VISION DISORDERS GENE PANEL DG 2.16 (432 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ABCA4	104,5	99.9%	98.9%	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	109,1	93.6%	92.8%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABHD12	93,1	100.0%	98.9%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACBD5	145,1	99.6%	98.0%	No OMIM phenotype Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) ?Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
ACO2	115,3	95.8%	89.5%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ADAM9	141,3	99.9%	99.0%	Cone-rod dystrophy 9, 612775
ADAMTS18	129,7	100.0%	99.9%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTSL4	122,7	100.0%	99.6%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADGRV1	126	99.7%	98.4%	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
ADIPOR1	87,8	99.4%	95.2%	No OMIM phenotype syndromic retinitis pigmentosa (Xy (2016) Hum Mutat 37(3):246-249
AGBL1	106,4	98.5%	98.4%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	104,9	100.0%	99.4%	Retinitis pigmentosa 75, 617023
AGK	108,5	99.5%	95.7%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AHI1	129,8	99.7%	98.3%	Joubert syndrome 3, 608629

AHR	184	100.0%	99.6%	?Retinitis pigmentosa 85, 618345
AIPL1	113	100.0%	99.9%	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
ALDH1A3	102,4	99.7%	97.0%	Microphthalmia, isolated 8, 615113
ALMS1	172,8	100.0%	99.7%	Alstrom syndrome, 203800
AP3B1	112,1	99.5%	96.5%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	125,2	98.4%	97.9%	?Hermansky-Pudlak syndrome 10, 617050
APOPT1	NC	NC	NC	Mitochondrial complex IV deficiency, 220110
ARHGEF18	140,4	99.5%	97.3%	Retinitis pigmentosa 78, 617433
ARL13B	102,2	100.0%	99.4%	Joubert syndrome 8, 612291
ARL2BP	63,8	92.4%	83.1%	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	73,5	99.8%	95.7%	Joubert syndrome 35, 618161 Retinitis pigmentosa 83, 618173
ARL6	100,3	99.8%	98.2%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARR3	83,7	99.9%	99.2%	Myopia 26, X-linked, female-limited, 301010
ARSG	113,1	99.9%	98.6%	Usher syndrome, type IV, 618144
ASPH	111,3	99.9%	98.8%	Traboulsi syndrome, 601552
ASRGL1	122,3	100.0%	100.0%	No OMIM phenotype Retinal degeneration (Biswas (2016) Hum Mol Genet 25,2483)
ATF6	125,2	100.0%	99.3%	Achromatopsia 7, 616517
ATOH7	176,2	98.6%	97.0%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
B3GLCT	96,6	99.7%	99.1%	Peters-plus syndrome, 261540
BBIP1	119,7	97.3%	90.3%	?Bardet-Biedl syndrome 18, 615995
BBS1	146,4	100.0%	100.0%	Bardet-Biedl syndrome 1, 209900
BBS10	158,1	100.0%	99.9%	Bardet-Biedl syndrome 10, 615987
BBS12	187,1	100.0%	100.0%	Bardet-Biedl syndrome 12, 615989
BBS2	150,7	99.9%	99.6%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	110,2	99.9%	99.2%	Bardet-Biedl syndrome 4, 615982
BBS5	98,5	98.0%	93.3%	Bardet-Biedl syndrome 5, 615983
BBS7	142,9	99.1%	96.5%	Bardet-Biedl syndrome 7, 615984
BBS9	113,6	98.6%	94.4%	Bardet-Biedl syndrome 9, 615986

BCOR	102,7	98.8%	95.3%	Microphthalmia, syndromic 2, 300166
BEST1	127,2	99.6%	97.1%	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	100,1	100.0%	99.1%	Cataract 33, multiple types, 611391
BFSP2	99,3	99.9%	98.2%	Cataract 12, multiple types, 611597
BLOC1S3	67,4	100.0%	99.9%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	101,1	99.2%	95.1%	?Hermansky-pudlak syndrome 9, 614171
BMP4	173,4	100.0%	100.0%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C12orf65	110,4	100.0%	99.6%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	104,2	100.0%	99.8%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
C1QTNF5	154,2	97.2%	90.6%	Retinal degeneration, late-onset, autosomal dominant, 605670
C21orf2	NC	NC	NC	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C2orf71	NC	NC	NC	Retinitis pigmentosa 54, 613428
C5orf42	NC	NC	NC	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C8orf37	144,1	99.8%	99.4%	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
CA4	162,1	100.0%	100.0%	Retinitis pigmentosa 17, 600852
CABP4	148,3	100.0%	100.0%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1F	84,9	99.8%	97.1%	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA2D4	95,9	99.0%	97.1%	Retinal cone dystrophy 4, 610478
CAPN5	153,6	100.0%	99.8%	Vitreoretinopathy, neovascular inflammatory, 193235
CC2D2A	111,7	99.0%	97.1%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284

CCT2	145,8	100.0%	99.8%	No OMIM phenotype
CDH23	172,7	100.0%	100.0%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 {Pituitary adenoma 5, multiple types}, 617540
CDH3	140,5	100.0%	99.8%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	143,6	99.9%	99.0%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK10	131,3	100.0%	100.0%	Al Kaissi syndrome, 617694
CEP164	89,3	99.8%	98.0%	Nephronophthisis 15, 614845
CEP250	99,8	99.9%	98.8%	Cone-rod dystrophy and hearing loss 2, 618358
CEP290	82,6	97.3%	91.7%	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	78,2	98.9%	94.4%	Joubert syndrome 15, 614464
CEP78	120,1	99.7%	97.6%	Cone-rod dystrophy and hearing loss, 617236
CEP83	108,8	99.4%	96.6%	Nephronophthisis 18, 615862
CERKL	114,1	99.4%	97.2%	Retinitis pigmentosa 26, 608380
CFH	155,4	99.4%	97.9%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CHD7	137	99.9%	99.4%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHM	100,3	98.3%	92.0%	Choroideremia, 303100
CHMP4B	139	100.0%	98.9%	Cataract 31, multiple types, 605387
CHRDL1	88,1	99.9%	98.8%	Megalocornea 1, X-linked, 309300
CHST6	279,8	100.0%	100.0%	Macular corneal dystrophy, 217800
CIB2	198	99.9%	99.4%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CISD2	113,6	83.4%	83.3%	Wolfram syndrome 2, 604928
CLCC1	99,4	99.9%	98.3%	No OMIM phenotype
CLN3	114,7	92.6%	91.9%	Ceroid lipofuscinosis, neuronal, 3, 204200

CLN5	138,7	99.9%	98.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	132,3	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	144,5	83.5%	83.5%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLRN1	135,3	100.0%	99.4%	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CLUAP1	133,3	99.9%	99.5%	No OMIM phenotype Leber congenital amaurosis (Soens (2016) Genet Med 18,1044)
CNGA1	110,4	92.5%	86.5%	Retinitis pigmentosa 49, 613756
CNGA3	149,6	100.0%	99.7%	Achromatopsia 2, 216900
CNGB1	107,4	99.5%	98.0%	Retinitis pigmentosa 45, 613767
CNGB3	101,5	98.6%	93.9%	Achromatopsia 3, 262300 Macular degeneration, juvenile, 248200
CNNM4	161,6	100.0%	99.5%	Jalili syndrome, 217080
COL11A1	96,6	97.9%	94.0%	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL18A1	133,5	99.5%	96.9%	Knobloch syndrome, type 1, 267750
COL25A1	131,9	99.5%	98.9%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL2A1	112,2	100.0%	99.7%	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 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, 0
COL8A2	119,3	100.0%	99.6%	Corneal dystrophy, Fuchs endothelial, 1, 136800

				Corneal dystrophy, posterior polymorphous 2, 609140
COL9A1	132,3	100.0%	99.7%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	95,2	99.9%	98.8%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
CRB1	155	100.0%	100.0%	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105
CRX	196,6	100.0%	100.0%	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	131	95.8%	90.7%	Cataract 9, multiple types, 604219
CRYAB	94	99.7%	96.8%	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CRYBA1	106	100.0%	99.7%	Cataract 10, multiple types, 600881
CRYBA2	168,4	100.0%	100.0%	?Cataract 42, 115900
CRYBA4	121,5	100.0%	100.0%	Cataract 23, 610425
CRYBB1	125,2	100.0%	99.8%	Cataract 17, multiple types, 611544
CRYBB2	137,6	100.0%	100.0%	Cataract 3, multiple types, 601547
CRYBB3	139,5	100.0%	100.0%	Cataract 22, 609741
CRYGB	96,8	99.8%	97.4%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	127,3	100.0%	99.6%	Cataract 2, multiple types, 604307
CRYGD	103,2	100.0%	99.4%	Cataract 4, multiple types, 115700
CRYGS	79,8	93.9%	83.5%	Cataract 20, multiple types, 116100
CSPP1	119	100.0%	99.1%	Joubert syndrome 21, 615636
CTDP1	128,1	95.1%	88.0%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA1	109,6	99.1%	97.2%	Macular dystrophy, patterned, 2, 608970
CTNNB1	127,4	100.0%	99.9%	Colorectal cancer, somatic, 114500 Exudative vitreoretinopathy 7, 617572 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTSD	171	99.8%	97.8%	Ceroid lipofuscinosis, neuronal, 10, 610127

CWC27	84,5	99.8%	97.5%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CYP1B1	134,4	100.0%	100.0%	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300
CYP4V2	137,2	99.9%	98.3%	Bietti crystalline corneoretinal dystrophy, 210370
DCN	129,7	95.7%	95.2%	Corneal dystrophy, congenital stromal, 610048
DDHD1	161,6	99.9%	98.4%	Spastic paraplegia 28, autosomal recessive, 609340
DHDDS	81	97.1%	93.8%	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861
DHX38	104,9	100.0%	99.5%	Retinitis pigmentosa 84, 618220
DKC1	91,2	99.8%	97.7%	Dyskeratosis congenita, X-linked, 305000
DNM1L	119,5	99.9%	98.7%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DRAM2	130,4	100.0%	100.0%	Cone-rod dystrophy 21, 616502
DTNBP1	113,4	99.8%	97.9%	Hermansky-Pudlak syndrome 7, 614076
EFEMP1	147,8	99.9%	99.4%	Doyne honeycomb degeneration of retina, 126600
ELOVL1	85,9	99.8%	97.5%	No OMIM phenotype
ELOVL4	104,4	99.9%	99.1%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
EMC1	105,7	100.0%	98.9%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EPG5	110,3	99.3%	97.9%	Vici syndrome, 242840
EPHA2	157	100.0%	99.8%	Cataract 6, multiple types, 116600
EXOSC2	110,3	100.0%	99.9%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EYA1	120,2	99.9%	99.8%	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
EYS	132,5	99.7%	97.5%	Retinitis pigmentosa 25, 602772
FA2H	92,7	98.8%	92.5%	Spastic paraplegia 35, autosomal recessive, 612319
FAM161A	136,9	99.9%	99.1%	Retinitis pigmentosa 28, 606068
FLVCR1	146,1	99.9%	99.2%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOXC1	80,4	99.7%	97.5%	Anterior segment dysgenesis 3, multiple subtypes, 601631 Axenfeld-Rieger syndrome, type 3, 602482
FOXE3	88,3	89.7%	82.5%	Anterior segment dysgenesis 2, multiple subtypes, 610256

				Cataract 34, multiple types, 612968 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
FREM1	110,7	99.8%	98.4%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FRMD7	101	99.8%	97.8%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FTL	145,2	99.7%	96.7%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FYCO1	123,2	100.0%	100.0%	Cataract 18, autosomal recessive, 610019
FZD4	179,1	100.0%	100.0%	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
GALK1	165,2	100.0%	99.9%	Galactokinase deficiency with cataracts, 230200
GALT	152,6	100.0%	100.0%	Galactosemia, 230400
GCNT2	151,2	99.5%	99.5%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GDF3	127,9	100.0%	100.0%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF6	156,3	100.0%	100.0%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Multiple synostoses syndrome 4, 617898
GFER	90,6	100.0%	99.6%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GJA1	156,2	100.0%	100.0%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 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	162,2	100.0%	100.0%	Cataract 14, multiple types, 601885
GJA8	156,6	100.0%	100.0%	Cataract 1, multiple types, 116200

GNAT1	176,4	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 3, 610444 Night blindness, congenital stationary, type 1G, 616389
GNAT2	100,7	99.8%	98.2%	Achromatopsia 4, 613856
GNB3	146	100.0%	99.9%	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNPTG	177,6	100.0%	98.5%	Mucolipidosis III gamma, 252605
GPR143	59,5	91.0%	79.1%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR179	146,8	100.0%	100.0%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GRHL2	116,8	100.0%	100.0%	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRK1	135,7	100.0%	100.0%	Oguchi disease-2, 613411
GRM6	138,3	98.0%	92.4%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GSN	115,5	95.6%	93.5%	Amyloidosis, Finnish type, 105120
GUCA1A	172,9	100.0%	100.0%	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	130	100.0%	99.9%	Retinitis pigmentosa 48, 613827
GUCY2D	108,9	100.0%	99.9%	?Choroidal dystrophy, central areolar 1, 215500 Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
HARS	134,8	100.0%	100.0%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HCCS	92,4	99.2%	95.2%	Linear skin defects with multiple congenital anomalies 1, 309801
HGSNAT	98,3	87.2%	86.2%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HK1	116,5	100.0%	99.7%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460
HMX1	39,8	85.4%	63.9%	Oculoauricular syndrome, 612109
HPS1	115,8	100.0%	99.9%	Hermansky-Pudlak syndrome 1, 203300
HPS3	132,7	99.9%	98.8%	Hermansky-Pudlak syndrome 3, 614072
HPS4	128,1	100.0%	99.9%	Hermansky-Pudlak syndrome 4, 614073
HPS5	122,8	99.9%	98.7%	Hermansky-Pudlak syndrome 5, 614074
HPS6	164,6	99.9%	97.8%	Hermansky-Pudlak syndrome 6, 614075
HRAS	182,3	100.0%	100.0%	Bladder cancer, somatic, 109800

				Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Nevus sebaceous or woolly hair nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Thyroid carcinoma, follicular, somatic, 188470
HSF4	148,7	99.8%	98.5%	Cataract 5, multiple types, 116800
HSPG2	119,8	99.5%	98.8%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
IDH3B	128,2	95.5%	95.4%	Retinitis pigmentosa 46, 612572
IFT140	117,6	99.9%	99.2%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	94,5	100.0%	99.4%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	115,8	100.0%	100.0%	?Bardet-Biedl syndrome 19, 615996
IFT43	112,4	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT74	84,7	99.1%	95.8%	?Bardet-Biedl syndrome 20, 617119
IFT81	92,3	93.6%	89.0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IMPDH1	53,6	95.3%	84.1%	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	90,7	99.9%	98.6%	Macular dystrophy, vitelliform, 4, 616151
IMPG2	125,6	99.4%	97.6%	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INPP5E	116,8	100.0%	98.6%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	143,7	100.0%	99.9%	Nephronophthisis 2, infantile, 602088
IQCB1	93,3	91.6%	80.0%	Senior-Loken syndrome 5, 609254
IRX1	151,6	96.7%	89.2%	No OMIM phenotype ?Macular dystrophy, North Carolina (Small (2016) Ophthalmology 123,9)
ITPR1	131,2	100.0%	99.7%	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
JAG1	133,7	99.2%	97.1%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500

JAM3	126,9	100.0%	99.9%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KCNJ13	138,9	100.0%	100.0%	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNV2	138,9	100.0%	100.0%	Retinal cone dystrophy 3B, 610356
KERA	175,3	100.0%	100.0%	Cornea plana 2, autosomal recessive, 217300
KIAA1549	118,4	98.5%	97.4%	No OMIM phenotype
KIF11	92,1	97.8%	94.5%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF21A	120,7	99.9%	99.1%	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF7	105,2	98.2%	93.5%	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KIZ	145	99.9%	98.1%	Retinitis pigmentosa 69, 615780
KLHL7	116,2	100.0%	99.5%	Cold-induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943
KRT12	134,2	99.7%	97.3%	Meesmann corneal dystrophy, 122100
KRT3	113,3	100.0%	99.7%	Meesmann corneal dystrophy, 122100
LAMA1	116	100.0%	99.5%	Poretti-Boltshauser syndrome, 615960
LAMB2	166,5	100.0%	99.6%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LCA5	139,9	99.8%	98.9%	Leber congenital amaurosis 5, 604537
LEMD2	103,2	100.0%	99.6%	Cataract 46, juvenile-onset, 212500
LIM2	112,9	100.0%	99.5%	Cataract 19, multiple types, 615277
LRAT	240,7	100.0%	100.0%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRIT3	108	94.3%	93.1%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	114,9	99.4%	97.6%	Albinism, oculocutaneous, type VII, 615179
LRP2	139,2	100.0%	99.9%	Donnai-Barrow syndrome, 222448
LRP5	168,1	99.8%	98.7%	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875

				van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LRPAP1	141	100.0%	99.6%	Myopia 23, autosomal recessive, 615431
LSS	127,6	100.0%	99.7%	Cataract 44, 616509 Hypotrichosis 14, 618275
LTBP2	112,9	99.9%	99.3%	?Weill-Marchesani syndrome 3, recessive, 614819 Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LYST	136,3	99.4%	97.8%	Chediak-Higashi syndrome, 214500
LZTFL1	117	99.8%	99.2%	Bardet-Biedl syndrome 17, 615994
MAB21L2	237,8	100.0%	100.0%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAF	88,9	87.2%	83.0%	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAK	131,8	99.5%	97.4%	Retinitis pigmentosa 62, 614181
MAPKAPK3	87,6	100.0%	99.6%	?Macular dystrophy, patterned, 3, 617111
MERTK	128,2	99.4%	98.7%	Retinitis pigmentosa 38, 613862
MFN2	122,8	100.0%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	128,1	100.0%	100.0%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	121,3	100.0%	99.6%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MIP	117,1	99.7%	97.0%	Cataract 15, multiple types, 615274
MIR184	NC	NC	NC	EDICT syndrome, 614303
MITF	141,1	100.0%	99.8%	COMMAD syndrome, 617306 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MKKS	155,7	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	92,4	99.6%	97.8%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000

MVK	121,4	91.0%	90.5%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYO7A	125,2	99.8%	98.5%	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYOC	153,4	99.9%	98.6%	Glaucoma 1A, primary open angle, 137750
NAA10	105	100.0%	98.8%	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NBAS	138,5	99.9%	99.1%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NDP	91,6	100.0%	99.5%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDUFS2	100,1	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NEK2	86,4	98.6%	92.4%	?Retinitis pigmentosa 67, 615565
NEUROD1	154,5	100.0%	99.4%	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853
NHS	111	98.5%	96.0%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NMNAT1	113,5	100.0%	98.5%	Leber congenital amaurosis 9, 608553
NPHP1	121,2	99.8%	98.5%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	121,4	99.8%	98.5%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	125,6	100.0%	99.7%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NR2E3	105,1	99.9%	98.6%	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	222,3	100.0%	100.0%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRL	114,8	99.9%	98.3%	Retinal degeneration, autosomal recessive, clumped pigment type, 0 Retinitis pigmentosa 27, 613750
NYX	131,7	99.6%	98.1%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	68,2	81.7%	70.1%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCA2	116,8	99.7%	97.7%	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	106,2	99.8%	98.3%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	51,9	85.8%	70.8%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OPA1	124,7	99.7%	97.4%	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 {Glaucoma, normal tension, susceptibility to}, 606657
OPA3	156,6	100.0%	99.2%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPN1LW	58,7	68.3%	61.3%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	56,9	66.1%	57.9%	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800
OTX2	127,4	100.0%	99.3%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OVOL2	120,8	99.8%	97.7%	Corneal dystrophy, posterior polymorphous, 1, 122000
P3H2	98,8	99.9%	99.4%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	114,8	99.9%	98.4%	Myopia 25, autosomal dominant, 617238
PANK2	154,1	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX2	184,1	100.0%	100.0%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX6	116,5	100.0%	99.8%	?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 ?Morning glory disc anomaly, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 Foveal hypoplasia 1, 136520 Keratitis, 148190

				Optic nerve hypoplasia, 165550
PCDH15	140,3	99.2%	99.0%	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCYT1A	95,6	97.9%	94.4%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE6A	102,6	100.0%	99.2%	Retinitis pigmentosa 43, 613810
PDE6B	157,3	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	116,2	99.5%	97.2%	Cone dystrophy 4, 613093
PDE6D	114,7	100.0%	99.9%	?Joubert syndrome 22, 615665
PDE6G	125,8	100.0%	99.5%	Retinitis pigmentosa 57, 613582
PDE6H	58,2	98.5%	76.0%	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024
PDZD7	93,9	99.6%	97.8%	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PET100	87,9	98.0%	87.6%	Mitochondrial complex IV deficiency, 220110
PEX1	127,9	99.9%	99.3%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX2	134,9	100.0%	100.0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	94,3	100.0%	99.6%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX7	111	91.2%	89.3%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGK1	44,7	90.9%	75.9%	Phosphoglycerate kinase 1 deficiency, 300653
PHOX2A	56	98.6%	88.9%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	74	99.9%	96.9%	Refsum disease, 266500
PIKFYVE	136,4	99.9%	99.7%	Corneal fleck dystrophy, 121850
PITX2	164,8	100.0%	99.5%	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PITX3	88,5	100.0%	99.5%	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, autosomal recessive, 610623

PLA2G5	104,8	100.0%	100.0%	[Fleck retina, familial benign], 228980
PLK4	149,7	99.8%	98.2%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PNPLA6	137,9	99.9%	99.5%	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
POC1B	79,6	99.7%	97.9%	Cone-rod dystrophy 20, 615973
POC5	133,7	99.1%	97.0%	No OMIM phenotype
POMGNT1	115,5	100.0%	99.6%	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 Retinitis pigmentosa 76, 617123
PPT1	136,6	90.2%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	95,6	100.0%	100.0%	Retinitis pigmentosa 36, 610599
PRDM13	186,6	99.9%	98.3%	No OMIM phenotype Macular dystrophy, North Carolina (Small (2016) Ophthalmology 123, 9)
PRDM5	137,8	99.6%	98.7%	Brittle cornea syndrome 2, 614170
PRIMPOL	118,1	97.7%	94.6%	Myopia 22, autosomal dominant, 615420
PROM1	107,4	97.8%	95.8%	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PRPF3	73,2	98.4%	94.8%	Retinitis pigmentosa 18, 601414
PRPF31	117,4	99.8%	97.1%	Retinitis pigmentosa 11, 600138
PRPF4	124,9	100.0%	99.5%	Retinitis pigmentosa 70, 615922
PRPF6	112,3	100.0%	99.6%	Retinitis pigmentosa 60, 613983
PRPF8	103,4	99.9%	98.9%	Retinitis pigmentosa 13, 600059
PRPH2	203	100.0%	100.0%	Choroidal 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 form, 608133 Retinitis punctata albescens, 136880
PRSS56	95,4	100.0%	99.2%	Microphthalmia, isolated 6, 613517
PXDN	138,4	100.0%	99.6%	Anterior segment dysgenesis 7, with sclerocornea, 269400
RAB28	66,6	98.9%	92.3%	Cone-rod dystrophy 18, 615374

RAB3GAP2	91,6	99.7%	96.9%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RARB	93,2	100.0%	100.0%	Microphthalmia, syndromic 12, 615524
RAX	135,9	99.9%	98.5%	Microphthalmia, isolated 3, 611038
RAX2	89,2	100.0%	99.9%	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RBP3	150	100.0%	100.0%	?Retinitis pigmentosa 66, 615233
RBP4	137,9	99.2%	95.8%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	95,9	99.8%	99.3%	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	165,7	100.0%	100.0%	Leber congenital amaurosis 12, 610612
RDH11	93	99.9%	98.5%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	80,7	99.8%	97.2%	Leber congenital amaurosis 13, 612712
RDH5	167,7	100.0%	100.0%	Fundus albipunctatus, 136880
REEP6	206,2	100.0%	99.7%	Retinitis pigmentosa 77, 617304
RGS9	107,8	98.7%	96.7%	Bradyopsia, 608415
RGS9BP	143,6	100.0%	100.0%	Bradyopsia, 608415
RHO	165,3	100.0%	100.0%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIMS1	126	99.8%	98.6%	Cone-rod dystrophy 7, 603649
RLBP1	120,4	100.0%	99.7%	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
ROM1	126,1	100.0%	99.9%	Retinitis pigmentosa 7, digenic form, 608133
RP1	109,4	91.4%	90.8%	Retinitis pigmentosa 1, 180100
RP1L1	135,2	100.0%	100.0%	Occult macular dystrophy, 613587
RP2	155,8	100.0%	99.8%	Retinitis pigmentosa 2, 312600
RP9	64,5	91.4%	78.5%	?Retinitis pigmentosa 9, 180104
RPE65	131,9	100.0%	99.8%	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	81,3	82.4%	75.5%	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	128	100.0%	99.7%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	123,4	96.7%	95.4%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RS1	51,4	98.7%	87.3%	Retinoschisis, 312700
RTN4IP1	79,6	100.0%	98.0%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
SAG	127	100.0%	99.9%	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SAMD11	98,9	94.8%	88.6%	No OMIM phenotype ?Autism spectrum disorder (Chapman (2015) Hum Genet 134, 1055)
SC5D	153,6	99.8%	99.3%	Lathosterolosis, 607330
SCAPER	138,7	98.2%	96.4%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCO2	115,7	100.0%	99.9%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SDCCAG8	124,1	100.0%	99.7%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEMA4A	124,3	100.0%	99.3%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SGSH	140,2	97.6%	94.7%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SHH	147,1	100.0%	100.0%	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SIPA1L3	171,8	100.0%	99.5%	?Cataract 45, 616851
SIX6	278,5	100.0%	100.0%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SLC16A12	128,5	100.0%	99.9%	Cataract 47, juvenile, with microcornea, 612018
SLC24A1	167	100.0%	100.0%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	104,1	99.9%	99.3%	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A46	173	99.8%	98.3%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC33A1	132	99.7%	97.7%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC38A8	71,5	99.4%	95.5%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A5	130,2	100.0%	99.4%	Myopia 24, autosomal dominant, 615946

SLC45A2	115,2	100.0%	99.8%	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	157,4	100.0%	100.0%	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal endothelial dystrophy, autosomal recessive, 217700
SLC52A2	185,4	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC7A14	145,3	100.0%	100.0%	Retinitis pigmentosa 68, 615725
SMOC1	115,1	99.8%	98.2%	Microphthalmia with limb anomalies, 206920
SNRNP200	118,8	99.7%	98.3%	Retinitis pigmentosa 33, 610359
SOX2	230	100.0%	100.0%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX5	89,4	99.7%	97.6%	Lamb-Shaffer syndrome, 616803
SPATA7	122,7	99.4%	97.4%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPP2	110,7	100.0%	99.8%	No OMIM phenotype Retinitis pigmentosa (Li (2015) Sci Rep 5,14867) ?Autism (Neale (2012) Nature 485,242)
STRA6	117,6	100.0%	99.8%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
TACSTD2	268,7	100.0%	99.9%	Corneal dystrophy, gelatinous drop-like, 204870
TCTN1	94,8	95.6%	92.3%	Joubert syndrome 13, 614173
TCTN3	116,3	100.0%	99.9%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDRD7	136,7	99.8%	98.7%	Cataract 36, 613887
TEAD1	132,3	100.0%	99.6%	Sveinsson chorioretinal atrophy, 108985
TENM3	148,6	99.8%	99.3%	Microphthalmia, isolated, with coloboma 9, 615145
TGFBI	114,1	99.9%	98.6%	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	46,3	94.6%	79.9%	Mohr-Tranebjaerg syndrome, 304700

TIMP3	137,5	100.0%	100.0%	Sorsby fundus dystrophy, 136900
TMCO3	121,6	100.0%	99.2%	No OMIM phenotype
TMEM126A	104,8	96.2%	82.8%	Optic atrophy 7, 612989
TMEM138	82,7	100.0%	99.2%	Joubert syndrome 16, 614465
TMEM216	88	99.7%	95.7%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	101,1	100.0%	99.3%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	117,7	100.0%	99.2%	Joubert syndrome 14, 614424
TMEM67	83,1	99.1%	94.6%	?RHYNS syndrome, 602152 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TOPORS	182,2	100.0%	100.0%	Retinitis pigmentosa 31, 609923
TPP1	123,7	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAF3IP1	84,2	99.4%	97.1%	Senior-Loken syndrome 9, 616629
TREX1	233,4	100.0%	100.0%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRIM32	123	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRNT1	101,5	99.2%	96.5%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPM1	128,8	100.0%	99.0%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TSPAN12	135,3	100.0%	99.4%	Exudative vitreoretinopathy 5, 613310
TTC8	115,2	99.8%	98.8%	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
TTLL5	136,1	100.0%	98.7%	Cone-rod dystrophy 19, 615860
TUB	103,4	100.0%	99.4%	?Retinal dystrophy and obesity, 616188
TUBA3D	103,6	99.3%	95.9%	Keratoconus 9, 617928
TUBB3	121,3	99.8%	98.4%	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638

TUBB4B	86,1	100.0%	99.9%	Leber congenital amaurosis with early-onset deafness, 617879
TUBGCP4	104,6	98.0%	94.7%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TULP1	117,9	100.0%	99.6%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TYR	147,9	100.0%	99.9%	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	152,3	100.0%	100.0%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBIAD1	187,7	99.8%	97.4%	Corneal dystrophy, Schnyder type, 121800
UNC119	115,7	100.0%	99.7%	?Cone-rod dystrophy, 0 ?Immunodeficiency 13, 615518
UNC45B	114,7	99.9%	99.2%	?Cataract 43, 616279
USH1C	92,5	99.9%	99.2%	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	191,3	99.9%	98.8%	Usher syndrome, type 1G, 606943
USH2A	129,3	100.0%	99.7%	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP45	96,7	99.6%	98.1%	No OMIM phenotype
VAX1	95,1	99.5%	95.7%	?Microphthalmia, syndromic 11, 614402
VCAN	153,1	100.0%	100.0%	Wagner syndrome 1, 143200
VIM	129,2	99.7%	98.3%	Cataract 30, pulverulent, 116300
VPS13B	134,5	99.3%	98.0%	Cohen syndrome, 216550
VSX1	66,6	97.8%	89.5%	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300
VSX2	120,2	100.0%	99.8%	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
WDPCP	106,7	97.8%	94.9%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	126,8	100.0%	99.2%	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307

WFS1	189,9	100.0%	99.9%	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WHRN	132,8	99.9%	99.0%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WRN	124,8	99.7%	98.8%	Werner syndrome, 277700
YAP1	91,1	98.2%	92.3%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YME1L1	103,9	98.2%	93.5%	?Optic atrophy 11, 617302
ZEB1	151,3	100.0%	99.8%	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZNF408	144,5	100.0%	100.0%	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469
ZNF423	192,9	100.0%	100.0%	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF469	157,6	100.0%	100.0%	Brittle cornea syndrome 1, 229200
ZNF513	135,3	100.0%	100.0%	?Retinitis pigmentosa 58, 613617
ZNF644	155,4	99.9%	99.8%	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 : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors