Whole Exome Sequencing
Gene package Vision disorders, version 3, 1-7-2017

Technical information
After DNA was enriched using Agilent Sureselect Clinical Research Exome (CRE) Capture, samples were run on the Illumina Hiseq platform. The aim is to obtain 50 million total reads per exome with a mapped fraction >0.98. The average coverage of the exome is ~50x. Data are demultiplexed by Illumina software bcl2fastq. Reads are mapped to the genome using BWA (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by Genome Analysis Toolkit (reference: http://www.broadinstitute.org/gatk/). Analysis is performed in Cartagenia using The Variant Calling File (VCF) followed by filtering. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ABCA4	Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 {Macular degeneration, age-related, 2}, 153800 Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Stargardt disease 1, 248200	601691	86	100	99
ABCB6	[Blood group, Langereis system], 111600 Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153	605452	79	100	100
ABCC6	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850	603234	66	93	92
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	613599	62	100	94
ACBD5	No OMIM phenotype	616618	89	100	100
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	91	98	92
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	77	100	100
ACTG1	Baraitser-Winter syndrome 2, 614583 Deafness 20/26, 604717	102560	104	100	100

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ADAM9	Cone-rod dystrophy 9, 612775	602713	78	100	100
ADAMTS10	Weill-Marchesani syndrome 1, recessive, 277600	608990	66	100	97
ADAMTS17	Weill-Marchesani-like syndrome, 613195	607511	82	92	89
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458	607512	83	100	98
ADAMTSL4	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, 225100	610113	72	100	99
ADGRV1	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472	602851	79	100	99
ADIPOR1	No OMIM phenotype	607945	66	99	95
AGBL1	Corneal dystrophy, Fuchs endothelial, 8, 615523	615496	43	99	91
AGBL5	Retinitis pigmentosa 75, 617023	615900	44	98	87
AGK	Cataract 38, 614691 Sengers syndrome, 212350	610345	84	97	90
AHI1	Joubert syndrome 3, 608629	608894	77	100	98
AIPL1	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393	604392	87	100	100
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	45	98	86
ALMS1	Alstrom syndrome, 203800	606844	105	100	100
ALX3	Frontonasal dysplasia 1, 136760	606014	65	87	80
ANTXR1	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089	606410	78	98	91
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	73	100	95
ARHGEF18	Retinitis pigmentosa 78, 617433	616432	52	98	86
ARL13B	Joubert syndrome 8, 612291	608922	61	100	100
ARL2BP	Retinitis pigmentosa with or without situs inversus, 615434	615407	28	80	63
ARL3	No OMIM phenotype	604695	83	100	97
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	48	95	81
ASPH	Traboulsi syndrome, 601552	600582	39	93	70
ASRGL1	No OMIM phenotype	609212	46	99	93
ATF6	Achromatopsia 7, 616517	605537	77	100	99
ATOH7	Persistent hyperplastic primary vitreous, 221900	609875	67	100	92
BBIP1	?Bardet-Biedl syndrome 18, 615995	613605	53	100	89
BBS1	Bardet-Biedl syndrome 1, 209900	209901	92	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	76	100	100
BBS12	Bardet-Biedl syndrome 12, 615989	610683	98	100	100

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BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	76	100	98
BBS4	Bardet-Biedl syndrome 4, 615982	600374	80	100	99
BBS5	Bardet-Biedl syndrome 5, 615983	603650	59	99	92
BBS7	Bardet-Biedl syndrome 7, 615984	607590	68	100	98
BBS9	Bardet-Biedl syndrome 9, 615986	607968	76	98	93
BCOR	Microphthalmia, syndromic 2, 300166	300485	52	99	94
BEST1	Bestrophinopathy, 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	607854	81	100	98
BFSP1	Cataract 33, multiple types, 611391	603307	69	100	98
BFSP2	Cataract 12, multiple types, 611597	603212	72	100	100
BLOC1S3	Hermansky-Pudlak syndrome 8, 614077	609762	48	100	100
BLOC1S6	Hermansky-pudlak syndrome 9, 614171	604310	67	100	100
BMP4	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625	112262	77	100	100
BMP7	No OMIM phenotype	112267	76	100	100
C12orf57	Temtamy syndrome, 218340	615140	85	100	100
C19ORF12	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, 615043	614297	71	100	100
C1QTNF5	Retinal degeneration, late-onset, 605670	608752	68	100	96
C21orf2	No OMIM phenotype	603191	52	98	92
C2ORF71	Retinitis pigmentosa 54, 613428	613425	67	100	99
C5ORF42	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	78	100	96
C8ORF37	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500	614477	73	100	98
CA4	Retinitis pigmentosa 17, 600852	114760	76	100	100
CABP4	Cone-rod synaptic disorder, congenital nonprogressive, 610427	608965	73	100	99
CACNA1F	Aland Island eye disease, 300600 Cone-rod dystrophy, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, 300071	300110	41	99	90
CACNA2D4	Retinal cone dystrophy 4, 610478	608171	81	100	98
CAPN5	Vitreoretinopathy, neovascular inflammatory, 193235	602537	64	100	92

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CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	82	100	99
CDH23	Deafness 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067	605516	119	100	100
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553	114021	94	100	100
CDHR1	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660	609502	103	98	98
CEP164	Nephronophthisis 15, 614845	614848	68	100	98
CEP250	No OMIM phenotype	609689	39	96	80
CEP290	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189	610142	55	98	92
CEP41	Joubert syndrome 15, 614464	610523	64	100	93
CEP78	Cone-rod dystrophy and hearing loss, 617236	617110	45	95	84
CERKL	Retinitis pigmentosa 26, 608380	608381	73	100	97
CFH	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698	134370	80	99	96
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	80	100	98
CHM	Choroideremia, 303100	300390	48	100	94
CHMP4B	Cataract 31, multiple types, 605387	610897	65	99	91
CHST6	Macular corneal dystrophy, 217800	605294	141	100	100
CIB2	Deafness 48, 609439 Usher syndrome, type IJ, 614869	605564	109	100	100
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	79	100	97
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	68	100	100
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	73	100	94
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	72	100	91
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	100	100	100

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CLRN1	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902	606397	105	100	100
CLUAP1	No OMIM phenotype	616787	44	98	81
CNGA1	Retinitis pigmentosa 49, 613756	123825	62	91	91
CNGA3	Achromatopsia 2, 216900	600053	98	100	100
CNGB1	Retinitis pigmentosa 45, 613767	600724	81	100	98
CNGB3	Achromatopsia 3, 262300 Macular degeneration, juvenile, 248200	605080	80	100	96
CNNM4	Jalili syndrome, 217080	607805	129	99	98
COL11A1	Fibrochondrogenesis 1, 228520 {Lumbar disc herniation, susceptibility to}, 603932 Marshall syndrome, 154780 Stickler syndrome, type II, 604841	120280	56	98	88
COL11A2	Deafness 13, 601868 Deafness 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, 215150 Stickler syndrome, type III, 184840 Weissenbacher-Zweymuller syndrome, 277610	120290	64	100	97
COL18A1	Knobloch syndrome, type 1, 267750	120328	71	100	95
COL2A1	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	120140	74	100	99
COL8A2	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140	120252	43	95	82
COL9A1	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134	120210	71	99	95

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COL9A2	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284	120260	50	100	94
CRB1	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105	604210	84	100	100
CRX	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829	602225	104	100	100
CRYAA	Cataract 9, multiple types, 604219	123580	126	100	100
CRYAB	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869	123590	77	100	100
CRYBA1	Cataract 10, multiple types, 600881	123610	93	100	98
CRYBA2	?Cataract 42, 115900	600836	65	100	100
CRYBA4	Cataract 23, 610425	123631	67	100	93
CRYBB1	Cataract 17, multiple types, 611544	600929	87	100	100
CRYBB2	Cataract 3, multiple types, 601547	123620	92	100	100
CRYBB3	Cataract 22, 609741	123630	92	100	100
CRYGB	Cataract 39, multiple types, 615188	123670	82	100	100
CRYGC	Cataract 2, multiple types, 604307	123680	85	100	100
CRYGD	Cataract 4, multiple types, 115700	123690	63	100	100
CRYGS	Cataract 20, multiple types, 116100	123730	74	100	93
CSPP1	Joubert syndrome 21, 615636	611654	38	96	81
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	74	95	88
CTNNA1	Macular dystrophy, patterned, 2, 608970	116805	46	97	87
CTNND1	No OMIM phenotype	601045	49	99	92
CWC27	Retinitis pigmentosa with or without skeletal anomalies, 250410	617170	25	85	49
CYP1B1	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300	601771	73	100	100
CYP4V2	Bietti crystalline corneoretinal dystrophy, 210370	608614	89	100	100
DCN	Corneal dystrophy, congenital stromal, 610048	125255	52	96	87
DFNB31	Retinitis pigmentosa 59, 613861	608172	98	100	100
DHDDS	Retinitis pigmentosa 59, 613861	608172	73	100	100
DHX38	No OMIM phenotype	605584	43	96	86
DKC1	Dyskeratosis congenita, 305000	300126	57	100	94
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	79	98	95
DRAM2	Cone-rod dystrophy 21, 616502	613360	45	100	93
DTHD1	No OMIM phenotype	616979	45	98	90

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DTNBP1	Hermansky-Pudlak syndrome 7, 614076	607145	73	100	97
EFEMP1	Doyne honeycomb degeneration of retina, 126600	601548	108	100	100
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 ?Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	57	100	98
ELP4	?Aniridia 2, 617141	606985	48	89	78
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875	616846	45	99	88
EPG5	Vici syndrome, 242840	615068	42	96	83
EPHA2	Cataract 6, multiple types, 116600	176946	83	100	99
EXOSC2	No OMIM phenotype	602238	41	100	86
EYA1	Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 ?Otofaciocervical syndrome, 166780	601653	78	100	96
EYS	Retinitis pigmentosa 25, 602772	612424	76	100	98
FA2H	Spastic paraplegia 35, 612319	611026	55	98	87
FAM161A	Retinitis pigmentosa 28, 606068	613596	70	100	100
FBN1	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328	134797	86	100	100
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	82	100	97
FOXC1	Anterior segment dysgenesis 3, multiple subtypes, 601631 Axenfeld-Rieger syndrome, type 3, 602482	601090	46	99	75
FOXE3	Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 Cataract 34, multiple types, 612968	601094	40	73	58
FOXL2	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996	605597	70	97	90
FRAS1	Fraser syndrome, 219000	607830	91	100	100
FREM1	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485	608944	88	100	99
FREM2	Fraser syndrome, 219000	608945	92	100	100

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FRMD7	Nystagmus 1, congenital, 310700 Nystagmus, infantile periodic alternating, 310700	300628	42	100	96
FSCN2	Retinitis pigmentosa 30, 607921	607643	82	100	100
FTL	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159	134790	62	94	76
FYCO1	Cataract 18, 610019	607182	85	100	100
FZD4	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780	604579	93	100	100
GALK1	Galactokinase deficiency with cataracts, 230200	604313	76	100	100
GALT	Galactosemia, 230400	606999	120	100	100
GCNT2	Adult i phenotype without cataract, 110800 [Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 116700	600429	105	100	100
GDF3	Klippel-Feil syndrome 3, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704	606522	101	100	100
GDF6	Klippel-Feil syndrome 1, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094	601147	86	100	100
GFER	Myopathy progressive, with congenital cataract, hearing loss, and developmental delay, 613076	600924	58	100	86
GJA1	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, 218400 Erythrokeratodermia variabilis et progressiva, 133200 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100	121014	66	100	100
GJA3	Cataract 14, multiple types, 601885	121015	95	100	97
GJA8	Cataract 1, multiple types, 116200	600897	97	100	100
GLI2	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829	165230	82	99	97
GLIS2	Nephronophthisis 7, 611498	608539	74	100	96
GNAT1	Night blindness, congenital stationary 3, 610444 ?Night blindness, congenital stationary, type 1G, 616389	139330	96	100	100
GNAT2	Achromatopsia 4, 613856	139340	100	100	98
GNPTG	Mucolipidosis III gamma, 252605	607838	87	95	88

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GPR143	Nystagmus 6, congenital, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500	300808	36	99	75
GPR179	Night blindness, congenital stationary (complete), 1E, 614565	614515	78	100	100
GRASP	No OMIM phenotype	612027	No coverage data		
GRK1	Oguchi disease-2, 613411	180381	95	100	100
GRM6	Night blindness, congenital stationary (complete), 1B, 257270	604096	73	92	81
GRN	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485	138945	95	100	100
GSN	Amyloidosis, Finnish type, 105120	137350	82	99	94
GUCA1A	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093	600364	135	100	100
GUCA1B	Retinitis pigmentosa 48, 613827	602275	89	100	100
GUCY2D	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000	600179	64	100	93
HARS	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504	142810	86	100	99
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801	300056	53	100	99
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	69	94	91
HK1	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460	142600	91	100	98
HMGB3	?Microphthalmia, syndromic 13, 300915	300193	12	49	21
HMX1	Oculoauricular syndrome, 612109	142992	37	77	59
HPS1	Hermansky-Pudlak syndrome 1, 203300	604982	85	100	100
HPS3	Hermansky-Pudlak syndrome 3, 614072	606118	89	100	100
HPS4	Hermansky-Pudlak syndrome 4, 614073	606682	82	100	100
HPS5	Hermansky-Pudlak syndrome 5, 614074	607521	82	100	99
HPS6	Hermansky-Pudlak syndrome 6, 614075	607522	65	99	89
HSF4	Cataract 5, multiple types, 116800	602438	66	99	95
IDH3A	No OMIM phenotype	601149	53	99	88
IDH3B	Retinitis pigmentosa 46, 612572	604526	80	100	100
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920	614620	82	100	99
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	77	100	99
IFT27	?Bardet-Biedl syndrome 19, 615996	615870	45	100	83
IFT74	?Bardet-Biedl syndrome 20, 617119	608040	28	83	49
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472	300139	56	100	100

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IMPDH1	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105	146690	72	97	92
IMPG1	Macular dystrophy, vitelliform, 4, 616151	602870	37	96	79
IMPG2	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581	607056	89	100	100
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	71	98	92
INVS	Nephronophthisis 2, infantile, 602088	243305	82	100	99
IQCB1	Senior-Loken syndrome 5, 609254	609237	65	99	89
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	102	100	100
KCNJ13	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230	603208	88	100	100
KCNV2	Retinal cone dystrophy 3B, 610356	607604	74	100	100
KERA	Cornea plana 2, 217300	603288	91	100	100
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	53	99	95
KIF21A	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700	608283	79	100	96
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	64	98	93
KIZ	Retinitis pigmentosa 69, 615780	615757	48	96	84
KLHL7	Cold-induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943	611119	78	100	99
KRT12	Meesmann corneal dystrophy, 122100	601687	67	100	97
KRT3	Meesmann corneal dystrophy, 122100	148043	73	100	100
LAMA1	Poretti-Boltshauser syndrome, 615960	150320	88	100	99
LCA5	Leber congenital amaurosis 5, 604537	611408	70	100	97
LEMD2	Cataract 46, juvenile-onset, 212500	616312	43	97	84
LIM2	Cataract 19, multiple types, 615277	154045	66	100	100
LRAT	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341	604863	137	100	100
LRIT3	Night blindness, congenital stationary (complete), 1F, 615058	615004	67	100	100
LRMDA	Albinism, oculocutaneous, type VII, 615179	614537	95	100	95
LRP2	Donnai-Barrow syndrome, 222448	600073	89	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
LRP5	[Bone mineral density variability 1], 601884 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 {Osteoporosis}, 166710 Osteosclerosis, 144750 van Buchem disease, type 2, 607636	603506	112	99	98
LSS	Cataract 44, 616509	600909	52	99	93
LTBP2	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	602091	72	100	99
LYST	Chediak-Higashi syndrome, 214500	606897	77	99	97
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	87	100	92
MAB21L2	Microphthalmia, syndromic 14, 615877	604357	96	100	100
MAF	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202	177075	53	81	74
MAK	Retinitis pigmentosa 62, 614181	154235	97	100	95
MAPKAPK3	?Macular dystrophy, patterned, 3, 617111	602130	34	98	80
MERTK	Retinitis pigmentosa 38, 613862	604705	86	100	100
MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152	608507	84	100	100
MFRP	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549	606227	74	100	100
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	75	100	97
MIP	Cataract 15, multiple types, 615274	154050	54	100	93
MIR184	EDICT syndrome, 614303	613146	No coverage data		
MIR204	?Retinal dystrophy and iris coloboma with or without cataract, 616722	610942	No coverage data		
MITF	COMMAD syndrome, 617306 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470	156845	54	100	98
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	106	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	73	100	98
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	78	100	96
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	94	100	100
MYO7A	Deafness 11, 601317 Deafness 2, 600060 Usher syndrome, type 1B, 276900	276903	100	100	98
MYOC	Glaucoma 1A, primary open angle, 137750	601652	101	100	100
NAA10	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855 dominant	300013	51	100	100
NDP	Exudative vitreoretinopathy 2, 305390 Norrie disease, 310600	300658	68	100	100
NEK2	?Retinitis pigmentosa 67, 615565	604043	32	91	65
NEK8	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415	609799	94	100	100
NEUROD1	{Diabetes mellitus, noninsulin-dependent}, 125853 Maturity-onset diabetes of the young 6, 606394	601724	66	100	100
NHS	Cataract 40, 302200 Nance-Horan syndrome, 302350	300457	48	95	92
NMNAT1	Leber congenital amaurosis 9, 608553	608700	119	100	100
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	78	100	99
NPHP3	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540	608002	77	100	100
NPHP4	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996	607215	87	100	100
NR2E3	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131	604485	65	100	100
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	123	100	100
NRL	Retinal degeneration, clumped pigment type Retinitis pigmentosa 27, 613750	162080	54	100	100
NYX	Night blindness, congenital stationary (complete), 1A, 310500	300278	43	100	97
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	72	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
OCA2	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	611409	75	100	98
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	45	99	93
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	43	99	88
OPA1	Behr syndrome, 210000 {Glaucoma, normal tension, susceptibility to}, 606657 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250	605290	69	100	93
OPA3	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300	606580	72	100	90
OPN1LW	Blue cone monochromacy, 303700 Colorblindness, protan, 303900	300822	27	59	46
OPN1MW	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800	300821	22	65	50
OPN1SW	Colorblindness, tritan, 190900	613522	89	100	100
OPTN	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657	602432	69	100	93
OR2W3	No OMIM phenotype	616729	76	100	98
OTX2	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125	600037	88	100	100
P3H2	Myopia, high, with cataract and vitreoretinal degeneration, 614292	610341	79	100	100
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	90	100	100
PAX2	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330	167409	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PAX6	Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 ?Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550	607108	62	100	99
PCDH15	Deafness 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083	605514	89	100	98
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940	123695	40	96	78
PDE6A	Retinitis pigmentosa 43, 613810	180071	75	100	99
PDE6B	Night blindness, congenital stationary 2, 163500 Retinitis pigmentosa-40, 613801	180072	101	100	100
PDE6C	Cone dystrophy 4, 613093	600827	75	100	99
PDE6G	Retinitis pigmentosa 57, 613582	180073	88	100	100
PDE6H	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024	601190	48	100	100
PDZD7	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472	612971	60	99	96
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	67	100	98
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	71	100	100
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	68	100	89
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	43	100	93
PHYH	Refsum disease, 266500	602026	57	99	85
PIGL	CHIME syndrome, 280000	605947	74	100	94
PIK3R1	?Agammaglobulinemia 7, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880	171833	81	100	100
PIKFYVE	Corneal fleck dystrophy, 121850	609414	88	100	98
PITPNM3	Cone-rod dystrophy 5, 600977	608921	75	99	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PITX2	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550	601542	74	98	91
PITX3	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623	602669	60	100	100
PLA2G5	[Fleck retina, familial benign], 228980	601192	105	100	100
PLK4	Microcephaly and chorioretinopathy, 2, 616171	605031	53	92	69
PNPLA6	Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, 612020	603197	72	100	98
POC1B	Cone-rod dystrophy 20, 615973	614784	30	93	72
POMGNT1	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	606822	82	100	95
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	81	100	100
PRCD	Retinitis pigmentosa 36, 610599	610598	51	100	100
PRDM13	No OMIM phenotype	616741	65	96	88
PRDM5	Brittle cornea syndrome 2, 614170	614161	74	100	99
PROM1	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786	604365	72	100	99
PRPF3	Retinitis pigmentosa 18, 601414	607301	81	100	100
PRPF31	Retinitis pigmentosa 11, 600138	606419	73	100	99
PRPF4	Retinitis pigmentosa 70, 615922	607795	47	98	90
PRPF6	Retinitis pigmentosa 60, 613983	613979	91	100	100
PRPF8	Retinitis pigmentosa 13, 600059	607300	89	100	99
PRPH2	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	179605	87	100	100
PRSS56	Microphthalmia, isolated 6, 613517	613858	52	100	99
PXDN	Anterior segment dysgenesis 7, with sclerocornea, 269400	605158	87	100	98
RAB28	Cone-rod dystrophy 18, 615374	612994	20	84	39

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
RARB	Microphthalmia, syndromic 12, 615524	180220	44	100	94
RAX	Microphthalmia, isolated 3, 611038	601881	77	96	80
RAX2	Cone-rod dystrophy 11, 610381 ?Macular degeneration, age-related, 6, 613757	610362	48	100	100
RBP3	?Retinitis pigmentosa 66, 615233	180290	82	100	100
RBP4	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147	180250	87	100	100
RCBTB1	Retinal dystrophy with or without extraocular anomalies, 617175	607867	88	100	100
RD3	Leber congenital amaurosis 12, 610612	180040	100	100	100
RDH11	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108	607849	37	97	83
RDH12	Leber congenital amaurosis 13, 612712	608830	80	100	100
RDH5	Fundus albipunctatus, 136880	601617	98	100	100
REEP6	Retinitis pigmentosa 77, 617304	609346	83	99	93
RGR	Retinitis pigmentosa 44, 613769	600342	94	100	99
RGS9	Bradyopsia, 608415	604067	67	100	99
RGS9BP	Bradyopsia, 608415	607814	58	100	100
RHO	Night blindness, congenital stationary 1, 610445 Retinitis pigmentosa 4 or recessive, 613731 Retinitis punctata albescens, 136880	180380	117	100	100
RIMS1	Cone-rod dystrophy 7, 603649	606629	73	100	96
RLBP1	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880	180090	90	100	98
ROM1	Retinitis pigmentosa 7, digenic, 608133	180721	65	100	100
RP1	Retinitis pigmentosa 1, 180100	603937	61	100	96
RP1L1	Occult macular dystrophy, 613587	608581	81	100	100
RP2	Retinitis pigmentosa 2, 312600	300757	47	100	96
RP9	?Retinitis pigmentosa 9, 180104	607331	63	99	92
RPE65	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794	180069	77	100	97
RPGR	Cone-rod dystrophy, 1, 304020 Macular degeneration atrophic, 300834 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, and sinorespiratory infections, with or without deafness, 300455	312610	30	71	64
RPGRIP1	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826	605446	86	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	77	97	95
RS1	Retinoschisis, 312700	300839	40	100	94
RTN4IP1	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732	610502	32	93	74
SAG	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758	181031	79	100	100
SALL2	?Coloboma, ocular, 216820	602219	59	100	99
SDCCAG8	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615	613524	70	100	99
SEMA4A	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282	607292	91	100	100
SHH	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250	600725	80	100	97
SIPA1L3	?Cataract 45, 616851	616655	68	98	91
SIX3	Holoprosencephaly 2, 157170 Schizencephaly, 269160	603714	103	100	100
SIX6	Optic disc anomalies with retinal and/or macular dystrophy, 212550	606326	108	100	100
SLC16A12	Cataract 47, juvenile, with microcornea, 612018	611910	72	100	99
SLC24A1	Night blindness, congenital stationary (complete), 1D, 613830	603617	85	100	100
SLC24A5	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750	609802	56	100	100
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, 612539	603690	62	100	96
SLC38A8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218	615585	29	94	75
SLC45A2	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	606202	78	100	99
SLC4A11	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal endothelial dystrophy, 217700	610206	79	100	97
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707	607882	103	100	100
SLC7A14	Retinitis pigmentosa 68, 615725	615720	55	100	96
SMOC1	Microphthalmia with limb anomalies, 206920	608488	77	100	100
SNRNP200	Retinitis pigmentosa 33, 610359	601664	105	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SOX2	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900	184429	113	100	100
SPATA7	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, 604232	609868	71	100	96
SPP2	No OMIM phenotype	602637	48	100	97
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	105	100	100
STRA6	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186	610745	58	100	100
TACSTD2	Corneal dystrophy, gelatinous drop-like, 204870	137290	107	100	100
TCOF1	Treacher Collins syndrome 1, 154500	606847	78	100	98
TCTN1	Joubert syndrome 13, 614173	609863	70	100	97
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	60	100	96
TDRD7	Cataract 36, 613887	611258	91	100	99
TEAD1	Sveinsson chorioretinal atrophy, 108985	189967	74	100	100
TENM3	Microphthalmia, isolated, with coloboma 9, 615145	610083	62	98	94
TFAP2A	Branchiooculofacial syndrome, 113620	107580	66	100	96
TGFBI	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471	601692	83	99	93
TGIF1	Holoprosencephaly 4, 142946	602630	97	100	100
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	42	100	100
TIMP3	Sorsby fundus dystrophy, 136900	188826	88	100	100
TMEM126A	Optic atrophy 7, 612989	612988	70	100	96
TMEM138	Joubert syndrome 16, 614465	614459	67	100	100
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	56	98	93
TMEM237	Joubert syndrome 14, 614424	614423	80	100	99
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550	609884	51	99	90
TOPORS	Retinitis pigmentosa 31, 609923	609507	76	100	100

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TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	79	100	98
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 {Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315	606609	129	100	100
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110	602290	77	100	100
TRNT1	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084	612907	36	90	67
TRPM1	Night blindness, congenital stationary (complete), 1C, 613216	603576	87	100	100
TSPAN12	Exudative vitreoretinopathy 5, 613310	613138	85	100	100
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	64	100	90
TTLL5	Cone-rod dystrophy 19, 615860	612268	50	97	89
TUB	?Retinal dystrophy and obesity, 616188	601197	73	100	100
TUBGCP4	Microcephaly and chorioretinopathy, 3, 616335	609610	42	96	82
TULP1	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132	602280	65	100	97
TYR	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 Waardenburg syndrome/albinism, digenic, 103470	606933	71	100	100
TYRP1	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271	115501	107	100	100
UBIAD1	Corneal dystrophy, Schnyder type, 121800	611632	83	100	100
UNC119	?Cone-rod dystrophy ?Immunodeficiency 13, 615518	604011	53	100	94
UNC45B	?Cataract 43, 616279	611220	46	97	86
USH1C	Deafness 18A, 602092 Usher syndrome, type 1C, 276904	605242	70	100	91
USH1G	Usher syndrome, type 1G, 606943	607696	86	100	99
USH2A	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901	608400	90	100	100
VAX1	?Microphthalmia, syndromic 11, 614402	604294	54	90	84
VCAN	Wagner syndrome 1, 143200	118661	78	100	100
VIM	Cataract 30, pulverulent, 116300	193060	96	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
VSX1	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300	605020	62	89	78
VSX2	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093	142993	74	100	100
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, 603563	610657	96	100	99
WDPCP	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	613580	67	100	93
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	73	100	98
WFS1	?Cataract 41, 116400 Deafness 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome, 222300 Wolfram-like syndrome, 614296	606201	102	100	100
WHRN	Deafness 31, 607084 Usher syndrome, type 2D, 611383	607928	68	100	98
WRN	Werner syndrome, 277700	604611	71	100	92
YAP1	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433	606608	40	86	73
YME1L1	?Optic atrophy 11, 617302	607472	36	90	71
ZEB2	Mowat-Wilson syndrome, 235730	605802	81	100	100
ZIC2	Holoprosencephaly 5, 609637	603073	92	93	88
ZNF408	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469	616454	70	100	99
ZNF423	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844	604557	102	100	100
ZNF469	Brittle cornea syndrome 1, 229200	612078	69	100	97
ZNF513	?Retinitis pigmentosa 58, 613617	613598	66	100	100
ZNF644	Myopia 21, 614167	614159	67	100	99

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping.
- The statistics above are based on a set of 50 samples
- Median depth is the median of the mean sequence depth over the protein coding exons of the transcript
- % Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x

- % 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