	100%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
TTLL5	128,8	100%	99%	Cone-Rod dystrophy 19, 615860
TUB	89,6	100%	99%	No OMIM phenotype Retinal dystrophy and obesity (Borman (2014) Hum Mutat 35,289)
TULP1	94,7	100%	96%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
UNC119	123,6	100%	100%	Cone-rod dystrophy
USH1C	79,4	99%	96%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092

USH1G	114	99%	91%	Usher syndrome, type 1G, 606943
USH2A	123,8	100%	99%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809 -3

VAX1	78,6	100%	97%	Microphthalmia, syndromic 11, 614402
VCAN	157,1	100%	100%	Wagner syndrome 1, 143200
VIM	98	100%	100%	Cataract 30, pulverulent, 116300
WDR19	130,6	100%	100%	Asphyxiating thoracic dystrophy 5, 614376 Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378
WFS1	155	100%	100%	Wolfram syndrome, 222300 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
ZNF423	144,8	100%	99%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF513	110,1	99%	97%	Retinitis pigmentosa 58, 613617
ZNF644	174,8	100%	100%	Myopia 21, autosomal dominant, 614167

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
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 : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors