Whole Exome Sequencing
Gene package Vision disorders, version 2, 23-9-2016

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	Stargardt disease 1, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 {Macular degeneration, age-related, 2}, 153800 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200	601691	86	100	99
ABCB6	Microphthalmia, isolated, with coloboma 7, 614497 [Blood group, Langereis system], 111600 Dyschromatosis universalis hereditaria 3, 615402 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153	605452	79	100	100
ABCC6	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473	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
ACTB	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310	102630	77	100	100
ACTG1	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583	102560	104	100	100
ADAM9	Cone-rod dystrophy 9, 612775	602713	78	100	100
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458	607512	83	100	98

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ADIPOR1	No OMIM phenotype	607945	41	88	73
AGBL1	Corneal dystrophy, Fuchs endothelial, 8, 615523	615496	79	100	99
AGK	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691	610345	66	99	95
AHI1	Joubert syndrome-3, 608629	608894	43	99	91
AIPL1	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393	604392	84	97	90
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	77	100	98
ALMS1	Alstrom syndrome, 203800	606844	87	100	100
ALX3	Frontonasal dysplasia 1, 136760	606014	45	98	86
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	105	100	100
ARL13B	Joubert syndrome 8, 612291	608922	65	87	80
ARL2BP	Retinitis pigmentosa with or without situs inversus, 615434	615407	73	100	95
ARL6	Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900 ?Retinitis pigmentosa 55, 613575	608845	61	100	100
ASPH	Traboulsi syndrome, 601552	600582	28	80	63
ATF6	Achromatopsia 7, 616517	605537	48	95	81
ATOH7	Persistent hyperplastic primary vitreous, autosomal recessive, 221900	609875	39	93	70
BBIP1	?Bardet-Biedl syndrome 18, 615995	613605	77	100	99
BBS1	Bardet-Biedl syndrome 1, 209900	209901	67	100	92
BBS10	Bardet-Biedl syndrome 10, 615987	610148	53	100	89
BBS12	Bardet-Biedl syndrome 12, 615989	610683	92	100	100
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	76	100	100
BBS4	Bardet-Biedl syndrome 4, 615982	600374	98	100	100
BBS5	Bardet-Biedl syndrome 5, 615983	603650	76	100	98
BBS7	Bardet-Biedl syndrome 7, 615984	607590	80	100	99
BBS9	Bardet-Biedl syndrome 9, 615986	607968	59	99	92
BCOR	Microphthalmia, syndromic 2, 300166	300485	68	100	98
BEST1	Macular dystrophy, vitelliform, 2, 153700 Bestrophinopathy, autosomal recessive, 611809 Vitreoretinochoroidopathy, 193220 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa-50, 613194 Retinitis pigmentosa, concentric, 613194	607854	76	98	93
BFSP1	Cataract 33, 611391	603307	52	99	94
BFSP2	Cataract 12, multiple types, 611597	603212	81	100	98

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BLOC1S3	Hermansky-Pudlak syndrome 8, 614077	609762	69	100	98
BLOC1S6	Hermansky-pudlak syndrome 9, 614171	604310	72	100	100
BMP4	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625	112262	48	100	100
BMP7	No OMIM phenotype	112267	67	100	100
C10orf11	Albinism, oculocutaneous, type VII, 615179	614537	77	100	100
C12orf57	Temtamy syndrome, 218340	615140	76	100	100
C19ORF12	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043	614297	95	100	95
C1QTNF5	Retinal degeneration, late-onset, autosomal dominant, 605670	608752	85	100	100
C21orf2	No OMIM phenotype	603191	71	100	100
C2ORF71	Retinitis pigmentosa 54, 613428	613425	68	100	96
C5ORF42	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	52	98	92
C8ORF37	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500	614477	67	100	99
CA4	Retinitis pigmentosa 17, 600852	114760	78	100	96
CABP4	Cone-rod synaptic disorder, congenital nonprogressive, 610427	608965	73	100	98
CACNA1F	Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Cone-rod dystrophy, X-linked, 3, 300476 Aland Island eye disease, 300600	300110	76	100	100
CACNA2D4	Retinal cone dystrophy 4, 610478	608171	73	100	99
CAPN5	Vitreoretinopathy, neovascular inflammatory, 193235	602537	41	99	90
CC2D2A	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360	612013	81	100	98
CDH23	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067	605516	64	100	92
CDH3	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280	114021	82	100	99
CDHR1	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660	609502	119	100	100
CEP164	Nephronophthisis 15, 614845	614848	94	100	100
CEP250	No OMIM phenotype	609689	103	98	98

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CEP290	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 ?Bardet-Biedl syndrome 14, 615991	610142	68	100	98
CERKL	Retinitis pigmentosa 26, 608380	608381	39	96	80
CFH	{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 Complement factor H deficiency, 609814 {Macular degeneration, age-related, 4}, 610698 Basal laminar drusen, 126700	134370	55	98	92
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	73	100	97
CHM	Choroideremia, 303100	300390	80	99	96
CHMP4B	Cataract 31, multiple types, 605387	610897	80	100	98
CHST6	Macular corneal dystrophy, 217800	605294	48	100	94
CIB2	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869	605564	65	99	91
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	141	100	100
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	109	100	100
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	79	100	97
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	68	100	100
CLRN1	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180	606397	73	100	94
CNGA1	Retinitis pigmentosa 49, 613756	123825	72	100	91
CNGA3	Achromatopsia-2, 216900	600053	105	100	100
CNGB1	Retinitis pigmentosa 45, 613767	600724	62	91	91
CNGB3	Achromatopsia-3, 262300 Macular degeneration, juvenile, 248200	605080	98	100	100
CNNM4	Jalili syndrome, 217080	607805	81	100	98
COL11A1	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis 1, 228520	120280	80	100	96

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COL11A2	Stickler syndrome, type III, 184840 Otospondylomegaepiphyseal dysplasia, 215150 Weissenbacher-Zweymuller syndrome, 277610 Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524	120290	129	99	98
COL18A1	Knobloch syndrome, type 1, 267750	120328	56	98	88
COL2A1	Stickler syndrome, type I, 108300 Kniest dysplasia, 156550 Achondrogenesis, type II or hypochondrogenesis, 200610 SED congenita, 183900 SMED Strudwick type, 184250 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Spondyloperipheral dysplasia, 271700 Osteoarthritis with mild chondrodysplasia, 604864 Vitreoretinopathy with phalangeal epiphyseal dysplasia Platyspondylic skeletal dysplasia, Torrance type, 151210 Otospondylomegaepiphyseal dysplasia, 215150 Avascular necrosis of the femoral head, 608805 Legg-Calve-Perthes disease, 150600 Stickler sydrome, type I, nonsyndromic ocular, 609508 Czech dysplasia, 609162 Spondyloepiphyseal dysplasia, Stanescu type, 616583	120140	64	100	97
COL8A2	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140	120252	71	100	95
COL9A1	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134	120210	74	100	99
CRB1	Retinitis pigmentosa-12, autosomal recessive, 600105 Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870	604210	43	95	82
CRX	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829	602225	71	99	95
CRYAA	Cataract 9, multiple types, 604219	123580	84	100	100
CRYAB	Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869 Cardiomyopathy, dilated, 1II, 615184	123590	104	100	100
CRYBA1	Cataract 10, multiple types, 600881	123610	126	100	100
CRYBA4	Cataract 23, 610425	123631	77	100	100

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CRYBB1	Cataract 17, multiple types, 611544	600929	93	100	98
CRYBB2	Cataract 3, multiple types, 601547	123620	67	100	93
CRYBB3	Cataract 22, autosomal recessive, 609741	123630	87	100	100
CRYGB	Cataract 39, multiple types, autosomal dominant, 615188	123670	92	100	100
CRYGC	Cataract 2, multiple types, 604307	123680	92	100	100
CRYGD	Cataract 4, multiple types, 115700	123690	82	100	100
CRYGS	Cataract 20, multiple types, 116100	123730	85	100	100
CSPP	Joubert syndrome 21, 615636	611654	63	100	100
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	74	100	93
CTNNA1	No OMIM phenotype	116805	38	96	81
CYP1B1	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Peters anomaly, 604229	601771	74	95	88
CYP4V2	Bietti crystalline corneoretinal dystrophy, 210370	608614	46	97	87
DHDDS	Retinitis pigmentosa 59, 613861	608172	73	100	100
DHX38	No OMIM phenotype	605584	89	100	100
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	68	100	98
DRAM2	Cone-rod dystrophy 21, 616502	613360	73	100	100
DTHD1	No OMIM phenotype	616979	43	96	86
DTNBP1	{Schizophrenia}, 181500 Hermansky-Pudlak syndrome 7, 614076	607145	79	98	95
EFEMP1	Doyne honeycomb degeneration of retina, 126600	601548	45	100	93
ELOVL4	Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457 ?Spinocerebellar ataxia 34, 133190	605512	45	98	90
ELP4	?Aniridia, 106210	606985	73	100	97
EMC1	No OMIM phenotype	616846	108	100	100
EPG5	Vici syndrome, 242840	615068	57	100	98
EPHA2	Cataract 6, multiple types, 116600	176946	48	89	78
EYA1	Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 ?Otofaciocervical syndrome, 166780	601653	45	99	88
EYS	Retinitis pigmentosa 25, 602772	612424	42	96	83
FA2H	Spastic paraplegia 35, autosomal recessive, 612319	611026	83	100	99
FADD	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759	602457	78	100	96
FAM161A	Retinitis pigmentosa 28, 606068	613596	76	100	98
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	55	98	87

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FOXE3	Anterior segment mesenchymal dysgenesis, 107250 Aphakia, congenital primary, 610256	601094	99	100	100
FREM1	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485	608944	70	100	100
FRMD7	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700	300628	82	100	97
FSCN2	Retinitis pigmentosa 30, 607921	607643	40	73	58
FTL	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604	134790	88	100	99
FYCO1	Cataract 18, autosomal recessive, 610019	607182	42	100	96
FZD4	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780	604579	82	100	100
GALK1	Galactokinase deficiency with cataracts, 230200	604313	62	94	76
GALT	Galactosemia, 230400	606999	85	100	100
GCNT2	[Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 116700 Adult i phenotype without cataract, 110800	600429	93	100	100
GDF3	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704	606522	76	100	100
GDF6	Klippel-Feil syndrome 1, autosomal dominant, 118100 Microphthalmia, isolated 4, 613094 Microphthalmia with coloboma 6, digenic, 613703 Leber congenital amaurosis 17, 615360	601147	120	100	100
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076	600924	105	100	100
GJA1	Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Hypoplastic left heart syndrome 1, 241550 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, autosomal recessive, 257850 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva, 133200 Palmoplantar keratoderma with congenital alopecia, 104100	121014	101	100	100
GJA3	Cataract 14, multiple types, 601885	121015	86	100	100
GJA8	Cataract 1, multiple types, 116200	600897	58	100	86
GLIS2	Nephronophthisis 7, 611498	608539	66	100	100

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GNAT1	Night blindness, congenital stationary, autosomal dominant 3, 610444 ?Night blindness, congenital stationary, type 1G, 616389	139330	95	100	97
GNAT2	Achromatopsia-4, 613856	139340	97	100	100
GNPTG	Mucolipidosis III gamma, 252605	607838	74	100	96
GPR125	No OMIM phenotype	612303	96	100	100
GPR143	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814	300808	100	100	98
GPR179	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565	614515	87	95	88
GPR98	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472	602851	36	99	75
GRCC10	No OMIM phenotype	0	78	100	100
GRK1	Oguchi disease-2, 613411	180381	95	100	100
GRM6	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270	604096	73	92	81
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	Leber congenital amaurosis 1, 204000 Cone-rod dystrophy 6, 601777	600179	64	100	93
HARS	Usher syndrome type 3B, 614504 Charcot-Marie-Tooth disease, axonal, type 2W, 616625	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
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
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
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	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 Retinitis pigmentosa 71, 616394	607386	77	100	99
IFT27	?Bardet-Biedl syndrome 19, 615996	615870	45	100	83
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472	300139	56	100	100

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IMPDH1	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837	146690	72	97	92
IMPG1	Macular dystrophy, vitelliform, 4, 616151	602870	37	96	79
IMPG2	Retinitis pigmentosa 56, 613581 Macular dystrophy, vitelliform, 5, 616152	607056	89	100	100
INPP5E	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300	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	Snowflake vitreoretinal degeneration, 193230 Leber congenital amaurosis 16, 614186	603208	88	100	100
KCNV2	Retinal cone dystrophy 3B, 610356	607604	74	100	100
KERA	Cornea plana congenita, recessive, 217300	603288	91	100	100
KIAA0196	Spastic paraplegia 8, autosomal dominant, 603563 Ritscher-Schinzel syndrome 1, 220210	610657	96	100	99
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	53	99	95
KIF7	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131	611254	64	98	93
KIZ	Retinitis pigmentosa 69, 615780	615757	48	96	84
KLHL7	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
LIM2	Cataract 19, multiple types, 615277	154045	66	100	100
LRAT	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341	604863	137	100	100
LRIT3	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058	615004	67	100	100
LRP2	Donnai-Barrow syndrome, 222448	600073	89	100	100

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LRP5	Osteoporosis-pseudoglioma syndrome, 259770 [Bone mineral density variability 1], 601884 Hyperostosis, endosteal, 144750 van Buchem disease, type 2, 607636 Osteosclerosis, 144750 {Osteoporosis}, 166710 Exudative vitreoretinopathy 4, 601813 Osteopetrosis, autosomal dominant 1, 607634	603506	112	99	98
LSS	Cataract 44, 616509	600909	52	99	93
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	Cataract 21, multiple types, 610202 Ayme-Gripp syndrome, 601088	177075	53	81	74
MAK	Retinitis pigmentosa 62, 614181	154235	97	100	95
MAPKAPK3	No OMIM phenotype	602130	34	98	80
MERTK	Retinitis pigmentosa 38, 613862	604705	86	100	100
MFN2	Charcot-Marie-Tooth disease, type 2A2, 609260 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	Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456	156845	54	100	98
MKKS	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231	604896	106	100	100
MKS1	Meckel syndrome 1, 249000 Bardet-Biedl syndrome 13, 615990	609883	73	100	98
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	78	100	96
MVK	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900	251170	94	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MYO7A	Usher syndrome, type 1B, 276900 Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317	276903	100	100	98
MYOC	Glaucoma 1A, primary open angle, 137750	601652	101	100	100
NAA10	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800	300013	51	100	100
NDP	Norrie disease, 310600 Exudative vitreoretinopathy 2, X-linked, 305390	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	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200	300457	48	95	92
NMNAT1	Leber congenital amaurosis 9, 608553	608700	119	100	100
NPHP1	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583	607100	78	100	99
NPHP3	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010	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	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type	162080	54	100	100
NYX	Night blindness, congenital stationary (complete), 1A, X-linked, 310500	300278	43	100	97
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	72	100	96
OCA2	Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220	611409	75	100	98
OFD1	Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804 ?Retinitis pigmentosa 23, 300424	300170	43	99	88

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OPA1	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 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	Colorblindness, protan, 303900 Blue cone monochromacy, 303700	300822	27	59	46
OPN1MW	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700	300821	22	65	50
OPN1SW	Colorblindness, tritan, 190900	613522	89	100	100
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	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236	606157	90	100	100
PAX2	Papillorenal syndrome, 120330 Glomerulosclerosis, focal segmental, 7, 616002	167409	100	100	98
PAX6	Aniridia, 106210 Peters anomaly, 604229 Cataract with late-onset corneal dystrophy, 106210 Keratitis, 148190 Foveal hypoplasia 1, 136520 ?Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550 Coloboma, ocular, 120200 Coloboma of optic nerve, 120430 Gillespie syndrome, 206700	607108	62	100	99
PCDH15	Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067	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, autosomal dominant 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

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PDE6H	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 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	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Heimler syndrome 1, 234580	602136	67	100	98
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	71	100	100
PEX7	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879	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
PIKFYVE	Corneal fleck dystrophy, 121850	609414	88	100	98
PITPNM3	Cone-rod dystrophy 5, 600977	608921	75	99	99
PITX2	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Ring dermoid of cornea, 180550 Peters anomaly, 604229	601542	74	98	91
PITX3	Anterior segment mesenchymal dysgenesis, 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, autosomal recessive, 2, 616171	605031	53	92	69
PNPLA6	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400	603197	72	100	98
POC1B	Cone-rod dystrophy 20, 615973	614784	30	93	72
PRCD	Retinitis pigmentosa 36, 610599	610598	51	100	100
PROM1	Retinitis pigmentosa 41, 612095 Cone-rod dystrophy 12, 612657 Stargardt disease 4, 603786 Macular dystrophy, retinal, 2, 608051	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

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PRPF8	Retinitis pigmentosa 13, 600059	607300	89	100	99
PRPH2	Retinitis pigmentosa 7 and digenic, 608133 Leber congenital amaurosis 18, 608133 Macular dystrophy, patterned, 1, 169150 Retinitis punctata albescens, 136880 Choriodal dystrophy, central areolar 2, 613105 Macular dystrophy, vitelliform, 3, 608161	179605	87	100	100
PRSS56	Microphthalmia, isolated 6, 613517	613858	52	100	99
PXDN	Corneal opacification and other ocular anomalies, 269400	605158	87	100	98
RAB28	Cone-rod dystrophy 18, 615374	612994	20	84	39
RARB	Microphthalmia, syndromic 12, 615524	180220	44	100	94
RAX	Microphthalmia, isolated 3, 611038	601881	77	96	80
RAX2	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381	610362	48	100	100
RBP3	?Retinitis pigmentosa 66, 615233	180290	82	100	100
RBP4	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 Microphthalmia, isolated, with coloboma 10, 616428	180250	87	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
RGR	Retinitis pigmentosa 44, 613769	600342	94	100	99
RGS9	Bradyopsia, 608415	604067	67	100	99
RGS9BP	Bradyopsia, 608415	607814	58	100	100
RHO	Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880	180380	117	100	100
RIMS1	Cone-rod dystrophy 7, 603649	606629	73	100	96
RLBP1	Fundus albipunctatus, 136880 Retinitis punctata albescens, 136880 Newfoundland rod-cone dystrophy, 607476 Bothnia retinal dystrophy, 607475	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

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
RPGR	Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834 Cone-rod dystrophy, X-linked, 1, 304020	312610	30	71	64
RPGRIP1	Leber congenital amaurosis 6, 613826 Cone-rod dystrophy 13, 608194	605446	86	100	99
RPGRIP1L	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360	610937	77	97	95
RS1	Retinoschisis, 312700	300839	40	100	94
SAG	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758	181031	79	100	100
SALL2	?Coloboma, ocular, autosomal recessive, 216820	602219	59	100	99
SDCCAG8	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993	613524	70	100	99
SEMA4A	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283	607292	91	100	100
SHH	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160	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
SLC24A1	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830	603617	85	100	100
SLC24A5	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 Albinism, oculocutaneous, type VI, 113750	609802	56	100	100
SLC33A1	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482	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 endothelial dystrophy 2, autosomal recessive, 217700 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal dystrophy, Fuchs endothelial, 4, 613268	610206	79	100	97
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707	607882	103	100	100

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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
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, autosomal recessive, 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, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 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
TDRD7	Cataract 36, 613887	611258	91	100	99
TEAD1	Sveinsson choreoretinal 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, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, epithelial basement membrane, 121820	601692	83	99	93
TGIF1	Holoprosencephaly-4, 142946	602630	97	100	100
TIMM8A	Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150	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

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TMEM67	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Nephronophthisis 11, 613550	609884	51	99	90
TOPORS	Retinitis pigmentosa 31, 609923	609507	76	100	100
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700	606609	129	100	100
TRIM32	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988	602290	77	100	100
TRNT1	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084	612907	36	90	67
TRPM1	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 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, autosomal recessive, 3, 616335	609610	42	96	82
TULP1	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843	602280	65	100	97
TYR	Albinism, oculocutaneous, type IA, 203100 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800	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
USH1C	Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092	605242	70	100	91
USH1G	Usher syndrome, type 1G, 606943	607696	86	100	99
USH2A	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809	608400	90	100	100
VAX1	?Microphthalmia, syndromic 11, 614402	604294	54	90	84

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VCAN	Wagner syndrome 1, 143200	118661	78	100	100
VIM	?Cataract 30, pulverulent, 116300	193060	96	100	100
VSX2	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093	142993	74	100	100
WDPCP	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	613580	67	100	93
WDR19	Nephronophthisis 13, 614377 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Senior-Loken syndrome 8, 616307	608151	73	100	98
WFS1	Wolfram syndrome, 222300 Deafness, autosomal dominant 6/14/38, 600965 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 ?Cataract 41, 116400	606201	102	100	100
WHRN	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383	607928	No coverage data		
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 Coloboma, ocular, 120433	606608	40	86	73
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	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844	604557	102	100	100
ZNF513	Retinitis pigmentosa 58, 613617	613598	66	100	100
ZNF644	Myopia 21, autosomal dominant, 614167	614159	67	100	99

- Gene symbols according HGCN
- OMIM release used: 17-3-2016
- "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
- % Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x