VISION DISORDERS GENE PANEL DG 2.12 (395 genes)

Gene	*Median*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ABCA4	128.2	100	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	116.6	93	92	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABHD12	107	97	87	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACBD5	145.5	97	96	No OMIM phenotype Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) ?Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
ACO2	129.6	95	91	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
ADAM9	146.3	98	93	Cone-rod dystrophy 9, 612775
ADAMTS18	147.6	99	98	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADIPOR1	111.4	100	98	No OMIM phenotype syndromic retinitis pigmentosa (Xy (2016) Hum Mutat 37(3):246-249
AGBL1	134.4	100	100	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	111.2	100	99	Retinitis pigmentosa 75,617023
AGK	112.2	99	96	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AHI1	139.1	99	95	Joubert syndrome-3, 608629
AIPL1	116.3	100	99	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
ALDH1A3	104.9	93	89	Microphthalmia, isolated 8, 615113

ALMS1	179.4	99	99	Alstrom syndrome, 203800
AP3B1	95	97	90	Hermansky-Pudlak syndrome 2, 608233
AP3D1	121.2	98	97	?Hermansky-Pudlak syndrome 10, 617050
APOPT1	67.9	86	83	Mitochondrial complex IV deficiency, 220110
ARHGEF18	114.3	97	93	Retinitis pigmentosa 78, 617433
ARL13B	97.3	98	92	Joubert syndrome 8, 612291
ARL2BP	66.4	88	79	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	84.9	98	93	No OMIM phenotype ?Retinitis pigmentosa (Strom (2016) PLoS One 11,e0150944)
ARL6	85.2	99	95	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ASPH	117.1	98	93	Traboulsi syndrome, 601552
ASRGL1	137.3	100	99	No OMIM phenotype Retinal degeneration (Biswas (2016) Hum Mol Genet 25,2483)
ATF6	133.9	100	99	Achromatopsia 7, 616517
B3GALTL	101.2	97	93	Peters-plus syndrome, 261540
BBIP1	131.9	99	94	?Bardet-Biedl syndrome 18, 615995
BBS1	149.1	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	172.3	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	208.7	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	181.9	100	99	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	135.2	99	97	Bardet-Biedl syndrome 4, 615982
BBS5	110.5	97	90	Bardet-Biedl syndrome 5, 615983
BBS7	120.5	98	91	Bardet-Biedl syndrome 7, 615984
BBS9	112.5	96	93	Bardet-Biedl syndrome 9, 615986
BCOR	111	99	96	Microphthalmia, syndromic 2, 300166
BEST1	144.6	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	98.1	98	89	Cataract 33, 611391
BFSP2	89.5	99	97	Cataract 12, multiple types, 611597
BLOC1S3	28.7	88	64	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	97.3	98	91	Hermansky-pudlak syndrome 9, 614171
BMP4	151.9	100	99	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C10orf11	154.5	99	99	Albinism, oculocutaneous, type VII, 615179
C12orf65	88.1	97	91	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	94.3	100	99	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1QTNF5	151.4	80	65	Retinal degeneration, late-onset, autosomal dominant, 605670
C21orf2	104.8	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	124.7	99	98	Retinitis pigmentosa 54, 613428
C5orf42	122.4	98	95	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C8orf37	126.4	100	99	Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
CA4	142.6	100	99	Retinitis pigmentosa 17, 600852
CABP4	98.5	99	97	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1F	100.2	99	98	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA2D4	112.1	99	97	Retinal cone dystrophy 4, 610478
CAPN5	166.4	100	99	Vitreoretinopathy, neovascular inflammatory, 193235
CC2D2A	127.4	99	97	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCDC41	96.9	98	89	Nephronophthisis 18, 615862

CDH23	195.1	100	99	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067
CDH3	159.6	99	97	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDH7	154.1	99	95	No OMIM phenotype
CDHR1	154.4	99	98	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CEP164	94.4	99	97	Nephronophthisis 15, 614845
CEP250	105.1	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	66	88	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	83.6	97	89	Joubert syndrome 15, 614464
CEP78	112.1	97	94	Cone-rod dystrophy and hearing loss, 617236
CERKL	100.2	98	92	Retinitis pigmentosa 26, 608380
CFH	183.1	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	103.1	96	87	Choroideremia, 303100
CHMP4B	139.8	99	99	Cataract 31, multiple types, 605387
CHST6	334.5	100	100	Macular corneal dystrophy, 217800
CIB2	230.2	99	99	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CLN3	121.3	98	95	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	146.3	98	92	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	131.8	98	95	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	196.4	100	100	Ceroid lipofuscinosis, neuronal, 8, 600143

				Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLRN1	157.2	100	99	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CLUAP1	152.8	99	99	No OMIM phenotype Leber congenital amaurosis (Soens (2016) Genet Med 18,1044)
CNGA1	127.2	89	84	Retinitis pigmentosa 49, 613756
CNGA3	167.8	100	99	Achromatopsia-2, 216900
CNGB1	102.7	98	94	Retinitis pigmentosa 45, 613767
CNGB3	101.3	97	93	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200
CNNM4	193.1	98	97	Jalili syndrome, 217080
COL11A1	90.8	94	89	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL18A1	88.8	93	87	Knobloch syndrome, type 1, 267750
COL2A1	103.6	99	98	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	37.4	84	69	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140

COL9A1	120.5	99	96	Stickler syndrome, type IV, 614134 /?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	65.1	98	88	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284 {Intervertebral disc disease, susceptibility to}, 603932
CRB1	193.6	100	100	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, autosomal recessive, 600105
CRX	114.4	99	98	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	135.7	92	86	Cataract 9, multiple types, 604219
CRYAB	125.9	99	98	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869
CRYBA1	135.6	100	99	Cataract 10, multiple types, 600881
CRYBA2	150.7	100	100	?Cataract 42, 115900
CRYBA4	117.7	100	100	Cataract 23, 610425
CRYBB1	129.3	100	99	Cataract 17, multiple types, 611544
CRYBB2	150.7	100	100	Cataract 3, multiple types, 601547
CRYBB3	144.4	100	100	Cataract 22, autosomal recessive, 609741
CRYGB	97.3	99	97	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	128.8	100	99	Cataract 2, multiple types, 604307
CRYGD	100.3	100	99	Cataract 4, multiple types, 115700
CRYGS	105.6	96	87	Cataract 20, multiple types, 116100
CSPP1	111.9	99	97	Joubert syndrome 21, 615636
CTDP1	105.3	86	83	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA1	125.7	99	99	Macular dystrophy,patterned,608970 Gastric cancer, diffuse (Majewski (2012) J Pathol epub)
CTNNB1	163.8	100	99	Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600

CTSD	164	97	95	Ceroid lipofuscinosis, neuronal, 10, 610127
CWC27	74.7	97	89	Retinitis pigmentosa with or without skeletal anomalies,250410
CYP1B1	134.9	100	100	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229
CYP4V2	147.7	99	98	Bietti crystalline corneoretinal dystrophy, 210370
DCN	140.1	95	94	Corneal dystrophy, congenital stromal, 610048
DDHD1	141.7	97	94	Spastic paraplegia 28, autosomal recessive, 609340
DFNB31	114.2	99	98	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DHDDS	93.5	97	94	Retinitis pigmentosa 59, 613861
DHX38	130.5	99	98	No OMIM phenotype Retinitis pigmentosa,early-onset with macular coloboma (Ajmal (2014) J Med Genet 51,444)
DKC1	112.6	99	98	Dyskeratosis congenita, X-linked, 305000
DRAM2	131.7	100	99	Cone-rod dystrophy 21, 616502
DTNBP1	113.9	99	95	Hermansky-Pudlak syndrome 7, 614076
EFEMP1	167.9	100	99	Doyne honeycomb degeneration of retina, 126600
ELOVL4	91.9	99	98	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190
EPG5	126.1	99	97	Vici syndrome, 242840
EPHA2	175.4	99	97	Cataract 6, multiple types, 116600
EXOSC2	142.3	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	144.4	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	135.7	98	94	Retinitis pigmentosa 25, 602772
FA2H	94.2	87	79	Spastic paraplegia 35, autosomal recessive, 612319
FAM161A	115.3	98	95	Retinitis pigmentosa 28, 606068
FLVCR1	139.4	99	95	Ataxia, posterior column, with retinitis pigmentosa, 609033

FOXC1	32.8	86	68	Axenfeld-Rieger syndrome, type 3, 602482 Iridogoniodysgenesis, type 1, 601631 Iris hypoplasia and glaucoma, 601631 Rieger or Axenfeld anomalies, 602482
FOXE3	20.8	69	48	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256 Cataract 34,multiple types, 612968
FRMD7	114.9	99	98	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FTL	148.1	98	93	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FYCO1	123.9	100	100	Cataract 18, autosomal recessive, 610019
FZD4	224.3	100	99	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
GALK1	122.7	99	97	Galactokinase deficiency with cataracts, 230200
GALT	155.5	100	99	Galactosemia, 230400
GCNT2	167.9	100	100	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GDF3	135.1	100	100	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF6	75	98	89	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GFER	76.3	93	75	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076

GJA1	246.5	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	165.2	100	99	Cataract 14, multiple types, 601885
GJA8	153.9	100	100	Cataract 1, multiple types, 116200
GNAT1	154.2	100	100	Night blindness, congenital stationary, autosomal dominant 3, 610444 ?Night blindness, congenital stationary, type 1G, 616389
GNAT2	130.9	99	99	Achromatopsia-4, 613856
GNB3	179.2	100	100	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNPTG	153.1	96	90	Mucolipidosis III gamma, 252605
GPR143	60.5	85	74	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR179	133.5	100	99	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR98	139.9	99	96	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
GRK1	126.4	100	99	Oguchi disease-2, 613411
GRM6	151.9	93	86	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GSN	120.7	97	91	Amyloidosis, Finnish type, 105120
GUCA1A	162.6	100	100	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	144.2	100	99	Retinitis pigmentosa 48, 613827
GUCY2D	91.6	98	91	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
HARS	159.6	100	100	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504

HCCS	107.4	99	99	Linear skin defects with multiple congenital anomalies 1, 309801
HGSNAT	95.2	81	80	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HK1	140.9	100	99	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285
HMX1	23	56	42	Oculoauricular syndrome, 612109
HPS1	118	100	99	Hermansky-Pudlak syndrome 1, 203300
HPS3	135.2	99	96	Hermansky-Pudlak syndrome 3, 614072
HPS4	144.5	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	133	99	98	Hermansky-Pudlak syndrome 5, 614074
HPS6	139.2	91	84	Hermansky-Pudlak syndrome 6, 614075
HRAS	164.9	99	98	Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Bladder cancer, somatic}, 109800 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 {Spitz nevus or nevus spilus, somatic}, 137550 {Thyroid carcinoma, follicular, somatic}, 188470
HSF4	103.1	97	94	Cataract 5, multiple types, 116800
IDH3B	173.6	100	100	Retinitis pigmentosa 46, 612572
IFT140	115	100	99	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	116.5	100	99	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	131.8	100	99	?Bardet-Biedl syndrome 19, 615996
IFT74	80.2	97	88	?Bardet-Biedl syndrome 20, 617119
IFT81	92.8	88	81	No OMIM phenotype Asphyxiating thoracic dystrophy (Duran (2016) Sci Rep 6, 34232) Short-rib polydactyly syndrome (Duran (2016) Sci Rep 6, 34232) Ciliopathy (Perrault (2015) J Med Genet 52,657)
IMPDH1	61.3	87	83	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	103.3	99	97	Macular dystrophy, vitelliform, 4, 616151
IMPG2	154.4	99	97	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581

INPP5E	89.3	95	90	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	159.6	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	92.2	89	75	Senior-Loken syndrome 5, 609254
IRX1	105.1	83	81	No OMIM phenotype ?Macular dystrophy, North Carolina (Small (2016) Ophthalmology 123,9)
JAG1	148.6	98	97	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
JAM3	144.9	99	98	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KCNJ13	210.6	100	99	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNV2	137.7	100	100	Retinal cone dystrophy 3B, 610356
KERA	191.2	100	100	Cornea plana congenita, recessive, 217300
KIF11	83.6	97	94	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF7	85.8	93	88	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
KIZ	177.7	98	95	Retinitis pigmentosa 69, 615780
KLHL7	120.5	99	97	Cold induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943
KRT12	126.2	98	95	Meesmann corneal dystrophy, 122100
KRT3	105.4	100	99	Meesmann corneal dystrophy, 122100
LAMA1	137.3	100	99	Poretti-Boltshauser syndrome, 615960
LAMB2	202	100	99	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LCA5	127.6	97	95	Leber congenital amaurosis 5, 604537
LEMD2	68.7	89	80	Cataract 46, juvenile-onset, 212500
LEPREL1	100.2	99	93	Myopia, high, with cataract and vitreoretinal degeneration, 614292
LIM2	103.6	100	98	Cataract 19, multiple types, 615277

LRAT	298.3	100	100	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRIT3	142.6	94	94	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRP5	190.1	98	97	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
LRPAP1	138.7	99	97	Myopia 23, autosomal recessive, 615431
LSS	128.1	100	99	Cataract 44, 616509
LTBP2	104.7	99	97	Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 Weill-Marchesani syndrome 3, recessive, 614819
LYST	134.6	97	93	Chediak-Higashi syndrome, 214500
LZTFL1	108.7	99	95	Bardet-Biedl syndrome 17, 615994
MAB21L2	245.9	100	100	Microphthalmia, syndromic 14, 615877
MAF	60.2	77	72	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAK	142.6	95	94	Retinitis pigmentosa 62, 614181
MAPKAPK3	93.1	98	96	?Macular dystrophy, patterned, 3, 617111
MERTK	170.1	100	99	Retinitis pigmentosa 38, 613862
MFN2	150.9	100	99	Charcot-Marie-Tooth disease, type 2A2A, 609260 Charcot-Marie-Tooth disease, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	121.5	100	100	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD8	124.8	99	98	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MIP	132.1	99	96	Cataract 15, multiple types, 615274

MIR184	NC	NC	NC	EDICT syndrome, 614303
MITF	155.2	100	99	Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MKKS	225.1	89	89	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	115.6	99	98	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000
MVK	136.7	100	99	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYO7A	135	99	98	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYOC	172.5	100	99	Glaucoma 1A, primary open angle, 137750
NAA10	103.3	98	96	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NBAS	145.3	99	97	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NDP	117.4	100	100	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDUFS2	117.8	100	100	Mitochondrial complex I deficiency, 252010
NEK2	114	99	95	?Retinitis pigmentosa 67, 615565
NEUROD1	166.9	100	100	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853
NHS	127.9	94	93	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NMNAT1	137.9	100	99	Leber congenital amaurosis 9, 608553
NPHP1	117.6	98	96	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	114.3	99	95	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387

				Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	137.3	99	99	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NR2E3	93.7	99	99	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	201.8	99	98	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRL	69.1	99	95	Retinal degeneration, autosomal recessive, clumped pigment type Retinitis pigmentosa 27, 613750
NYX	95.5	98	96	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	89.2	77	70	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCA2	139.8	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	123	98	96	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	53	84	68	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OPA1	122.5	99	94	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	128.2	99	97	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPN1LW	77.5	67	62	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	67.6	68	60	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800

OTX2	155.1	100	99	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
P4HA2	159	100	99	Myopia 25, autosomal dominant, 617238
PANK2	146.6	99	93	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX2	168.6	99	99	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX6	121.9	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	155	99	98	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCYT1A	113.6	98	94	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE6A	126	100	99	Retinitis pigmentosa 43, 613810
PDE6B	148.1	100	100	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	137	99	96	Cone dystrophy 4, 613093
PDE6D	106.1	100	99	?Joubert syndrome 22, 615665
PDE6G	95.6	99	96	Retinitis pigmentosa 57, 613582
PDE6H	68.4	97	77	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024
PDZD7	96.7	100	98	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PET100	94.5	88	74	Mitochondrial complex IV deficiency, 220110

PEX1	115.7	97	95	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX2	147	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX7	113.5	89	82	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PGK1	54.9	93	81	Phosphoglycerate kinase 1 deficiency, 300653
PHYH	74.7	97	90	Refsum disease, 266500
PIKFYVE	141.5	99	98	Corneal fleck dystrophy, 121850
PITX2	147.9	99	97	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PITX3	40.2	95	82	Anterior segment mesenchymal dysgenesis, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623
PLA2G5	125.4	100	100	[Fleck retina, familial benign], 228980
PLK4	145.6	99	96	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PNPLA6	122.4	99	98	Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020 ?Laurence-Moon syndrome, 245800
POC1B	77.6	96	92	Cone-rod dystrophy 20, 615973
POMGNT1	126.8	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	172.9	100	100	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	89.4	100	99	Retinitis pigmentosa 36, 610599
PRDM13	122.8	92	87	No OMIM phenotype Macular dystrophy, North Carolina (Small (2016) Ophthalmology 123, 9)
PRDM5	129.4	99	95	Brittle cornea syndrome 2, 614170

PRIMPOL	110	95	90	Myopia 22,autosomal dominant,615420
PROM1	111.5	95	92	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PRPF3	86	98	96	Retinitis pigmentosa 18, 601414
PRPF31	116.9	97	91	Retinitis pigmentosa 11, 600138
PRPF4	149.6	100	99	Retinitis pigmentosa 70, 615922
PRPF6	130.9	100	100	Retinitis pigmentosa 60, 613983
PRPF8	139.2	99	99	Retinitis pigmentosa 13, 600059
PRPH2	244.1	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	51.5	96	84	Microphthalmia, isolated 6, 613517
PXDN	164	99	98	Corneal opacification and other ocular anomalies, 269400
RAB28	52	96	87	Cone-rod dystrophy 18, 615374
RARB	138.7	100	100	Microphthalmia, syndromic 12, 615524
RAX	82.9	88	77	Microphthalmia, isolated 3, 611038
RAX2	52.4	90	67	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RBP3	155.4	100	100	?Retinitis pigmentosa 66, 615233
RBP4	99.7	99	96	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	122.5	100	99	[Beta-glycopyranoside tasting] {Alcohol dependence, susceptibility to}, 103780
RD3	161.6	100	99	Leber congenital amaurosis 12, 610612
RDH11	119.4	100	99	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	94.5	100	98	Leber congenital amaurosis 13, 612712
RDH5	160.2	100	99	Fundus albipunctatus, 136880
REEP6	172.2	99	96	Retinitis pigmentosa 77, 617304

RGR	126.9	100	99	Retinitis pigmentosa 44, 613769
RGS9	104	100	99	Bradyopsia, 608415
RGS9BP	93.9	100	99	Bradyopsia, 608415
RHO	210.5	100	100	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RIMS1	125.7	98	95	Cone-rod dystrophy 7, 603649
RLBP1	144.9	100	100	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
ROM1	115.3	100	99	Retinitis pigmentosa 7, digenic, 608133
RP1	140	99	99	Retinitis pigmentosa 1, 180100
RP1L1	94.9	100	99	Occult macular dystrophy, 613587
RP2	180.9	100	98	Retinitis pigmentosa 2, 312600
RP9	62.9	77	76	?Retinitis pigmentosa 9, 180104
RPE65	130.3	100	99	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	92	83	74	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	153.8	100	99	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	126.2	96	93	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RS1	60.4	97	88	Retinoschisis, 312700
RTN4IP1	98.2	99	99	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
SAG	130.8	100	99	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SCAPER	135.9	96	93	No OMIM phenotype

SCO2	113.3	100	100	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SDCCAG8	124.4	99	97	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEMA4A	127.6	99	98	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SHH	117.6	99	94	Holoprosencephaly-3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SIPA1L3	141.2	99	98	?Cataract 45, 616851
SIX6	229.1	100	100	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SLC16A12	164.4	100	99	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC24A1	218.5	100	100	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A5	114.4	99	97	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A46	204.3	95	87	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC33A1	140.7	96	90	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC38A8	76.5	99	95	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A5	119.5	100	99	Myopia 24, autosomal dominant, 615946
SLC45A2	144.2	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	153.4	100	99	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC52A2	177.8	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC7A14	192.1	100	100	Retinitis pigmentosa 68, 615725
SNRNP200	161.3	100	99	Retinitis pigmentosa 33, 610359

SOX2	129.2	98	93	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SPATA7	119.4	97	90	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPP2	144.3	100	100	No OMIM phenotype Retinitis pigmentosa (Li (2015) Sci Rep 5,14867) ?Autism (Neale (2012) Nature 485,242)
STRA6	116.6	100	99	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
TACSTD2	224	99	96	Corneal dystrophy, gelatinous drop-like, 204870
TCTN1	100.5	96	94	Joubert syndrome 13, 614173
TCTN3	127.5	100	99	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDRD7	168.4	98	97	Cataract 36, 613887
TEAD1	158.9	99	98	Sveinsson choreoretinal atrophy, 108985
TENM3	185.8	99	98	Microphthalmia, isolated, with coloboma 9, 615145
TGFBI	130.1	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	46.2	94	78	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700
TIMP3	147.4	100	100	Sorsby fundus dystrophy, 136900
TMEM126A	120.1	98	86	Optic atrophy 7, 612989
TMEM138	100.3	100	99	Joubert syndrome 16, 614465
TMEM231	111.8	100	99	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100.6	99	98	Joubert syndrome 14, 614424

TMEM67	72.9	93	83	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TOPORS	210.9	100	100	Retinitis pigmentosa 31, 609923
TPP1	146.4	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAF3IP1	90.2	96	92	Senior-Loken syndrome 9, 616629
TREX1	242.7	100	100	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRIM32	141.3	100	100	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRNT1	104.6	97	92	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPM1	161	100	99	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TSPAN12	129.3	100	99	Exudative vitreoretinopathy 5, 613310
TTC8	106.8	97	92	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTLL5	152.6	99	98	Cone-rod dystrophy 19, 615860
TUB	103.1	97	95	?Retinal dystrophy and obesity, 616188
TUBGCP4	130.6	99	96	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TULP1	98	96	91	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TYR	185.4	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	176.6	100	99	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBIAD1	249.3	98	95	Corneal dystrophy, Schnyder type, 121800
UNC119	93.1	97	90	?Cone-rod dystrophy ?Immunodeficiency 13, 615518
UNC45B	143.1	100	99	?Cataract 43, 616279
USH1C	97.7	100	99	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	195.6	98	96	Usher syndrome, type 1G, 606943
USH2A	149.1	100	99	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
VAX1	52.3	88	78	?Microphthalmia, syndromic 11, 614402
VCAN	186.4	100	100	Wagner syndrome 1, 143200
VIM	126.6	99	97	?Cataract 30, pulverulent, 116300
VPS13B	143.7	98	96	Cohen syndrome, 216550
VSX1	55.7	85	76	Keratoconus 1, 148300 ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195
VSX2	77.9	99	97	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
WDPCP	107.3	93	88	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	132	99	98	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WFS1	251.6	100	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	123.6	98	94	Werner syndrome, 277700

YAP1	95.7	87	81	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YME1L1	105.3	97	91	?Optic atrophy 11, 617302
ZEB1	192.5	100	99	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZNF408	135.9	100	100	Retinitis pigmentosa 72, 616469 ?Exudative vitreoretinopathy 6, 616468
ZNF423	251.1	100	100	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF469	93.2	98	96	Brittle cornea syndrome 1, 229200
ZNF513	110.9	100	99	Retinitis pigmentosa 58, 613617
ZNF644	156	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 version DG 2.12
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors