VISION GENE PANEL DGD141114

Gene	Median coverage	% covered >10x	% covered >20x	Associated Phenotype description and OMIM ID
ABCA4	87.8	99%	99%	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	118.6	100%	100%	Microphthalmia, isolated, with coloboma 7, 614497 [Blood group, Langereis system], 111600 Dyschromatosis universalis hereditaria 3, 615402
ABCC6	52.4	73%	73%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABHD12	61.0	98%	98%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACBD5	105.3	100%	100%	No OMIM disease ID Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) Cone-rod dystrophy (Aby-Safieh (2013) Genome Res 23,236)
ADAM9	118.1	100%	100%	Cone-rod dystrophy 9, 612775
ADAMTS18	99.3	100%	100%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
AGK	106.6	100%	100%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AHI1	115.0	100%	100%	Joubert syndrome-3, 608629
AIPL1	85.4	100%	100%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
ALMS1	195.4	98%	98%	Alstrom syndrome, 203800
APOPT1	100.9	100%	100%	Mitochondrial complex IV deficiency, 220110
ARL13B	121.2	99%	99%	Joubert syndrome 8, 612291
ARL2BP	85.2	100%	100%	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	88.6	99%	99%	Traboulsi syndrome, 601552
BBS1	114.8	100%	100%	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	115.0	100%	100%	Bardet-Biedl syndrome 2, 209900
BBS4	97.2	97%	97%	Bardet-Biedl syndrome 4, 209900
BBS5	150.6	100%	100%	Bardet-Biedl syndrome 5, 209900
BBS7	129.6	100%	100%	Bardet-Biedl syndrome 7, 209900
BBS9	124.3	100%	100%	Bardet-Biedl syndrome 9, 209900
BCOR	61.8	99%	99%	Microphthalmia, syndromic 2, 300166
BEST1	106.5	100%	100%	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	125.0	100%	100%	Cataract 33, 611391
BFSP2	62.7	94%	94%	Cataract 12, multiple types, 611597
BMP4	114.2	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625 -3
C19ORF12	72.8	100%	100%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
C1QTNF5	119.3	98%	98%	Retinal degeneration, late-onset, autosomal dominant, 605670
C21orf2	63.2	99%	99%	No OMIM disease ID Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
C2ORF71	105.5	99%	99%	Retinitis pigmentosa 54, 613428
C5ORF42	129.8	100%	100%	Joubert syndrome 17, 614615
C8ORF37	95.2	100%	100%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500
CA4	83.8	98%	98%	Retinitis pigmentosa 17, 600852
CABP4	64.3	100%	100%	Night blindness, congenital stationary (incomplete), 2B, autosomal recessive, 610427
CACNA1F	42.4	94%	94%	Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Cone-rod dystropy, X-linked, 3, 300476 Aland Island eye disease, 300600
CACNA2D4	78.1	98%	98%	Retinal cone dystrophy 4, 610478

CAPN5	78.6	100%	100%	Vitreoretinopathy, neovascular inflammatory, 193235
CC2D2A	94.3	99%	99%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CDH23	94.7	99%	99%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CDH3	96.9	97%	97%	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	76.6	98%	98%	Nephronophthisis 15, 614845
CEP250	81.0	99%	99%	No OMIM disease ID Usher syndrome, atypical (Khateb (2014) J Med Genet 51,460)
CEP290	101.5	100%	100%	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900
CEP41	84.5	100%	100%	Joubert syndrome 15, 614464
CERKL	137.4	100%	100%	Retinitis pigmentosa 26, 608380
CFH	107.3	95%	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	52.7	98%	98%	Choroideremia, 303100
CHMP4B	114.9	100%	100%	Cataract 31, multiple types, 605387
CIB2	110.1	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLN3	86.5	100%	100%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	139.7	100%	100%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	63.7	98%	98%	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	165.9	100%	100%	?digenic interaction with MYO7A Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180

CNGA1	119.8	91%	91%	Retinitis pigmentosa 49, 613756
CNGA3	135.6	100%	100%	Achromatopsia-2, 216900
CNGB1	84.8	96%	96%	Retinitis pigmentosa 45, 613767
CNGB3	108.7	100%	100%	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200
CNNM4	170.4	99%	99%	Jalili syndrome, 217080
COL11A1	98.0	98%	98%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	14.4	61%	61%	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%	99%	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	108.4	100%	100%	Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	72.9	98%	98%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284

CRB1	165.5	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	108.4	100%	100%	Cataract 9, multiple types, 604219
CRYAB	133.3	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	91.0	100%	100%	Cataract 10, multiple types, 600881
CRYBA4	79.0	100%	100%	Cataract 23, 610425
CRYBB1	56.9	100%	100%	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%	100%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	84.1	100%	100%	Cataract 2, multiple types, 604307
CRYGD	76.2	90%	90%	Cataract 4, multiple types, 115700
CRYGS	92.6	98%	98%	Cataract 20, multiple types, 116100
CSPP1	122.7	100%	100%	Joubert syndrome 21, 615636
CTDP1	70.3	89%	89%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTSD	96.2	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CYP4V2	124.7	100%	100%	Bietti crystalline corneoretinal dystrophy, 210370
DFNB31	85.0	100%	100%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DHDDS	76.9	100%	100%	Retinitis pigmentosa 59, 613861
DHX38	81.7	97%	97%	No OMIM disease ID Retinitis pigmentosa, early-onset with macular coloboma (Ajmal (2014) J Med Genet 51,444)
DTHD1	147.5	100%	100%	No OMIM disease ID Leber congenital amaurosis with myopathy (Aby-Safieh (2013) Genome Res 23,236)
EFEMP1	124.3	100%	100%	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

EMC1	97.9	99%	99%	No OMIM disease ID Retinitis pigmentosa (Abu-Safieh (2013) Genome Res 23, 236) Schizophrenia (Fromer (2014) Nature 506,179)
EPG5	89.4	100%	100%	Vici syndrome, 242840
EPHA2	89.5	97%	97%	Cataract 6, multiple types, 116600
EYS	127.6	100%	100%	Retinitis pigmentosa 25, 602772
FA2H	64.6	87%	87%	Spastic paraplegia 35, autosomal recessive, 612319
FAM161A	145.1	100%	100%	Retinitis pigmentosa 28, 606068
FLVCR1	95.8	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOXE3	14.2	70%	70%	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256
FSCN2	82.2	100%	100%	Retinitis pigmentosa 30, 607921
FTL	95.3	100%	100%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
FYCO1	86.2	99%	99%	Cataract 18, autosomal recessive, 610019
FZD4	142.2	100%	100%	Exudative vitreoretinopathy, 133780 Retinopathy of prematurity, 133780
GALK1	96.4	100%	100%	Galactokinase deficiency with cataracts, 230200
GALT	115.6	100%	100%	Galactokinase deficiency with cataracts, 230200
GCNT2	161.3	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	63.9	99%	99%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GJA1	63.1	91%	91%	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	100.0	100%	100%	Cataract 14, multiple types, 601885
GNAT1	81.7	100%	100%	Night blindness, congenital stationary, autosomal dominant 3, 610444
GNAT2	125.7	100%	100%	Achromatopsia-4, 613856
GNPTG	89.9	86%	86%	Mucolipidosis III gamma, 252605
GPR125	91.2	90%	90%	No OMIM phenotype Retinitis pigmentosa (Abu-afieh (2013) Genome Res 23,236)
GPR179	144.9	100%	100%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR98	115.4	99%	99%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
GRK1	95.1	100%	100%	Oguchi disease-2, 613411
GRM6	96.1	95%	95%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	115.7	100%	100%	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
GUCA1A	67.7	65%	65%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	118.6	100%	100%	Retinitis pigmentosa 48, 613827
GUCY2D	82.5	99%	99%	Leber congenital amaurosis 1, 204000 Cone-rod dystrophy 6, 601777
HARS	123.3	100%	100%	Usher syndrome type 3B, 614504
HCCS	63.9	100%	100%	Microphthalmia, syndromic 7, 309801
HMX1	37.6	99%	99%	Oculoauricular syndrome, 612109
HSF4	100.0	100%	100%	Cataract 5, multiple types, 116800
IFT140	83.3	99%	99%	Mainzer-Saldino syndrome, 266920
IFT172	97.6	100%	100%	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)
IFT172	97.6	100%	100%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	101.6	92%	92%	?Bardet-Biedl syndrome 19, 615996
IMPDH1	37.5	87%	87%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
IMPG2	125.8	99%	99%	Retinitis pigmentosa 56, 613581 Maculopathy, IMPG2-related, 613581
INPP5E	74.2	100%	100%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300

IQCB1	95.1	99%	99%	Senior-Loken syndrome 5, 609254
JAG1	110.2	99%	99%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 Deafness, congenital heart defects, and posterior embryotoxon
JAM3	71.1	94%	94%	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%	100%	Retinal cone dystrophy 3B, 610356
KIAA1549	113.3	96%	96%	No OMIM phenotype Autism (Neale (2012) Nature 485,242) Retinitis pigmentosa (Abu-Safieh (2013) Genome Res 23,236)
KIF11	98.6	99%	99%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF7	67.0	93%	93%	Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KIZ	109.0	100%	100%	Retinitis pigmentosa 69
KLHL7	118.9	100%	100%	Retinitis pigmentosa 42, 612943
LAMA1	97.2	99%	99%	Poretti-Boltshauser syndrome, 615960
LCA5	137.4	100%	100%	Leber congenital amaurosis 5, 604537
LIM2	62.6	78%	78%	Cataract 19, 615277
LRAT	196.5	100%	100%	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LRIT3	140.3	94%	94%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP5	89.4	98%	98%	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	89.4	100%	100%	Bardet-Biedl syndrome 17, 615994
MAB21L2	134.3	100%	100%	Microphthalmia, syndromic 14, 615877
MAF	79.8	81%	81%	Cataract, pulverulent or cerulean, with or without microcornea, 610202
MAK	85.9	96%	96%	Retinitis pigmentosa 62, 614181
MERTK	114.9	100%	100%	Retinitis pigmentosa 38, 613862

MFN2	103.5	100%	100%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VI, 601152
MFRP	87.8	100%	100%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	120.0	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MFSD8	120.0	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951
MIP	76.1	100%	100%	Cataract 15, multiple types, 615274
MKKS	148.8	100%	100%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 209900
MKS1	110.6	100%	100%	Meckel syndrome 1, 249000
MVK	88.3	100%	100%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
MVK	88.3	100%	100%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
MYO7A	77.0	95%	95%	Usher syndrome, type 1B, 276900 Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317
NAA10	51.9	97%	97%	?Microphthalmia, syndromic 1, 309800 N-terminal acetyltransferase deficiency, 300855
NDP	52.2	86%	86%	Norrie disease, 310600 Exudative vitreoretinopathy, X-linked, 305390
NEK2	27.6	47%	47%	?Retinitis pigmentosa 67, 615565
NHS	67.6	93%	93%	Nance-Horan syndrome, 302350
NMNAT1	96.0	100%	100%	Leber congenital amaurosis 9, 608553
NPHP1	169.1	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	110.4	100%	100%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	89.5	98%	98%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NR2E3	82.2	98%	98%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131

NR2F1	164.8	100%	100%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRL	47.6	100%	100%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type
NYX	47.7	97%	97%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	44.9	83%	83%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OFD1	37.0	88%	88%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OFD1	37.0	88%	88%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804
OPA1	131.0	99%	99%	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250
OPA3	108.0	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPN1LW	.4	%	%	Colorblindness, protan, 303900 Blue cone monochromacy, 303700
OPN1MW	.5	%	%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700
OTX2	158.2	100%	100%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, and pituitary dysfunction, 610125
PANK2	112.1	90%	90%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PAX2	104.7	97%	97%	Papillorenal syndrome, 120330 Renal hypoplasia, isolated, 191830
PAX6	83.7	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 Coloboma of optic nerve, 120430

				Gillespie syndrome, 206700
PCDH15	137.6	100%	100%	Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067
PCYT1A	90.8	100%	100%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE6A	100.7	99%	99%	Retinitis pigmentosa 43, 613810
PDE6B	100.3	100%	100%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	109.6	100%	100%	Cone dystrophy 4, 613093
PDE6D	110.2	100%	100%	?Joubert syndrome 22, 615665
PDE6G	99.1	100%	100%	Retinitis pigmentosa 57, 613582
PDE6H	40.0	87%	87%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDZD7	69.4	95%	95%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PET100	70.5	100%	100%	Mitochondrial complex IV deficiency, 220110
PITPNM3	77.4	99%	99%	Cone-rod dystrophy 5, 600977
PITX3	37.2	99%	99%	Anterior segment mesenchymal dysgenesis, 107250
PLA2G5	108.2	100%	100%	Fleck retina, familial benign, 228980
POC1B	101.9	99%	99%	Cone-rod dystrophy 20, 615973
PPT1	72.0	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	87.0	100%	100%	Retinitis pigmentosa 36, 610599
PROM1	86.1	99%	99%	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%	86%	Retinitis pigmentosa 11, 600138
PRPF4	114.4	100%	100%	Retinitis pigmentosa 70
PRPF6	88.8	100%	100%	Retinitis pigmentosa 60, 613983
PRPF8	120.7	99%	99%	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 Macular dystrophy Retinitis pigmentosa, digenic, 608133 Choriodal dystrophy, central areolar 2, 613105
PRSS56	60.5	97%	97%	Microphthalmia, isolated 6, 613517
RAB28	79.0	96%	96%	Cone-rod dystrophy 18, 615374
RARB	143.3	100%	100%	Microphthalmia, syndromic 12, 615524
RAX2	57.2	100%	100%	Cone-rod dystrophy 11, 610381 Macular degeneration, age-related, 6,613757
RBP3	103.7	100%	100%	?Retinitis pigmentosa 66, 615233
RBP4	80.1	95%	95%	Retinol dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RD3	56.9	100%	100%	Leber congenital amaurosis 12, 610612
RDH12	63.6	91%	91%	Leber congenital amaurosis 13, 612712
RDH5	107.3	100%	100%	Fundus albipunctatus, 136880
RGR	79.9	94%	94%	Retinitis pigmentosa 44, 613769
RGS9	104.9	98%	98%	Bradyopsia, 608415
RGS9BP	37.4	100%	100%	Bradyopsia, 608415
RHO	125.6	100%	100%	Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880
RIMS1	103.9	100%	100%	Cone-rod dystrophy 7, 603649
RLBP1	101.2	100%	100%	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%	100%	Retinitis pigmentosa 2, 312600
RP9	25.0	50%	50%	Retinitis pigmentosa 9, 180104
RPE65	114.7	100%	100%	Leber congenital amaurosis 2, 204100

RPGR	51.9	83%	83%	Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, sinorespiratory infections, with/without deafness, 300455 Macular degeneration, X-linked atrophic, 300834 Cone-rod dystrophy, X-linked, 1, 304020
RPGRIP1	118.5	100%	100%	Leber congenital amaurosis 6, 613826 Cone-rod dystrophy 13, 608194
RPGRIP1L	101.3	98%	98%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RS1	39.8	92%	92%	Retinoschisis, 312700
SAG	111.5	100%	100%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SDCCAG8	105.0	100%	100%	Senior-Loken syndrome 7, 613615
SEMA4A	100.0	99%	99%	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283 -3
SHH	95.9	99%	99%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SIX6	128.1	100%	100%	Microphthalmia with cataract 2, 212550
SLC24A1	142.7	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
SLC7A14	142.2	100%	100%	Retinitis pigmentosa 68, 615725
SNRNP200	112.4	100%	100%	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
STRA6	68.9	100%	100%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
TCTN1	108.9	100%	100%	Joubert syndrome 13, 614173
TCTN3	99.6	100%	100%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TDRD7	116.4	100%	100%	Cataract 36, 613887
TEAD1	88.6	100%	100%	Sveinsson choreoretinal atrophy, 108985
TENM3	131.3	100%	100%	Microphthalmia, isolated, with coloboma 9, 61545

TIMM8A	26.2	73%	73%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TIMP3	123.0	100%	100%	Sorsby fundus dystrophy, 136900
TMEM126A	85.8	100%	100%	Optic atrophy-7, 612989
TMEM138	97.1	100%	100%	Joubert syndrome 16, 614465
TMEM231	68.7	96%	96%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397
TMEM237	101.8	100%	100%	Joubert syndrome 14, 614424
TMEM67	119.5	100%	100%	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	135.3	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
TRPM1	132.4	100%	100%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TSPAN12	117.9	100%	100%	Exudative vitreoretinopathy 5, 613310
TTC8	111.8	100%	100%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
TTLL5	118.9	100%	100%	Cone-rod dystrophy 19,615860
TUB	83.7	100%	100%	No OMIM phenotype Retinal dystrophy and obesity (Borman (2014) Hum Mutat 35,289)
TULP1	84.8	100%	100%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
UNC119	103.5	100%	100%	?Cone-rod dystrophy ?Immunodeficiency 13, 615518
USH1C	79.4	99%	99%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904
USH1G	98.7	95%	95%	Usher syndrome, type 1G, 606943
USH2A	116.3	100%	100%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809

VAX1	64.1	100%	100%	Microphthalmia, syndromic 11, 614402
VCAN	147.5	100%	100%	Wagner syndrome 1, 143200
VIM	97.7	100%	100%	Cataract 30, pulverulent, 116300
VSX2	62.5	100%	100%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
WDPCP	93.8	100%	100%	?Bardet-Biedl syndrome 15, 615992
WDR19	123.9	100%	100%	Asphyxiating thoracic dystrophy 5, 614376 Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378
WDR19	123.9	100%	100%	Asphyxiating thoracic dystrophy 5, 614376 Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378
WFS1	145.6	100%	100%	?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}
YAP1	76.6	96%	96%	Coloboma, ocular, 120433 Coloboma, ocular, with/without hearing impairment, cleft lip/palate, and mental retardation
ZNF408	86.3	96%	96%	No OMIM ID Exudative vitreoretinopathy (Collin (2013) Proc Natl Acad Sci USA 110, 9856)
ZNF423	127.9	100%	100%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF513	99.1	100%	100%	Retinitis pigmentosa 58, 613617
ZNF644	169.0	100%	100%	Myopia 21, autosomal dominant, 614167

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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 : 31 october 2014
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors