

Whole Exome Sequencing

Gene package Vision disorders, version 6.1, 31-1-2020



Technical information

DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: <http://bio-bwa.sourceforge.net/>). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: <http://www.broadinstitute.org/gatk/>). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. 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).



Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	94	100	100	97
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	110	100	100	100
ABCC6	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850	603234	118	100	100	99
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	613599	61	100	100	94
ACBD5	No OMIM phenotype	616618	85	100	99	89
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	146	100	98	94

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ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	192	100	100	100
ACTG1	Baraitser-Winter syndrome 2, 614583 Deafness 20/26, 604717	102560	188	100	100	100
ADAM9	Cone-rod dystrophy 9, 612775	602713	57	100	99	92
ADAMTS10	Weill-Marchesani syndrome 1, recessive, 277600	608990	112	100	100	100
ADAMTS17	Weill-Marchesani 4 syndrome, recessive, 613195	607511	100	97	95	92
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458	607512	79	100	100	98
ADAMTSL4	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, 225100	610113	115	100	100	99
ADGRV1	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472	602851	80	100	100	97
ADIPOR1	No OMIM phenotype	607945	100	100	100	100
AGBL1	Corneal dystrophy, Fuchs endothelial, 8, 615523	615496	74	98	98	97
AGBL5	Retinitis pigmentosa 75, 617023	615900	90	100	99	96
AGK	Cataract 38, 614691 Sengers syndrome, 212350	610345	57	100	100	93
AHI1	Joubert syndrome 3, 608629	608894	67	100	98	90
AHR	?Retinitis pigmentosa 85, 618345	600253	63	100	100	95
AIPL1	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393	604392	141	100	100	100
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	88	100	100	97
ALMS1	Alstrom syndrome, 203800	606844	100	100	100	99
ALX3	Frontonasal dysplasia 1, 136760	606014	116	98	91	86
ANTXR1	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089	606410	60	99	97	92
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	66	100	98	87
AP3D1	?Hermansky-Pudlak syndrome 10, 617050	607246	103	99	98	97
ARHGEF18	Retinitis pigmentosa 78, 617433	616432	123	100	99	93
ARL13B	Joubert syndrome 8, 612291	608922	63	100	100	94
ARL2BP	Retinitis pigmentosa with or without situs inversus, 615434	615407	57	100	100	96
ARL3	Joubert syndrome 35, 618161 Retinitis pigmentosa 83, 618173	604695	111	100	100	100

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ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	43	96	95	83
ARMC9	Joubert syndrome 30, 617622	617612	76	100	98	93
ARR3	Myopia 26, female-limited, 301010	301770	62	100	100	96
ARSG	Usher syndrome, type IV, 618144	610008	88	100	100	97
ASPH	Traboulsi syndrome, 601552	600582	63	100	99	92
ASRGL1	No OMIM phenotype	609212	72	100	98	93
ATF6	Achromatopsia 7, 616517	605537	65	100	100	97
ATOH7	Persistent hyperplastic primary vitreous, 221900	609875	131	95	88	85
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	144	100	100	100
B3GLCT	Peters-plus syndrome, 261540	610308	67	100	99	85
B9D1	Joubert syndrome 27, 617120 ?Meckel syndrome 9, 614209	614144	90	100	100	100
B9D2	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175	611951	82	100	100	100
BBIP1	?Bardet-Biedl syndrome 18, 615995	613605	45	100	96	86
BBS1	Bardet-Biedl syndrome 1, 209900	209901	126	100	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	66	100	100	100
BBS12	Bardet-Biedl syndrome 12, 615989	610683	59	100	100	99
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	77	100	100	97
BBS4	Bardet-Biedl syndrome 4, 615982	600374	76	100	100	95
BBS5	Bardet-Biedl syndrome 5, 615983	603650	65	100	98	89
BBS7	Bardet-Biedl syndrome 7, 615984	607590	64	100	99	95
BBS9	Bardet-Biedl syndrome 9, 615986	607968	59	96	95	90
BCOR	Microphthalmia, syndromic 2, 300166	300485	78	100	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 Vitreoretinopathy, 193220	607854	94	100	99	98
BFSP1	Cataract 33, multiple types, 611391	603307	98	100	100	96

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BFSP2	Cataract 12, multiple types, 611597	603212	73	100	99	92
BLOC1S3	Hermansky-Pudlak syndrome 8, 614077	609762	104	100	100	100
BLOC1S6	?Hermansky-pudlak syndrome 9, 614171	604310	46	100	100	84
BMP4	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625	112262	103	100	96	93
BMP7	No OMIM phenotype	112267	116	100	100	100
BSG	[Blood group, OK], 111380	109480	107	100	100	99
C12orf57	Temtamy syndrome, 218340	615140	141	100	100	100
C19orf12	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, 615043	614297	186	100	100	100
C1QTNF5	Retinal degeneration, late-onset, 605670	608752	102	100	100	93
C3	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378	120700	127	100	100	99
C8orf37	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500	614477	78	100	100	96
CA4	Retinitis pigmentosa 17, 600852	114760	100	100	100	100
CABP4	Cone-rod synaptic disorder, congenital nonprogressive, 610427	608965	149	100	100	100
CACNA1F	Aland Island eye disease, 300600 Cone-rod dystrophy, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, 300071	300110	76	100	100	96
CACNA2D4	Retinal cone dystrophy 4, 610478	608171	86	100	100	98
CAPN5	Vitreoretinopathy, neovascular inflammatory, 193235	602537	143	100	100	99
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	66	100	100	95
CDH23	Deafness 12, 601386 {Pituitary adenoma 5, multiple types}, 617540 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067	605516	135	100	100	100
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553	114021	124	100	100	100
CDHR1	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660	609502	116	100	100	98

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CEP104	Joubert syndrome 25, 616781	616690	71	100	98	93
CEP120	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300	613446	75	100	100	96
CEP164	Nephronophthisis 15, 614845	614848	93	100	100	98
CEP250	Cone-rod dystrophy and hearing loss 2, 618358	609689	73	100	99	95
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	71	100	98	89
CEP41	Joubert syndrome 15, 614464	610523	69	100	100	94
CEP78	Cone-rod dystrophy and hearing loss, 617236	617110	69	100	100	94
CEP83	Nephronophthisis 18, 615862	615847	60	100	96	81
CERKL	Retinitis pigmentosa 26, 608380	608381	73	100	99	94
CFAP410	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271	603191	141	100	100	100
CFB	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489	138470	106	100	100	100
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	78	100	100	96
CFI	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439	217030	67	100	99	93
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	87	100	100	97
CHM	Choroideremia, 303100	300390	50	100	94	80
CHMP4B	Cataract 31, multiple types, 605387	610897	178	100	100	100
CHRD1	Megalocornea 1, 309300	300350	48	100	100	92
CHST6	Macular corneal dystrophy, 217800	605294	246	100	100	100
CIB2	Deafness 48, 609439 Usher syndrome, type II, 614869	605564	195	100	100	100
CLCC1	No OMIM phenotype	617539	55	100	96	80

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CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	149	100	100	100
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	111	100	100	100
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	70	100	100	100
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780	606725	120	100	100	99
CLN8	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	129	100	100	100
CLRN1	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902	606397	83	100	100	97
CLUAP1	No OMIM phenotype	616787	58	100	99	89
CNGA1	Retinitis pigmentosa 49, 613756	123825	62	96	91	86
CNGA3	Achromatopsia 2, 216900	600053	118	100	100	100
CNGB1	Retinitis pigmentosa 45, 613767	600724	112	100	100	99
CNGB3	Achromatopsia 3, 262300 Macular degeneration, juvenile, 248200	605080	62	100	100	95
CNNM4	Jalili syndrome, 217080	607805	140	100	100	99
COL11A1	?Deafness 37, 618533 Fibrochondrogenesis 1, 228520 {Lumbar disc herniation, susceptibility to}, 603932 Marshall syndrome, 154780 Stickler syndrome, type II, 604841	120280	62	100	99	93
COL11A2	Deafness 13, 601868 Deafness 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegapiphyseal dysplasia, 184840 Otospondylomegapiphyseal dysplasia, 215150	120290	110	100	100	100
COL18A1	Knobloch syndrome, type 1, 267750	120328	143	100	100	97
COL25A1	Fibrosis of extraocular muscles, congenital, 5, 616219	610004	73	100	98	91

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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 syndrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia	120140	97	100	100	99
COL8A2	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140	120252	95	100	100	98
COL9A1	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134	120210	65	100	99	91
COL9A2	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284	120260	100	100	99	96
CPAMD8	Anterior segment dysgenesis 8, 617319	608841	84	98	94	88
CPLANE1	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	71	100	99	95
CRB1	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105	604210	71	100	100	99
CRX	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829	602225	196	100	100	100
CRYAA	Cataract 9, multiple types, 604219	123580	136	100	100	100
CRYAB	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869	123590	79	100	100	100
CRYBA1	Cataract 10, multiple types, 600881	123610	74	100	100	100

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CRYBA2	?Cataract 42, 115900	600836	113	100	100	100
CRYBA4	Cataract 23, 610425	123631	106	100	100	100
CRYBB1	Cataract 17, multiple types, 611544	600929	97	100	100	100
CRYBB2	Cataract 3, multiple types, 601547	123620	181	100	100	100
CRYBB3	Cataract 22, 609741	123630	105	100	100	100
CRYGB	Cataract 39, multiple types, 615188	123670	87	100	100	100
CRYGC	Cataract 2, multiple types, 604307	123680	86	100	100	100
CRYGD	Cataract 4, multiple types, 115700	123690	97	100	100	100
CRYGS	Cataract 20, multiple types, 116100	123730	104	100	100	100
CSPP1	Joubert syndrome 21, 615636	611654	83	100	100	97
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	125	98	89	87
CTNNA1	Macular dystrophy, patterned, 2, 608970	116805	93	100	100	96
CTNND1	Blepharocheilodontic syndrome 2, 617681	601045	74	100	100	96
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	137	100	100	100
CTSH	No OMIM phenotype	116820	92	100	100	96
CWC27	Retinitis pigmentosa with or without skeletal anomalies, 250410	617170	51	100	96	84
CYP1B1	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300	601771	97	100	100	100
CYP4V2	Bietti crystalline corneoretinal dystrophy, 210370	608614	82	100	100	98
DCN	Corneal dystrophy, congenital stromal, 610048	125255	69	100	100	100
DHDDS	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861	608172	69	100	100	99
DHX38	Retinitis pigmentosa 84, 618220	605584	100	100	100	100
DKC1	Dyskeratosis congenita, 305000	300126	53	100	98	89
DNM1L	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708	603850	70	100	99	94
DNMBP	Cataract 48, 618415	611282	81	100	99	94
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	66	100	99	95
DRAM2	Cone-rod dystrophy 21, 616502	613360	53	100	99	92
DTHD1	No OMIM phenotype	616979	81	100	100	99
DTNBP1	Hermansky-Pudlak syndrome 7, 614076	607145	94	100	100	96
EFEMP1	Doyme honeycomb degeneration of retina, 126600	601548	99	100	100	99

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ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	71	100	100	98
ELP4	?Aniridia 2, 617141	606985	66	98	91	83
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875	616846	117	100	100	99
EPG5	Vici syndrome, 242840	615068	69	100	100	97
EPHA2	Cataract 6, multiple types, 116600	176946	141	100	100	99
EXOSC2	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763	602238	69	100	100	97
EYA1	Anterior segment anomalies with or without cataract, 602588 Branchiootoc syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 ?Otofaciocervical syndrome, 166780	601653	86	100	100	98
EYS	Retinitis pigmentosa 25, 602772	612424	72	100	100	96
FA2H	Spastic paraplegia 35, 612319	611026	83	100	99	91
FAM161A	Retinitis pigmentosa 28, 606068	613596	64	100	100	96
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	186	100	100	100
FBN2	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118	612570	74	100	100	97
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	98	100	100	96
FOXC1	Anterior segment dysgenesis 3, multiple subtypes, 601631 Axenfeld-Rieger syndrome, type 3, 602482	601090	80	100	95	89
FOXE3	Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 Cataract 34, multiple types, 612968	601094	74	87	79	74
FOXL2	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996	605597	114	100	94	88
FRAS1	Fraser syndrome 1, 219000	607830	79	100	100	97

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FREM1	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485	608944	83	100	100	98
FREM2	Cryptophthalmos, unilateral or bilateral, isolated, 123570 Fraser syndrome 2, 617666	608945	104	100	100	100
FRMD7	Nystagmus 1, congenital, 310700 Nystagmus, infantile periodic alternating, 310700	300628	50	100	98	89
FSCN2	Retinitis pigmentosa 30, 607921	607643	139	100	100	100
FTL	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159	134790	144	100	100	100
FYCO1	Cataract 18, 610019	607182	115	100	100	99
FZD4	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780	604579	90	100	100	99
GALK1	Galactokinase deficiency with cataracts, 230200	604313	133	100	100	99
GALT	Galactosemia, 230400	606999	151	100	100	100
GCNT2	Adult i phenotype without cataract, 110800 [Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 116700	600429	81	100	100	98
GDF3	Klippel-Feil syndrome 3, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704	606522	120	100	100	100
GDF6	Klippel-Feil syndrome 1, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Multiple synostoses syndrome 4, 617898	601147	124	100	100	100
GFER	Myopathy progressive, with congenital cataract, hearing loss, and developmental delay, 613076	600924	86	100	100	100

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GJA1	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, 218400 Erythrokeratoderma variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100	121014	101	100	100	100
GJA3	Cataract 14, multiple types, 601885	121015	138	100	100	100
GJA8	Cataract 1, multiple types, 116200	600897	145	100	100	100
GLI2	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829	165230	163	100	100	99
GLIS2	Nephronophthisis 7, 611498	608539	117	100	100	100
GNAT1	Night blindness, congenital stationary 3, 610444 Night blindness, congenital stationary, type 1G, 616389	139330	166	100	100	100
GNAT2	Achromatopsia 4, 613856	139340	72	100	100	98
GNB3	{Hypertension, essential, susceptibility to}, 145500 Night blindness, congenital stationary, type 1H, 617024	139130	118	100	100	100
GNPTG	Mucopolipidosis III gamma, 252605	607838	153	100	99	94
GPR143	Nystagmus 6, congenital, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500	300808	43	100	89	76
GPR179	Night blindness, congenital stationary (complete), 1E, 614565	614515	135	100	100	100
GRASP	No OMIM phenotype	612027	75	94	87	79
GRHL2	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029	608576	81	100	100	99
GRIP1	Fraser syndrome 3, 617667	604597	84	100	100	99
GRK1	Oguchi disease-2, 613411	180381	114	100	100	99
GRM6	Night blindness, congenital stationary (complete), 1B, 257270	604096	127	100	95	90
GRN	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485	138945	155	100	100	100
GSN	Amyloidosis, Finnish type, 105120	137350	95	100	100	99
GUCA1A	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093	600364	108	100	100	100

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GUCA1B	Retinitis pigmentosa 48, 613827	602275	156	100	100	100
GUCY2D	?Choroidal dystrophy, central areolar 1, 215500 Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1I, 618555	600179	112	100	100	99
HARS1	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504	142810	104	100	100	99
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801	300056	45	100	99	84
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	70	94	94	92
HK1	Hemolytic anemia due to hexokinase deficiency, 235700 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460	142600	102	100	100	99
HMGB3	?Microphthalmia, syndromic 13, 300915	300193	40	90	90	79
HMX1	Oculoauricular syndrome, 612109	142992	38	94	72	49
HPS1	Hermansky-Pudlak syndrome 1, 203300	604982	103	100	100	97
HPS3	Hermansky-Pudlak syndrome 3, 614072	606118	66	100	100	95
HPS4	Hermansky-Pudlak syndrome 4, 614073	606682	103	100	100	98
HPS5	Hermansky-Pudlak syndrome 5, 614074	607521	68	100	100	97
HPS6	Hermansky-Pudlak syndrome 6, 614075	607522	130	100	100	93
HSF4	Cataract 5, multiple types, 116800	602438	114	100	100	97
IDH3A	No OMIM phenotype	601149	67	100	99	95
IDH3B	Retinitis pigmentosa 46, 612572	604526	117	100	100	100
IFT140	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920	614620	113	100	100	98
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	75	100	100	96
IFT27	?Bardet-Biedl syndrome 19, 615996	615870	84	100	100	99
IFT43	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866	614068	75	100	100	100
IFT74	?Bardet-Biedl syndrome 20, 617119	608040	46	100	96	83
IFT81	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895	605489	47	99	94	82
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472	300139	69	100	100	90

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IMPDH1	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105	146690	99	100	96	91
IMPG1	Macular dystrophy, vitelliform, 4, 616151	602870	64	100	99	93
IMPG2	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581	607056	68	100	100	97
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	113	100	100	97
INVS	Nephronophthisis 2, infantile, 602088	243305	81	100	100	97
IPO13	No OMIM phenotype	610411	108	100	100	98
IQCB1	Senior-Loken syndrome 5, 609254	609237	54	100	94	78
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	70	100	100	97
KCNJ13	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230	603208	73	100	100	99
KCNV2	Retinal cone dystrophy 3B, 610356	607604	130	100	100	100
KERA	Cornea plana 2, 217300	603288	61	100	100	98
KIAA0556	Joubert syndrome 26, 616784	616650	97	100	99	97
KIAA0586	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546	610178	72	100	98	94
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	73	100	99	93
KIF21A	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700	608283	68	100	99	93
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydroletharus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	107	98	96	93
KIZ	Retinitis pigmentosa 69, 615780	615757	69	100	100	96
KLHL7	Cold-induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943	611119	70	100	100	98
KRT12	Meesmann corneal dystrophy, 122100	601687	94	100	100	100
KRT3	Meesmann corneal dystrophy, 122100	148043	134	100	100	99
LAMA1	Poretti-Boltshauser syndrome, 615960	150320	87	100	100	97
LCA5	Leber congenital amaurosis 5, 604537	611408	74	100	100	99
LEMD2	Cataract 46, juvenile-onset, 212500	616312	99	100	99	95
LIM2	Cataract 19, multiple types, 615277	154045	120	100	100	100
LOXL3	No OMIM phenotype	607163	140	100	100	100

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LRAT	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341	604863	159	100	100	100
LRIT3	Night blindness, congenital stationary (complete), 1F, 615058	615004	78	100	100	100
LRMDA	Albinism, oculocutaneous, type VII, 615179	614537	91	100	95	93
LRP2	Donnai-Barrow syndrome, 222448	600073	69	100	100	96
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 Polycystic liver disease 4 with or without kidney cysts, 617875 van Buchem disease, type 2, 607636	603506	146	100	99	98
LRPAP1	Myopia 23, 615431	104225	135	100	100	100
LSS	Cataract 44, 616509 Hypotrichosis 14, 618275	600909	91	100	100	96
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	106	100	100	99
LYST	Chediak-Higashi syndrome, 214500	606897	72	100	99	95
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	79	100	100	93
MAB21L2	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877	604357	170	100	100	100
MAF	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202	177075	80	84	80	76
MAK	Retinitis pigmentosa 62, 614181	154235	82	100	100	97
MAPKAPK3	?Macular dystrophy, patterned, 3, 617111	602130	73	100	100	97
MERTK	Retinitis pigmentosa 38, 613862	604705	97	100	100	96
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	114	100	100	98
MFRP	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549	606227	100	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	68	100	100	95
MIP	Cataract 15, multiple types, 615274	154050	123	100	100	100
MIR184	EDICT syndrome, 614303	613146	No coverage data	0	0	0
MIR204	?Retinal dystrophy and iris coloboma with or without cataract, 616722	610942	No coverage data	0	0	0
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	87	100	100	99
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	80	100	100	100
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	117	100	100	99
MLPH	Griscelli syndrome, type 3, 609227	606526	107	100	100	100
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	67	100	100	97
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	100	100	100	100
MYO5A	Griscelli syndrome, type 1, 214450	160777	71	100	99	94
MYO7A	Deafness 11, 601317 Deafness 2, 600060 Usher syndrome, type 1B, 276900	276903	109	100	100	99
MYOC	Glaucoma 1A, primary open angle, 137750	601652	140	100	100	100
NAA10	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855 dominant	300013	89	100	100	94
NDP	Exudative vitreoretinopathy 2, 305390 Norrie disease, 310600	300658	77	100	100	100
NEK2	?Retinitis pigmentosa 67, 615565	604043	84	100	98	84
NEK8	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415	609799	132	100	100	100
NEUROD1	{Diabetes mellitus, noninsulin-dependent}, 125853 Maturity-onset diabetes of the young 6, 606394	601724	115	100	100	100

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NHS	Cataract 40, 302200 Nance-Horan syndrome, 302350	300457	57	100	96	90
NMNAT1	Leber congenital amaurosis 9, 608553	608700	90	100	100	98
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	58	100	98	88
NPHP3	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540	608002	66	100	99	94
NPHP4	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996	607215	110	100	100	100
NR2E3	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131	604485	123	100	100	99
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	197	100	100	100
NRL	Retinal degeneration, clumped pigment type Retinitis pigmentosa 27, 613750	162080	108	100	100	100
NYX	Night blindness, congenital stationary (complete), 1A, 310500	300278	132	100	100	100
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	64	100	96	85
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	93	100	100	97
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	43	100	95	80
OFD1	Joubert syndrome 10, 300804 Orofacioidigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	43	100	95	75
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	59	100	98	89
OPA3	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300	606580	130	100	100	100

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OPN1LW	Blue cone monochromacy, 303700 Colorblindness, protan, 303900	300822	121	89	89	88
OPN1MW	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800	300821	47	54	45	42
OPN1SW	Colorblindness, tritan, 190900	613522	78	100	100	100
OPTN	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657	602432	81	100	100	96
OR2W3	No OMIM phenotype	616729	140	100	100	100
OTX2	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125	600037	100	100	100	100
OVOL2	Corneal dystrophy, posterior polymorphous, 1, 122000	616441	107	100	99	94
P3H2	Myopia, high, with cataract and vitreoretinal degeneration, 614292	610341	65	100	100	95
P4HA2	Myopia 25, 617238	600608	101	100	100	98
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	81	100	100	99
PAX2	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330	167409	144	100	100	98
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	73	100	100	96
PCARE	Retinitis pigmentosa 54, 613428	613425	105	100	100	98
PCDH15	Deafness 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083	605514	72	100	100	95
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940	123695	74	100	99	85
PDE6A	Retinitis pigmentosa 43, 613810	180071	74	100	100	96

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PDE6B	Night blindness, congenital stationary 2, 163500 Retinitis pigmentosa-40, 613801	180072	125	100	100	100
PDE6C	Cone dystrophy 4, 613093	600827	67	100	100	96
PDE6D	?Joubert syndrome 22, 615665	602676	86	100	100	100
PDE6G	Retinitis pigmentosa 57, 613582	180073	147	100	100	100
PDE6H	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024	601190	47	100	100	96
PDZD7	Deafness 57, 618003 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472	612971	84	100	100	99
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	62	100	99	94
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	63	100	100	100
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	57	100	100	95
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	47	100	100	89
PHOX2A	Fibrosis of extraocular muscles, congenital, 2, 602078	602753	61	100	100	86
PHYH	Refsum disease, 266500	602026	121	100	100	94
PIGL	CHIME syndrome, 280000	605947	98	100	100	100
PIK3R1	?Agammaglobulinemia 7, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880	171833	80	100	100	97
PIKFYVE	Corneal fleck dystrophy, 121850	609414	67	100	99	95
PITPNM3	Cone-rod dystrophy 5, 600977	608921	99	100	99	97
PITX2	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550	601542	149	100	100	100
PITX3	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623	602669	96	100	100	100
PLA2G5	[Fleck retina, familial benign], 228980	601192	89	100	100	100
PLK4	Microcephaly and chorioretinopathy, 2, 616171	605031	62	100	99	91

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PNPLA6	Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, 612020	603197	123	100	100	99
POC1B	Cone-rod dystrophy 20, 615973	614784	64	100	98	83
POC5	No OMIM phenotype	617880	57	100	99	91
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	93	100	100	99
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	84	100	100	100
PRCD	Retinitis pigmentosa 36, 610599	610598	83	100	100	100
PRDM13	No OMIM phenotype	616741	143	100	100	95
PRDM5	Brittle cornea syndrome 2, 614170	614161	76	100	100	96
PRIMPOL	Myopia 22, 615420	615421	49	95	89	78
PROM1	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786	604365	84	100	99	93
PRPF3	Retinitis pigmentosa 18, 601414	607301	77	100	98	94
PRPF31	Retinitis pigmentosa 11, 600138	606419	101	100	100	99
PRPF4	Retinitis pigmentosa 70, 615922	607795	83	100	100	94
PRPF6	Retinitis pigmentosa 60, 613983	613979	108	100	100	99
PRPF8	Retinitis pigmentosa 13, 600059	607300	102	100	100	98
PRPH2	Choroidal dystrophy, central areolar 2, 613105 Leber congenital amaurosis 18, 608133 Macular dystrophy, patterned, 1, 169150 Macular dystrophy, vitelliform, 3, 608161 Retinitis pigmentosa 7 and digenic form, 608133 Retinitis punctata albescens, 136880	179605	162	100	100	100
PRSS56	Microphthalmia, isolated 6, 613517	613858	95	100	100	100
PXDN	Anterior segment dysgenesis 7, with sclerocornea, 269400	605158	108	100	100	98
RAB28	Cone-rod dystrophy 18, 615374	612994	55	100	100	92
RARB	Microphthalmia, syndromic 12, 615524	180220	85	100	100	99
RAX	Microphthalmia, isolated 3, 611038	601881	124	100	100	93

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RAX2	Cone-rod dystrophy 11, 610381 ?Macular degeneration, age-related, 6, 613757	610362	78	100	100	100
RBP3	?Retinitis pigmentosa 66, 615233	180290	145	100	100	100
RBP4	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147	180250	128	100	100	100
RD3	Leber congenital amaurosis 12, 610612	180040	153	100	100	100
RDH11	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108	607849	82	100	100	99
RDH12	Leber congenital amaurosis 13, 612712	608830	104	100	100	100
RDH5	Fundus albipunctatus, 136880	601617	149	100	100	100
REEP6	Retinitis pigmentosa 77, 617304	609346	174	100	100	99
RGR	Retinitis pigmentosa 44, 613769	600342	101	100	98	91
RGS9	Bradyopsia, 608415	604067	108	100	100	98
RGS9BP	Bradyopsia, 608415	607814	153	100	100	100
RHO	Night blindness, congenital stationary 1, 610445 Retinitis pigmentosa 4 or recessive, 613731 Retinitis punctata albescens, 136880	180380	131	100	100	100
RIMS1	Cone-rod dystrophy 7, 603649	606629	94	100	100	98
RLBP1	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880	180090	111	100	100	100
ROM1	Retinitis pigmentosa 7, digenic form, 608133	180721	110	100	100	100
RP1	Retinitis pigmentosa 1, 180100	603937	73	100	100	97
RP1L1	Occult macular dystrophy, 613587	608581	165	100	100	100
RP2	Retinitis pigmentosa 2, 312600	300757	67	100	100	98
RP9	?Retinitis pigmentosa 9, 180104	607331	44	92	78	72
RPE65	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794	180069	72	100	100	98
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	39	81	72	60
RPGRIP1	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826	605446	88	100	100	96

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RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	60	98	96	89
RS1	Retinoschisis, 312700	300839	44	100	99	74
RTN4IP1	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732	610502	50	100	96	89
SAG	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758	181031	97	100	100	98
SALL2	?Coloboma, ocular, 216820	602219	121	100	100	100
SALL4	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750	607343	155	100	100	97
SAMD11	No OMIM phenotype	616765	92	94	87	79
SCAPER	Intellectual developmental disorder and retinitis pigmentosa, 618195	611611	57	100	98	89
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	125	100	100	100
SDCCAG8	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615	613524	92	100	100	96
SEMA4A	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282	607292	108	100	100	100
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	103	100	94	90
SHH	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250	600725	119	100	100	99
SIPA1L3	?Cataract 45, 616851	616655	149	100	99	97
SIX3	Holoprosencephaly 2, 157170 Schizencephaly, 269160	603714	158	100	100	96
SIX6	Optic disc anomalies with retinal and/or macular dystrophy, 212550	606326	233	100	100	100
SLC16A12	Cataract 47, juvenile, with microcornea, 612018	611910	89	100	100	100
SLC24A1	Night blindness, congenital stationary (complete), 1D, 613830	603617	105	100	100	99
SLC24A5	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750	609802	77	100	100	100
SLC25A46	Neuropathy, hereditary motor and sensory, type VIB, 616505	610826	70	100	100	94
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, 612539	603690	68	100	98	86
SLC38A8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218	615585	83	100	99	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SLC39A5	Myopia 24, 615946	608730	131	100	100	100
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	93	100	100	97
SLC4A11	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal endothelial dystrophy, 217700	610206	125	100	100	99
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707	607882	173	100	100	100
SLC7A14	Retinitis pigmentosa 68, 615725	615720	112	100	100	100
SMOC1	Microphthalmia with limb anomalies, 206920	608488	106	100	100	97
SNRNP200	Retinitis pigmentosa 33, 610359	601664	102	100	100	98
SOX2	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900	184429	183	100	100	100
SOX5	Lamb-Shaffer syndrome, 616803	604975	78	100	100	96
SPATA7	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, 604232	609868	62	100	99	94
SPP2	No OMIM phenotype	602637	73	100	100	100
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	111	100	100	96
STRA6	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186	610745	98	100	100	100
SUFU	Basal cell nevus syndrome, 109400 Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174	607035	104	100	100	100
TACSTD2	Corneal dystrophy, gelatinous drop-like, 204870	137290	228	100	100	100
TCOF1	Treacher Collins syndrome 1, 154500	606847	115	100	100	99
TCTN1	Joubert syndrome 13, 614173	609863	94	100	100	98
TCTN2	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885	613846	84	100	100	97
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	66	100	100	97
TDRD7	Cataract 36, 613887	611258	70	100	100	98
TEAD1	Sveinsson chorioretinal atrophy, 108985	189967	82	100	100	98

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TENM3	?Microphthalmia, isolated, with coloboma 9, 615145 Microphthalmia, syndromic 15, 615145	610083	86	100	100	99
TFAP2A	Branchiooculofacial syndrome, 113620	107580	95	100	100	100
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	82	100	100	98
TGIF1	Holoprosencephaly 4, 142946	602630	152	100	100	100
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	138	100	100	100
TIMP3	Sorsby fundus dystrophy, 136900	188826	104	100	100	100
TMEM107	?Joubert syndrome 29, 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563	616183	100	100	100	94
TMEM126A	Optic atrophy 7, 612989	612988	68	100	100	93
TMEM138	Joubert syndrome 16, 614465	614459	54	100	100	100
TMEM216	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194	613277	87	100	100	95
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	96	100	95	90
TMEM237	Joubert syndrome 14, 614424	614423	57	100	98	87
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 ?RHYS syndrome, 602152	609884	77	100	100	94
TOPORS	Retinitis pigmentosa 31, 609923	609507	73	100	100	100
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	97	100	100	100
TRAF3IP1	Senior-Loken syndrome 9, 616629	607380	55	100	97	86

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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	234	100	100	100
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle 8, 254110	602290	107	100	100	100
TRNT1	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084	612907	68	100	99	92
TRPM1	Night blindness, congenital stationary (complete), 1C, 613216	603576	85	100	100	99
TSPAN12	Exudative vitreoretinopathy 5, 613310	613138	67	100	100	92
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	66	100	100	93
TLL5	Cone-rod dystrophy 19, 615860	612268	77	100	100	98
TUB	?Retinal dystrophy and obesity, 616188	601197	123	100	100	100
TUBA3D	Keratoconus 9, 617928	617878	198	100	100	100
TUBB3	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638	602661	269	100	99	95
TUBB4B	Leber congenital amaurosis with early-onset deafness, 617879	602660	188	100	100	100
TUBGCP4	Microcephaly and chorioretinopathy, 3, 616335	609610	71	100	98	94
TULP1	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132	602280	108	100	100	100
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	83	100	100	98
TYRP1	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271	115501	73	100	100	98
UBIAD1	Corneal dystrophy, Schnyder type, 121800	611632	173	100	100	100
UNC119	?Cone-rod dystrophy ?Immunodeficiency 13, 615518	604011	97	100	100	100
UNC45B	?Cataract 43, 616279	611220	99	100	100	100
USH1C	Deafness 18A, 602092 Usher syndrome, type 1C, 276904	605242	80	100	98	92

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USH1G	Usher syndrome, type 1G, 606943	607696	180	100	100	100
USH2A	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901	608400	82	100	100	98
VAX1	?Microphthalmia, syndromic 11, 614402	604294	104	97	91	87
VCAN	Wagner syndrome 1, 143200	118661	67	100	100	99
VIM	Cataract 30, pulverulent, 116300	193060	106	100	100	96
VSX1	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300	605020	87	100	100	99
VSX2	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093	142993	96	100	100	100
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, 603563	610657	62	100	100	94
WDPCP	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	613580	72	100	99	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	71	100	100	97
WFS1	?Cataract 41, 116400 Deafness 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome 1, 222300 Wolfram-like syndrome, 614296	606201	196	100	100	100
WHRN	Deafness 31, 607084 Usher syndrome, type 2D, 611383	607928	119	100	100	100
WRN	Werner syndrome, 277700	604611	63	100	100	95
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433	606608	73	100	99	94
YME1L1	?Optic atrophy 11, 617302	607472	48	100	95	83
ZEB1	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141	189909	84	100	100	98
ZIC2	Holoprosencephaly 5, 609637	603073	161	96	95	93
ZNF408	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469	616454	127	100	100	100
ZNF423	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844	604557	166	100	100	100

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ZNF469	Brittle cornea syndrome 1, 229200	612078	169	100	100	100
ZNF513	?Retinitis pigmentosa 58, 613617	613598	121	100	100	100
ZNF644	Myopia 21, 614167	614159	63	100	100	99

- Gene symbols according HGNC
- OMIM release used: 8-9-2019
- "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 100 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (± 10 bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (± 10 bp flanking introns) that is covered at least 10x, 20x or 30x