VISION DISORDERS GENE PANEL DG 2.9 / DG 2.10 (375 genes)

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ABCA4	138.6	99%	99%	Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Stargardt disease 1, 248200 {Macular degeneration, age-related, 2}, 153800
ABCC6	121.4	93%	92%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABHD12	113.2	98%	90%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACBD5	171.2	98%	96%	No OMIM phenotype Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) ?Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
ACO2	141.2	97%	94%	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
ADAM9	168.8	99%	97%	Cone-rod dystrophy 9, 612775
ADAMTS18	168.8	99%	99%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADIPOR1	111.2	99%	98%	No OMIM phenotype syndromic retinitis pigmentosa (Xy (2016) Hum Mutat 37(3):246-249
AGBL1	145.6	100%	99%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	126.7	100%	99%	Retinitis pigmentosa 75,617023
AGK	152.2	99%	97%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AHI1	176.9	99%	98%	Joubert syndrome-3, 608629
AIPL1	133.9	100%	100%	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
ALMS1	208.7	99%	99%	Alstrom syndrome, 203800
AP3B1	128.1	99%	95%	Hermansky-Pudlak syndrome 2, 608233
APOPT1	90.9	87%	86%	Mitochondrial complex IV deficiency, 220110

ARHGEF18	128.1	99%	96%	Retinitis pigmentosa 78, 617433
ARL13B	117.5	99%	97%	Joubert syndrome 8, 612291
ARL2BP	82.9	90%	82%	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	101.4	99%	95%	No OMIM phenotype ?Retinitis pigmentosa (Strom (2016) PLoS One 11,e0150944)
ARL6	121.4	100%	97%	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ASPH	142.1	99%	96%	Traboulsi syndrome, 601552
ASRGL1	154.1	100%	99%	No OMIM phenotype Retinal degeneration (Biswas (2016) Hum Mol Genet 25,2483)
ATF6	161.2	100%	99%	Achromatopsia 7, 616517
B3GALTL	120.7	98%	95%	Peters-plus syndrome, 261540
BBIP1	170.9	99%	95%	?Bardet-Biedl syndrome 18, 615995
BBS1	178.6	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	195.4	100%	99%	Bardet-Biedl syndrome 10, 615987
BBS12	237	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	201.1	100%	99%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	153.5	99%	98%	Bardet-Biedl syndrome 4, 615982
BBS5	144	98%	96%	Bardet-Biedl syndrome 5, 615983
BBS7	156.1	99%	95%	Bardet-Biedl syndrome 7, 615984
BBS9	139.9	97%	95%	Bardet-Biedl syndrome 9, 615986
BCOR	117.5	99%	97%	Microphthalmia, syndromic 2, 300166
BEST1	159.3	99%	97%	Bestrophinopathy, autosomal recessive, 611809 Macular dystrophy, vitelliform, 2, 153700 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa, concentric, 613194 Retinitis pigmentosa-50, 613194 Vitreoretinochoroidopathy, 193220
BFSP1	119.9	99%	96%	Cataract 33, 611391
BFSP2	101.3	99%	97%	Cataract 12, multiple types, 611597
BLOC1S3	56	99%	95%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	111.3	99%	96%	Hermansky-pudlak syndrome 9, 614171

BMP4	164.2	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C10orf11	165.3	99%	99%	Albinism, oculocutaneous, type VII, 615179
C12orf65	94.5	98%	93%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	110.3	100%	99%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1QTNF5	175.6	91%	78%	Retinal degeneration, late-onset, autosomal dominant, 605670
C21orf2	112.5	99%	98%	No OMIM phenotype Retinal dystrophy,early-onset with macular staphyloma (Khan (2015) Br J Ophtalmol 99,1725) Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236) Jeune syndrome (Wheway (2015) Nat Cell Biol 17,1074)
C2orf71	137.3	99%	98%	Retinitis pigmentosa 54, 613428
C5orf42	154.1	99%	97%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C8orf37	149.6	100%	99%	Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
CA4	176.1	99%	99%	Retinitis pigmentosa 17, 600852
CABP4	116.6	99%	97%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1F	105.9	99%	97%	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA2D4	119.3	98%	97%	Retinal cone dystrophy 4, 610478
CAPN5	185.4	100%	99%	Vitreoretinopathy, neovascular inflammatory, 193235
CC2D2A	144.7	99%	97%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCDC41	126.7	99%	96%	Nephronophthisis 18, 615862
CDH23	225.8	99%	99%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067
CDH3	164.1	99%	98%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	168.7	99%	98%	Cone-rod dystrophy 15, 613660

				Retinitis pigmentosa 65, 613660
CEP164	100.3	99%	97%	Nephronophthisis 15, 614845
CEP250	102	99%	98%	No OMIM phenotype Usher syndrome,atypical (Khateb (2014) J Med Genet 51,460) ?Miscarriage, recurrent (Filges (2014) Mol Hum Reprod epub,epub)
CEP290	92.8	95%	87%	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	98.2	98%	93%	Joubert syndrome 15, 614464
CEP78	133.5	99%	96%	Cone-rod dystrophy and hearing loss, 617236
CERKL	121.8	99%	96%	Retinitis pigmentosa 26, 608380
CFH	216.6	99%	97%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CHM	123.8	97%	90%	Choroideremia, 303100
CHMP4B	158.5	99%	98%	Cataract 31, multiple types, 605387
CHST6	338.2	100%	100%	Macular corneal dystrophy, 217800
CIB2	235.2	100%	99%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLN3	133	99%	96%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	155.4	99%	97%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	144.8	98%	94%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	273.8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLRN1	171.9	100%	99%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CLUAP1	159.7	100%	99%	No OMIM phenotype Leber congenital amaurosis (Soens (2016) Genet Med 18,1044)
CNGA1	149.9	90%	85%	Retinitis pigmentosa 49, 613756
CNGA3	177.3	99%	99%	Achromatopsia-2, 216900

CNGB1	115.4	99%	97%	Retinitis pigmentosa 45, 613767
CNGB3	129.7	97%	92%	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200
CNNM4	212.9	98%	98%	Jalili syndrome, 217080
COL11A1	112.4	96%	92%	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	14.6	59%	23%	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, 215150 Stickler syndrome, type III, 184840 Weissenbacher-Zweymuller syndrome, 277610
COL18A1	94.8	96%	89%	Knobloch syndrome, type 1, 267750
COL2A1	119.3	99%	99%	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Otospondylomegaepiphyseal dysplasia, 215150 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia
COL8A2	42.4	90%	78%	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140
COL9A1	143.9	99%	97%	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135

COL9A2	77.1	99%	94%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284 {Intervertebral disc disease, susceptibility to}, 603932
CRB1	232	100%	100%	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, autosomal recessive, 600105
CRX	147.6	100%	99%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	158.8	97%	93%	Cataract 9, multiple types, 604219
CRYAB	120.6	99%	96%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869
CRYBA1	142.5	100%	99%	Cataract 10, multiple types, 600881
CRYBA2	164.3	100%	99%	?Cataract 42, 115900
CRYBA4	131.7	100%	100%	Cataract 23, 610425
CRYBB1	141.9	100%	99%	Cataract 17, multiple types, 611544
CRYBB2	175.3	100%	100%	Cataract 3, multiple types, 601547
CRYBB3	180.7	100%	99%	Cataract 22, autosomal recessive, 609741
CRYGB	106.7	100%	97%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	136.8	100%	99%	Cataract 2, multiple types, 604307
CRYGD	112	100%	99%	Cataract 4, multiple types, 115700
CRYGS	131.8	97%	91%	Cataract 20, multiple types, 116100
CSPP1	133.7	99%	98%	Joubert syndrome 21, 615636
CTDP1	120.5	91%	84%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA1	155.3	99%	99%	Macular dystrophy,patterned,608970 Gastric cancer, diffuse (Majewski (2012) J Pathol epub)
CTSD	197.2	99%	98%	Ceroid lipofuscinosis, neuronal, 10, 610127
CYP1B1	129.2	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP4V2	193.8	100%	99%	Bietti crystalline corneoretinal dystrophy, 210370
DCN	167.6	95%	95%	Corneal dystrophy, congenital stromal, 610048
DFNB31	121.4	99%	98%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383

DHDDS	103.9	96%	94%	Retinitis pigmentosa 59, 613861
DHX38	133.9	99%	97%	No OMIM phenotype Retinitis pigmentosa,early-onset with macular coloboma (Ajmal (2014) J Med Genet 51,444)
DKC1	122.5	100%	98%	Dyskeratosis congenita, X-linked, 305000
DRAM2	164.6	100%	100%	Cone-rod dystrophy 21, 616502
DTNBP1	127.3	99%	97%	Hermansky-Pudlak syndrome 7, 614076
EFEMP1	210.5	99%	99%	Doyne honeycomb degeneration of retina, 126600
ELOVL4	115.5	100%	99%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190
EPG5	138.8	99%	98%	Vici syndrome, 242840
EPHA2	193.2	99%	97%	Cataract 6, multiple types, 116600
EXOSC2	146.2	100%	100%	No OMIM phenotype Retinitis pigmentosa, hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt (Di Donato (2016) J Med Genet 53,419)
EYA1	167.2	100%	99%	Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 ?Otofaciocervical syndrome, 166780
EYS	172.3	99%	97%	Retinitis pigmentosa 25, 602772
FA2H	101.9	95%	89%	Spastic paraplegia 35, autosomal recessive, 612319
FAM161A	148.8	99%	98%	Retinitis pigmentosa 28, 606068
FLVCR1	166.7	99%	98%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOXC1	39.3	94%	79%	Axenfeld-Rieger syndrome, type 3, 602482 Iridogoniodysgenesis, type 1, 601631 Iris hypoplasia and glaucoma, 601631 Rieger or Axenfeld anomalies, 602482
FOXE3	24.7	73%	55%	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256 Cataract 34,multiple types, 612968
FRMD7	140.9	99%	99%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FTL	149.2	99%	93%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604

				Neurodegeneration with brain iron accumulation 3, 606159
FYCO1	141.9	100%	99%	Cataract 18, autosomal recessive, 610019
FZD4	226.6	99%	99%	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
GALK1	127.8	99%	96%	Galactokinase deficiency with cataracts, 230200
GALT	182.8	100%	100%	Galactosemia, 230400
GCNT2	201.6	100%	100%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GDF3	137.7	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF6	88.9	98%	92%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GFER	91.5	99%	92%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GJA1	238.2	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	172.5	99%	99%	Cataract 14, multiple types, 601885
GJA8	179.7	99%	99%	Cataract 1, multiple types, 116200
GNAT1	180.9	100%	100%	Night blindness, congenital stationary, autosomal dominant 3, 610444 ?Night blindness, congenital stationary, type 1G, 616389
GNAT2	157.8	100%	99%	Achromatopsia-4, 613856
GNB3	211.1	100%	99%	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNPTG	169.7	96%	92%	Mucolipidosis III gamma, 252605
GPR143	73.2	90%	83%	Nystagmus 6, congenital, X-linked, 300814

				Ocular albinism, type I, Nettleship-Falls type, 300500
GPR179	160.1	100%	99%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR98	172.4	99%	98%	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
GRK1	137.1	100%	99%	Oguchi disease-2, 613411
GRM6	166.2	94%	87%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	214.6	100%	100%	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GSN	141.9	97%	92%	Amyloidosis, Finnish type, 105120
GUCA1A	195.7	100%	100%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	154.9	100%	99%	Retinitis pigmentosa 48, 613827
GUCY2D	104.8	99%	94%	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
HARS	167	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HCCS	115.6	99%	98%	Linear skin defects with multiple congenital anomalies 1, 309801
HGSNAT	122.6	81%	81%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HK1	152.9	100%	99%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285
HMX1	25.4	72%	51%	Oculoauricular syndrome, 612109
HPS1	136.6	100%	99%	Hermansky-Pudlak syndrome 1, 203300
HPS4	151.7	100%	99%	Hermansky-Pudlak syndrome 4, 614073
HPS5	160.7	99%	98%	Hermansky-Pudlak syndrome 5, 614074
HPS6	156.4	97%	90%	Hermansky-Pudlak syndrome 6, 614075
HSF4	119.8	98%	95%	Cataract 5, multiple types, 116800
IDH3B	170.6	100%	100%	Retinitis pigmentosa 46, 612572
IFT140	124.5	99%	98%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	125.7	100%	99%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	133.2	100%	98%	?Bardet-Biedl syndrome 19, 615996

IFT74	100	99%	93%	?Bardet-Biedl syndrome 20, 617119
IMPDH1	59.3	93%	82%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	125.5	99%	98%	Macular dystrophy, vitelliform, 4, 616151
IMPG2	191.4	99%	97%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INPP5E	109	97%	92%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	166	100%	100%	Nephronophthisis 2, infantile, 602088
IQCB1	122	92%	81%	Senior-Loken syndrome 5, 609254
JAG1	160.7	99%	98%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
JAM3	158.3	99%	98%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KCNJ13	204.7	100%	99%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNV2	154.5	100%	99%	Retinal cone dystrophy 3B, 610356
KERA	208.7	100%	100%	Cornea plana congenita, recessive, 217300
KIF11	98	97%	95%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF7	95.3	95%	89%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
KIZ	174.5	99%	97%	Retinitis pigmentosa 69, 615780
KLHL7	154.6	99%	97%	Cold induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943
KRT12	130.8	98%	95%	Meesmann corneal dystrophy, 122100
KRT3	112.1	100%	99%	Meesmann corneal dystrophy, 122100
LAMA1	154	100%	99%	Poretti-Boltshauser syndrome, 615960
LCA5	178	98%	96%	Leber congenital amaurosis 5, 604537
LEMD2	80.2	95%	87%	Cataract 46, juvenile-onset, 212500
LEPREL1	117.6	99%	97%	Myopia, high, with cataract and vitreoretinal degeneration, 614292

LIM2	98.4	99%	98%	Cataract 19, multiple types, 615277
LRAT	345.6	100%	100%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRIT3	160.4	94%	93%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP5	210	98%	98%	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	137.3	100%	99%	Cataract 44, 616509
LYST	171	98%	96%	Chediak-Higashi syndrome, 214500
LZTFL1	142	99%	98%	Bardet-Biedl syndrome 17, 615994
MAB21L2	261.2	100%	100%	Microphthalmia, syndromic 14, 615877
MAF	67	79%	74%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAK	181.6	96%	94%	Retinitis pigmentosa 62, 614181
MAPKAPK3	91.1	99%	97%	?Macular dystrophy, patterned, 3, 617111
MERTK	182.5	100%	99%	Retinitis pigmentosa 38, 613862
MFN2	157.5	100%	100%	Charcot-Marie-Tooth disease, type 2A2A, 609260 Charcot-Marie-Tooth disease, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	140.1	100%	100%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	143.4	100%	99%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MIP	134.2	99%	95%	Cataract 15, multiple types, 615274
MIR184	NC	NC	NC	EDICT syndrome, 614303
MITF	173.3	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	216.9	89%	89%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	114.6	99%	99%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000
MVK	167.2	100%	99%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYO7A	149.2	99%	98%	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYOC	196.1	100%	99%	Glaucoma 1A, primary open angle, 137750
NAA10	109.4	99%	96%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NBAS	176.4	99%	98%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NDP	126.5	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NEK2	111.5	99%	97%	?Retinitis pigmentosa 67, 615565
NEUROD1	166.6	100%	100%	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853
NHS	138.9	96%	93%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NMNAT1	144.4	100%	99%	Leber congenital amaurosis 9, 608553
NPHP1	154.6	99%	98%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	146	99%	97%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	152.6	100%	99%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NR2E3	117.7	100%	100%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	216.5	99%	98%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRL	67.2	99%	96%	Retinal degeneration, autosomal recessive, clumped pigment type

				Retinitis pigmentosa 27, 613750
NYX	81.5	98%	95%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	96.2	80%	71%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCA2	141.6	99%	97%	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
OCRL	140.5	99%	98%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	59.2	87%	75%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OPA1	146.6	99%	97%	Behr syndrome,210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type),616896 {Glaucoma, normal tension, susceptibility to}, 606657
OPA3	125.8	99%	97%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPN1LW	85.7	68%	62%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	69.7	65%	58%	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800
OTX2	140.5	100%	99%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
PANK2	178.4	99%	98%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX2	200.3	100%	99%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX6	135.8	100%	100%	Aniridia, 106210 Cataract with late-onset corneal dystrophy, 106210 Coloboma of optic nerve, 120430 Coloboma, ocular, 120200

				Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550 Peters anomaly, 604229 ?Morning glory disc anomaly, 120430
PCDH15	179.5	99%	99%	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCYT1A	131.4	99%	96%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE6A	140.4	100%	99%	Retinitis pigmentosa 43, 613810
PDE6B	174.5	100%	100%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	148.4	98%	96%	Cone dystrophy 4, 613093
PDE6D	126.1	100%	99%	?Joubert syndrome 22, 615665
PDE6G	113.4	99%	97%	Retinitis pigmentosa 57, 613582
PDE6H	74.7	93%	65%	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024
PDZD7	113.2	99%	98%	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PET100	98.1	98%	86%	Mitochondrial complex IV deficiency, 220110
PEX1	139.9	98%	97%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX2	187.6	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX7	152.7	90%	87%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGK1	57.8	92%	80%	Phosphoglycerate kinase 1 deficiency, 300653
PHYH	90	98%	93%	Refsum disease, 266500
PIKFYVE	177.4	99%	98%	Corneal fleck dystrophy, 121850
PITX2	146.1	99%	98%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PITX3	64.5	99%	95%	Anterior segment mesenchymal dysgenesis, 107250

				Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623
PLA2G5	130.5	100%	100%	[Fleck retina, familial benign], 228980
PLK4	172.8	99%	97%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PNPLA6	150.4	99%	98%	Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020 ?Laurence-Moon syndrome, 245800
POC1B	99.5	97%	93%	Cone-rod dystrophy 20, 615973
POMGNT1	136.7	99%	97%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157
PPT1	203.5	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	103.1	100%	100%	Retinitis pigmentosa 36, 610599
PRDM13	128.2	95%	90%	No OMIM phenotype Macular dystrophy, North Carolina (Small (2016) Ophthalmology 123, 9)
PRDM5	163.3	99%	98%	Brittle cornea syndrome 2, 614170
PROM1	141	96%	93%	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PRPF3	92	98%	95%	Retinitis pigmentosa 18, 601414
PRPF31	130.5	98%	93%	Retinitis pigmentosa 11, 600138
PRPF4	160.8	99%	99%	Retinitis pigmentosa 70, 615922
PRPF6	130.5	100%	99%	Retinitis pigmentosa 60, 613983
PRPF8	143.2	99%	99%	Retinitis pigmentosa 13, 600059
PRPH2	249.9	100%	100%	Choriodal 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	59.9	98%	91%	Microphthalmia, isolated 6, 613517

PXDN	172.6	99%	99%	Corneal opacification and other ocular anomalies, 269400
RAB28	68.8	98%	93%	Cone-rod dystrophy 18, 615374
RARB	147.3	100%	100%	Microphthalmia, syndromic 12, 615524
RAX2	73.8	98%	91%	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RBP3	159.5	100%	99%	?Retinitis pigmentosa 66, 615233
RBP4	124.5	99%	95%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	135.4	100%	99%	[Beta-glycopyranoside tasting] {Alcohol dependence, susceptibility to}, 103780
RD3	182.9	100%	100%	Leber congenital amaurosis 12, 610612
RDH11	130.5	99%	99%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	102.7	99%	94%	Leber congenital amaurosis 13, 612712
RDH5	174.3	100%	99%	Fundus albipunctatus, 136880
REEP6	210.8	99%	96%	Retinitis pigmentosa 77, 617304
RGR	145.7	100%	99%	Retinitis pigmentosa 44, 613769
RGS9	117.1	100%	99%	Bradyopsia, 608415
RGS9BP	89.6	99%	99%	Bradyopsia, 608415
RHO	233	100%	100%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIMS1	140.2	98%	95%	Cone-rod dystrophy 7, 603649
RLBP1	143.9	100%	99%	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
ROM1	128.9	100%	99%	Retinitis pigmentosa 7, digenic, 608133
RP1	145.9	100%	99%	Retinitis pigmentosa 1, 180100
RP1L1	109.5	100%	99%	Occult macular dystrophy, 613587
RP2	189.5	100%	99%	Retinitis pigmentosa 2, 312600
RP9	77.9	78%	76%	?Retinitis pigmentosa 9, 180104
RPE65	165.8	100%	99%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	115.9	85%	78%	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	158.2	100%	99%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	160.8	96%	95%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RS1	65.5	99%	90%	Retinoschisis, 312700
RTN4IP1	109.9	99%	97%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
SAG	159.1	100%	100%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SDCCAG8	155	99%	98%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEMA4A	153.4	99%	99%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SHH	114.4	99%	95%	Holoprosencephaly-3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SIPA1L3	155.3	99%	98%	?Cataract 45, 616851
SIX6	251.7	100%	99%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SLC16A12	179.4	100%	100%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC24A1	222.7	100%	100%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	130.5	99%	97%	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A46	206.3	97%	90%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC33A1	152.6	98%	94%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC38A8	80.4	99%	95%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC45A2	152.5	99%	99%	Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 [Skin/hair/eye pigmentation 5, dark/fair skin], 227240

				[Skin/hair/eye pigmentation 5, dark/light eyes], 227240
SLC4A11	160.3	100%	99%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC52A2	206.8	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC7A14	198.4	100%	100%	Retinitis pigmentosa 68, 615725
SNRNP200	165.8	99%	99%	Retinitis pigmentosa 33, 610359
SOX2	127.1	99%	98%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SPATA7	149.2	98%	95%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPP2	169.8	100%	100%	No OMIM phenotype Retinitis pigmentosa (Li (2015) Sci Rep 5,14867) ?Autism (Neale (2012) Nature 485,242)
STRA6	120.5	100%	99%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
TACSTD2	232.7	99%	97%	Corneal dystrophy, gelatinous drop-like, 204870
TCTN1	116.8	96%	93%	Joubert syndrome 13, 614173
TCTN3	133.6	100%	99%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDRD7	176.2	99%	98%	Cataract 36, 613887
TEAD1	173.5	100%	99%	Sveinsson choreoretinal atrophy, 108985
TENM3	217.1	99%	99%	Microphthalmia, isolated, with coloboma 9, 615145
TGFBI	152.7	99%	94%	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-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082
TIMM8A	43.7	88%	71%	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700
TIMP3	169.1	100%	100%	Sorsby fundus dystrophy, 136900
TMEM126A	112.9	98%	88%	Optic atrophy 7, 612989

TMEM138	128	100%	99%	Joubert syndrome 16, 614465
TMEM231	105.5	99%	98%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	126	99%	98%	Joubert syndrome 14, 614424
TMEM67	93.3	95%	89%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TOPORS	261.3	100%	100%	Retinitis pigmentosa 31, 609923
TPP1	155.4	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAF3IP1	97.5	96%	93%	Senior-Loken syndrome 9, 616629
TREX1	302.9	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	147.3	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRNT1	123.4	98%	94%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPM1	185.9	100%	99%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TSPAN12	163.4	100%	99%	Exudative vitreoretinopathy 5, 613310
TTC8	118.3	99%	98%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTLL5	169.1	99%	97%	Cone-rod dystrophy 19, 615860
TUB	112.8	98%	96%	?Retinal dystrophy and obesity, 616188
TUBGCP4	149	99%	96%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TULP1	116.2	98%	95%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TYR	205.9	100%	100%	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800

				[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800
TYRP1	200.1	100%	100%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBIAD1	263.9	99%	97%	Corneal dystrophy, Schnyder type, 121800
UNC119	103	99%	93%	?Cone-rod dystrophy ?Immunodeficiency 13, 615518
UNC45B	140.1	99%	99%	?Cataract 43, 616279
USH1C	119.4	100%	99%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	185.8	98%	96%	Usher syndrome, type 1G, 606943
USH2A	178.6	100%	99%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
VAX1	68.4	90%	83%	?Microphthalmia, syndromic 11, 614402
VCAN	213.9	100%	100%	Wagner syndrome 1, 143200
VIM	136.8	99%	96%	?Cataract 30, pulverulent, 116300
VSX1	67.7	92%	79%	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
VSX2	85.2	99%	96%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
WDPCP	137.3	95%	91%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	170	100%	99%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WFS1	260.1	99%	99%	Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome, 222300 Wolfram-like syndrome, autosomal dominant, 614296 ?Cataract 41, 116400 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WRN	149.6	99%	96%	Werner syndrome, 277700
YAP1	112.2	90%	84%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433

YME1L1	125	98%	95%	?Optic atrophy 11, 617302
ZEB1	197.2	100%	99%	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZNF408	151.6	100%	100%	Retinitis pigmentosa 72, 616469 ?Exudative vitreoretinopathy 6, 616468
ZNF423	268.6	100%	100%	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF513	122.2	100%	99%	Retinitis pigmentosa 58, 613617
ZNF644	213.7	100%	99%	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 14th 2017
This list is accurate for panel versions DG 2.9 and DG 2.10

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