VISION DISORDERS GENE PANEL DG 2.7/DG 2.8

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ABCA4	112.8	100%	98%	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	94.8	93%	91%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABHD12	93.7	89%	78%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACBD5	150.6	96%	95%	No OMIM phenotype Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) ?Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
ADAM9	149.9	98%	93%	Cone-rod dystrophy 9, 612775
ADAMTS18	147.5	98%	97%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADIPOR1	88.4	100%	94%	No OMIM phenotype syndromic retinitis pigmentosa (Xy (2016) Hum Mutat 37(3):246-249
AGBL1	126.5	100%	100%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	98.5	100%	99%	Retinitis pigmentosa 75,617023
AGK	115.1	99%	94%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AHI1	133	99%	93%	Joubert syndrome-3, 608629
AIPL1	111.6	100%	100%	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
ALMS1	167.8	99%	99%	Alstrom syndrome, 203800
AP3B1	93.9	99%	92%	Hermansky-Pudlak syndrome 2, 608233
APOPT1	62.9	87%	82%	Mitochondrial complex IV deficiency, 220110
ARL13B	78.8	99%	81%	Joubert syndrome 8, 612291

ARL2BP	72.8	78%	71%	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	84.6	93%	86%	No OMIM phenotype ?Retinitis pigmentosa (Strom (2016) PLoS One 11)
ARL6	94.9	99%	96%	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ASPH	110.5	99%	94%	Traboulsi syndrome, 601552
ATF6	134.8	100%	98%	Achromatopsia 7, 616517
B3GALTL	100.3	95%	94%	Peters-plus syndrome, 261540
BBIP1	121	99%	87%	?Bardet-Biedl syndrome 18, 615995
BBS1	133.8	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	160.2	100%	99%	Bardet-Biedl syndrome 10, 615987
BBS12	196.3	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	173	100%	98%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	136.1	99%	95%	Bardet-Biedl syndrome 4, 615982
BBS5	103.3	97%	91%	Bardet-Biedl syndrome 5, 615983
BBS7	121.3	96%	90%	Bardet-Biedl syndrome 7, 615984
BBS9	105.1	94%	93%	Bardet-Biedl syndrome 9, 615986
BCOR	71	98%	95%	Microphthalmia, syndromic 2, 300166
BEST1	111.7	99%	96%	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	90.6	94%	89%	Cataract 33, 611391
BFSP2	80.3	98%	94%	Cataract 12, multiple types, 611597
BLOC1S3	38.5	99%	84%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	89.9	99%	84%	Hermansky-pudlak syndrome 9, 614171
BMP4	120.8	100%	98%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C10orf11	135.8	99%	99%	Albinism, oculocutaneous, type VII, 615179

C12orf65	74.3	99%	97%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	78.4	100%	96%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1QTNF5	141.5	78%	60%	Retinal degeneration, late-onset, autosomal dominant, 605670
C21orf2	81.5	99%	96%	No OMIM disease Retinal dystrophy,early-onset with macular staphyloma (Khan (2015) Br J Ophtalmol 99,1725) Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236) Jeune syndrome (Wheway (2015) Nat Cell Biol 17,1074)
C2orf71	116.9	100%	99%	Retinitis pigmentosa 54, 613428
C5orf42	119.9	95%	91%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C8orf37	116.7	100%	99%	Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
CA4	133.6	100%	97%	Retinitis pigmentosa 17, 600852
CABP4	90.2	96%	92%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1F	62.7	98%	91%	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA2D4	100.4	98%	97%	Retinal cone dystrophy 4, 610478
CAPN5	142.6	100%	99%	Vitreoretinopathy, neovascular inflammatory, 193235
CC2D2A	121.8	99%	94%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCDC41	103.8	99%	96%	Nephronophthisis 18, 615862
CDH23	183	100%	99%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067
CDH3	132.2	100%	97%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	141.1	100%	99%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CEP164	75.2	99%	94%	Nephronophthisis 15, 614845

CEP250	84.5	100%	97%	No OMIM phenotype Usher syndrome,atypical (Khateb (2014) J Med Genet 51,460) ?Miscarriage, recurrent (Filges (2014) Mol Hum Reprod epub,epub)
CEP290	69.5	89%	76%	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	83.4	97%	91%	Joubert syndrome 15, 614464
CEP78	119.3	98%	94%	No OMIM phenotype Usher syndrome (Fu et al. (2016) J Med Genet)
CERKL	108.2	99%	95%	Retinitis pigmentosa 26, 608380
CFH	176.8	98%	95%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CHM	66.7	90%	77%	Choroideremia, 303100
CHMP4B	125.3	99%	96%	Cataract 31, multiple types, 605387
CHST6	280	100%	100%	Macular corneal dystrophy, 217800
CIB2	188.6	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLN3	107.2	99%	97%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	144.8	97%	91%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	119.2	100%	92%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	206.6	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLRN1	141.9	100%	100%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CLUAP1	133.1	100%	100%	No OMIM phenotype Leber congenital amaurosis (Soens et al. (2016) Genet. Med.)
CNGA1	128.2	84%	84%	Retinitis pigmentosa 49, 613756
CNGA3	149.8	99%	98%	Achromatopsia-2, 216900
CNGB1	91.5	95%	92%	Retinitis pigmentosa 45, 613767

CNGB3	104.7	91%	86%	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200
CNNM4	174.3	98%	97%	Jalili syndrome, 217080
COL11A1	85.5	92%	83%	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	10.9	46%	9%	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	66.8	90%	83%	Knobloch syndrome, type 1, 267750
COL2A1	93.5	99%	96%	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	21.8	76%	53%	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140
COL9A1	107.6	99%	95%	Stickler syndrome, type IV, 614134 /?Epiphyseal dysplasia, multiple, 6, 614135

COL9A2	55.5	98%	86%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284 {Intervertebral disc disease, susceptibility to}, 603932
CRB1	199.9	100%	99%	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, autosomal recessive, 600105
CRX	91.4	100%	98%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	121.6	84%	82%	Cataract 9, multiple types, 604219
CRYAB	120.2	97%	95%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869
CRYBA1	130.8	100%	100%	Cataract 10, multiple types, 600881
CRYBA2	126	100%	100%	?Cataract 42, 115900
CRYBA4	98.4	100%	97%	Cataract 23, 610425
CRYBB1	108.3	100%	99%	Cataract 17, multiple types, 611544
CRYBB2	152	100%	100%	Cataract 3, multiple types, 601547
CRYBB3	134.1	100%	98%	Cataract 22, autosomal recessive, 609741
CRYGB	95	100%	95%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	114.1	100%	100%	Cataract 2, multiple types, 604307
CRYGD	89	100%	100%	Cataract 4, multiple types, 115700
CRYGS	130.9	95%	91%	Cataract 20, multiple types, 116100
CSPP1	99.3	99%	92%	Joubert syndrome 21, 615636
CTDP1	93.2	90%	85%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA1	127.1	100%	98%	Macular dystrophy,patterned,608970 Gastric cancer, diffuse (Majewski (2012) J Pathol epub)
CTSD	152.8	98%	94%	Ceroid lipofuscinosis, neuronal, 10, 610127
CYP1B1	106.3	100%	99%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP4V2	157	100%	98%	Bietti crystalline corneoretinal dystrophy, 210370
DCN	141.1	95%	94%	Corneal dystrophy, congenital stromal, 610048
DFNB31	102.8	100%	97%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383

DHDDS	84.7	95%	93%	Retinitis pigmentosa 59, 613861
DHX38	122.8	100%	98%	No OMIM phenotype Retinitis pigmentosa,early-onset with macular coloboma (Ajmal (2014) J Med Genet 51,444)
DKC1	77.4	99%	92%	Dyskeratosis congenita, X-linked, 305000
DRAM2	144.9	100%	100%	Cone-rod dystrophy 21, 616502
DTNBP1	100.5	100%	95%	Hermansky-Pudlak syndrome 7, 614076
EFEMP1	176.8	100%	98%	Doyne honeycomb degeneration of retina, 126600
ELOVL4	77.5	100%	98%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190
EPG5	123.6	99%	94%	Vici syndrome, 242840
EPHA2	170.6	97%	97%	Cataract 6, multiple types, 116600
EXOSC2	135.8	100%	100%	No OMIM phenotype Syndromic retinitis pigmentosa (Di Donato (2016) J Med Genet 53,419)
EYA1	139.1	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	143.4	98%	94%	Retinitis pigmentosa 25, 602772
FA2H	87.6	93%	81%	Spastic paraplegia 35, autosomal recessive, 612319
FAM161A	110.4	99%	93%	Retinitis pigmentosa 28, 606068
FLVCR1	121.4	99%	97%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOXC1	31.1	95%	72%	Axenfeld-Rieger syndrome, type 3, 602482 Iridogoniodysgenesis, type 1, 601631 Iris hypoplasia and glaucoma, 601631 Rieger or Axenfeld anomalies, 602482
FOXE3	9.3	49%	33%	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256
FRMD7	79.4	99%	93%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FTL	104.5	99%	82%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FYCO1	114.3	100%	99%	Cataract 18, autosomal recessive, 610019

FZD4	207.1	100%	98%	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
GALK1	88.7	96%	91%	Galactokinase deficiency with cataracts, 230200
GALT	141.5	100%	99%	Galactosemia, 230400
GCNT2	164.9	100%	100%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GDF3	119.6	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF6	56.8	93%	82%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GFER	69.7	98%	71%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GJA1	205.3	100%	100%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva, 133200 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJA3	152.3	100%	100%	Cataract 14, multiple types, 601885
GJA8	136.5	100%	100%	Cataract 1, multiple types, 116200
GNAT1	153.1	100%	100%	Night blindness, congenital stationary, autosomal dominant 3, 610444 ?Night blindness, congenital stationary, type 1G, 616389
GNAT2	142.9	100%	98%	Achromatopsia-4, 613856
GNB3	193.2	100%	100%	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNPTG	125.2	90%	87%	Mucolipidosis III gamma, 252605
GPR143	39.9	90%	67%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR179	119.3	100%	97%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565

GPR98	145.8	99%	94%	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
GRK1	100.1	100%	100%	Oguchi disease-2, 613411
GRM6	139.7	93%	83%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	171.5	100%	100%	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GSN	109	91%	87%	Amyloidosis, Finnish type, 105120
GUCA1A	147.4	100%	100%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	130.1	100%	98%	Retinitis pigmentosa 48, 613827
GUCY2D	79.9	96%	84%	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
HARS	143.6	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HCCS	63.8	99%	92%	Linear skin defects with multiple congenital anomalies 1, 309801
HGSNAT	95	81%	81%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HK1	134.8	100%	99%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa (Wang (2014) Invest Ophthalmol Vis Sci 55,7159)
HMX1	19.6	63%	39%	Oculoauricular syndrome, 612109
HPS1	102.9	100%	98%	Hermansky-Pudlak syndrome 1, 203300
HPS4	130.4	100%	98%	Hermansky-Pudlak syndrome 4, 614073
HPS5	137.7	99%	98%	Hermansky-Pudlak syndrome 5, 614074
HPS6	100.4	100%	92%	Hermansky-Pudlak syndrome 6, 614075
HSF4	89.4	96%	87%	Cataract 5, multiple types, 116800
IDH3B	166.6	100%	100%	Retinitis pigmentosa 46, 612572
IFT140	105.2	99%	96%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	108.6	99%	97%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	106.4	100%	99%	?Bardet-Biedl syndrome 19, 615996
IMPDH1	47.6	91%	74%	Leber congenital amaurosis 11, 613837

				Retinitis pigmentosa 10, 180105
IMPG1	98.9	100%	95%	Macular dystrophy, vitelliform, 4, 616151
IMPG2	166.8	100%	97%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INPP5E	83.9	96%	88%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	153.4	100%	99%	Nephronophthisis 2, infantile, 602088
IQCB1	96.9	78%	74%	Senior-Loken syndrome 5, 609254
JAG1	139.8	100%	97%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
JAM3	136.4	100%	99%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KCNJ13	194	100%	98%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNV2	120	100%	100%	Retinal cone dystrophy 3B, 610356
KERA	185.9	100%	100%	Cornea plana congenita, recessive, 217300
KIF11	76.6	97%	95%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF7	70	91%	84%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
KIZ	150.4	98%	94%	Retinitis pigmentosa 69, 615780
KLHL7	139.1	99%	98%	Cold induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943
KRT12	88	95%	92%	Meesmann corneal dystrophy, 122100
KRT3	90.9	100%	96%	Meesmann corneal dystrophy, 122100
LAMA1	134.2	99%	98%	Poretti-Boltshauser syndrome, 615960
LCA5	130	95%	94%	Leber congenital amaurosis 5, 604537
LEMD2	60.2	99%	75%	Cataract 46, juvenile-onset, 212500
LEPREL1	87.8	93%	87%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
LIM2	88.4	100%	99%	Cataract 19, multiple types, 615277
LRAT	285.3	100%	100%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341

LRIT3	148.4	94%	93%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP5	165.5	98%	96%	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LSS	108.7	100%	98%	Cataract 44, 616509
LYST	129.6	97%	92%	Chediak-Higashi syndrome, 214500
LZTFL1	121.2	98%	90%	Bardet-Biedl syndrome 17, 615994
MAB21L2	210	100%	100%	Microphthalmia, syndromic 14, 615877
MAF	51.8	75%	72%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAK	146.6	94%	93%	Retinitis pigmentosa 62, 614181
MAPKAPK3	76.9	98%	95%	No OMIM phenotype Martinique crinkled retinal pigment epitheliopathy (Meunier (2016) Hum Mol Gene 25,916)
MERTK	162.2	100%	99%	Retinitis pigmentosa 38, 613862
MFN2	138	100%	100%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	117.5	100%	100%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	120.9	100%	98%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MIP	104.3	94%	87%	Cataract 15, multiple types, 615274
MIR184	NC	NC	NC	EDICT syndrome, 614303
MITF	134.3	100%	100%	Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MKKS	222.1	89%	89%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	95.9	99%	96%	Bardet-Biedl syndrome 13, 615990

				Meckel syndrome 1, 249000
MVK	126.3	100%	100%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYO7A	127	98%	95%	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYOC	171.9	100%	100%	Glaucoma 1A, primary open angle, 137750
NAA10	58	100%	91%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NBAS	134.1	99%	97%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NDP	80.3	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NEK2	81.6	95%	86%	?Retinitis pigmentosa 67, 615565
NEUROD1	153.6	100%	100%	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853 Retinitis pigmentosa, autosomal recessive (Wang (2015) Invest Ophthalmol Vis Sci 56,150)
NHS	83.7	93%	88%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NMNAT1	128.7	100%	99%	Leber congenital amaurosis 9, 608553
NPHP1	122	99%	94%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	111.4	98%	91%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	127.5	99%	97%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NR2E3	95.9	100%	100%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	182.2	99%	92%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRL	61.1	98%	86%	Retinal degeneration, autosomal recessive, clumped pigment type Retinitis pigmentosa 27, 613750

NYX	50.7	96%	95%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	73.6	68%	61%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCA2	126.1	97%	95%	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
OFD1	29.6	74%	53%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OPA1	115.4	98%	89%	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	89.2	99%	93%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPN1LW	48.9	66%	44%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	64.3	65%	50%	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800
OTX2	116.2	100%	97%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
PANK2	142.9	95%	86%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX2	148.6	100%	99%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330

PAX6	129.3	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	164.2	99%	98%	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCYT1A	123.1	97%	94%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE6A	120.3	100%	99%	Retinitis pigmentosa 43, 613810
PDE6B	136.8	100%	99%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	131.5	96%	94%	Cone dystrophy 4, 613093
PDE6D	93.9	100%	100%	?Joubert syndrome 22, 615665
PDE6G	85.3	99%	94%	Retinitis pigmentosa 57, 613582
PDE6H	58.1	51%	48%	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024
PDZD7	77.7	100%	89%	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PET100	110.5	89%	72%	Mitochondrial complex IV deficiency, 220110
PEX1	106.9	97%	97%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX2	147.6	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX7	121.4	91%	82%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGK1	35.1	80%	65%	Phosphoglycerate kinase 1 deficiency, 300653
PHYH	87.5	97%	92%	Refsum disease, 266500
PIKFYVE	137.7	99%	98%	Corneal fleck dystrophy, 121850

PITX2	128.1	96%	93%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PITX3	56	100%	92%	Anterior segment mesenchymal dysgenesis, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623
PLA2G5	117.5	100%	100%	[Fleck retina, familial benign], 228980
PLK4	135.2	99%	86%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PNPLA6	111.2	99%	97%	Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020 ?Laurence-Moon syndrome, 245800
POC1B	93.5	92%	90%	Cone-rod dystrophy 20, 615973
POMGNT1	107	100%	95%	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	150.6	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	89.1	100%	100%	Retinitis pigmentosa 36, 610599
PROM1	112.9	96%	93%	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PRPF3	73.3	98%	91%	Retinitis pigmentosa 18, 601414
PRPF31	99.1	100%	93%	Retinitis pigmentosa 11, 600138
PRPF4	137.2	99%	98%	Retinitis pigmentosa 70, 615922
PRPF6	114.1	100%	99%	Retinitis pigmentosa 60, 613983
PRPF8	132.4	100%	99%	Retinitis pigmentosa 13, 600059
PRPH2	209.9	100%	98%	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	38.9	93%	73%	Microphthalmia, isolated 6, 613517
PXDN	153.8	99%	97%	Corneal opacification and other ocular anomalies, 269400
RAB28	50.6	93%	77%	Cone-rod dystrophy 18, 615374
RARB	124.5	100%	100%	Microphthalmia, syndromic 12, 615524
RAX2	45.5	90%	69%	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RBP3	128	100%	99%	?Retinitis pigmentosa 66, 615233
RBP4	87	93%	91%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	116.9	99%	98%	[Beta-glycopyranoside tasting] {Alcohol dependence, susceptibility to}, 103780
RD3	136.9	100%	100%	Leber congenital amaurosis 12, 610612
RDH11	125.6	100%	97%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	87.6	97%	83%	Leber congenital amaurosis 13, 612712
RDH5	144	100%	100%	Fundus albipunctatus, 136880
RGR	123.5	100%	100%	Retinitis pigmentosa 44, 613769
RGS9	94.3	100%	99%	Bradyopsia, 608415
RGS9BP	59	100%	97%	Bradyopsia, 608415
RHO	224.6	100%	100%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIMS1	113.5	96%	93%	Cone-rod dystrophy 7, 603649
RLBP1	127.9	100%	100%	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
ROM1	97.7	100%	98%	Retinitis pigmentosa 7, digenic, 608133
RP1	123.7	99%	96%	Retinitis pigmentosa 1, 180100
RP1L1	81.7	100%	98%	Occult macular dystrophy, 613587
RP2	117.2	100%	98%	Retinitis pigmentosa 2, 312600
RP9	46	77%	77%	?Retinitis pigmentosa 9, 180104
RPE65	138.6	100%	99%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794

RPGR	59.2	81%	66%	Cone-rod dystrophy, X-linked, 1, 304020 Macular degeneration, X-linked atrophic, 300834 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455
RPGRIP1	145.2	100%	99%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	133.2	95%	93%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RS1	32.2	83%	72%	Retinoschisis, 312700
RTN4IP1	94.8	100%	97%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
SAG	126.2	100%	100%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SDCCAG8	116.7	100%	96%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEMA4A	118.9	100%	97%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SHH	84.8	95%	91%	Holoprosencephaly-3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SIPA1L3	121.9	98%	96%	?Cataract 45, 616851
SIX6	180.4	100%	100%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SLC16A12	167.6	100%	100%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC24A1	199.4	100%	99%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	119.9	95%	93%	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A46	175.2	96%	94%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC33A1	133.8	98%	89%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC38A8	74	100%	93%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218

SLC45A2	128.1	100%	98%	Albinism, oculocutaneous, type IV, 606574 [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	133.3	100%	99%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy 2, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC52A2	168	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC7A14	179.5	100%	100%	Retinitis pigmentosa 68, 615725
SNRNP200	144.9	100%	98%	Retinitis pigmentosa 33, 610359
SOX2	80.8	99%	96%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SPATA7	128	96%	86%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPP2	155.5	100%	100%	No OMIM phenotype Retinitis pigmentosa (Li (2015) Sci Rep 5,14867) ?Autism (Neale (2012) Nature 485,242)
STRA6	105.6	100%	100%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
TACSTD2	176.2	99%	97%	Corneal dystrophy, gelatinous drop-like, 204870
TCTN1	98.8	94%	92%	Joubert syndrome 13, 614173
TCTN3	116.3	99%	98%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDRD7	170.6	98%	97%	Cataract 36, 613887
TEAD1	161.4	100%	96%	Sveinsson choreoretinal atrophy, 108985
TENM3	185.5	99%	98%	Microphthalmia, isolated, with coloboma 9, 615145
TGFBI	121.8	100%	92%	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082
TIMM8A	17.5	64%	30%	Jensen syndrome, 311150

				Mohr-Tranebjaerg syndrome, 304700
TIMP3	160.3	100%	100%	Sorsby fundus dystrophy, 136900
TMEM126A	108.6	99%	91%	Optic atrophy 7, 612989
TMEM138	102.7	99%	97%	Joubert syndrome 16, 614465
TMEM231	86.1	99%	95%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	96.1	99%	97%	Joubert syndrome 14, 614424
TMEM67	70.3	91%	84%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TOPORS	234.8	100%	100%	Retinitis pigmentosa 31, 609923
TPP1	124	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAF3IP1	71.5	92%	90%	Senior-Loken syndrome 9, 616629
TREX1	214.3	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRIM32	134.2	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRNT1	93.1	95%	91%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPM1	161.4	100%	98%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TSPAN12	135.9	100%	98%	Exudative vitreoretinopathy 5, 613310
TTC8	88.4	98%	88%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTLL5	149.6	100%	96%	Cone-rod dystrophy 19, 615860
TUB	91.6	99%	94%	?Retinal dystrophy and obesity, 616188
TUBGCP4	135.5	98%	95%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TULP1	88.6	98%	93%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132

TYR	189.7	100%	100%	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [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	190.1	100%	100%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBIAD1	225.9	98%	93%	Corneal dystrophy, Schnyder type, 121800
UNC119	91.1	98%	72%	?Cone-rod dystrophy ?Immunodeficiency 13, 615518
UNC45B	116.5	100%	98%	?Cataract 43, 616279
USH1C	99.1	98%	95%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	153.6	95%	93%	Usher syndrome, type 1G, 606943
USH2A	156.8	99%	98%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
VAX1	44.7	87%	70%	?Microphthalmia, syndromic 11, 614402
VCAN	185.9	100%	100%	Wagner syndrome 1, 143200
VIM	117	100%	93%	?Cataract 30, pulverulent, 116300
VSX1	54.9	91%	73%	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
VSX2	67.6	98%	88%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
WDPCP	107.1	94%	93%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	133.5	100%	98%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376

WFS1	218.6	98%	97%	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	128.3	97%	94%	Werner syndrome, 277700
YAP1	95.5	85%	79%	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
ZEB1	188	100%	99%	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZNF408	112	100%	100%	Retinitis pigmentosa 72, 616469 ?Exudative vitreoretinopathy 6, 616468
ZNF423	226.6	100%	100%	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF513	90.9	100%	97%	Retinitis pigmentosa 58, 613617
ZNF644	163.1	100%	100%	Myopia 21, autosomal dominant, 614167

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

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