Whole Exome Sequencing
Gene package Vision disorders, version 5, 30-7-2018

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate 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).

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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	85	100	99	95
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	96	100	100	98
ABCC6	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850	603234	124	100	100	99
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	613599	57	100	100	91
ACBD5	No OMIM phenotype	616618	76	100	98	82
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	138	100	97	94
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	208	100	100	100

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ACTG1	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717	102560	174	100	100	100
ADAM9	Cone-rod dystrophy 9, 612775	602713	50	100	98	84
ADAMTS10	Weill-Marchesani syndrome 1, recessive, 277600	608990	103	100	100	99
ADAMTS17	Weill-Marchesani 4 syndrome, recessive, 613195	607511	96	97	96	93
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458	607512	69	100	99	96
ADAMTSL4	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100	610113	110	100	100	99
ADGRV1	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472	602851	67	100	99	93
ADIPOR1	No OMIM phenotype	607945	86	100	100	100
AGBL1	Corneal dystrophy, Fuchs endothelial, 8, 615523	615496	63	98	98	95
AGBL5	Retinitis pigmentosa 75, 617023	615900	84	100	99	95
AGK	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350	610345	49	100	98	88
AHI1	Joubert syndrome 3, 608629	608894	58	100	96	82
AIPL1	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393	604392	131	100	100	100
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	82	100	100	92
ALMS1	Alstrom syndrome, 203800	606844	84	100	99	97
ALX3	Frontonasal dysplasia 1, 136760	606014	113	98	92	88
ANTXR1	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089	606410	49	98	96	84
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	57	100	96	80
ARHGEF18	Retinitis pigmentosa 78, 617433	616432	112	100	99	93
ARL13B	Joubert syndrome 8, 612291	608922	53	100	99	90
ARL2BP	Retinitis pigmentosa with or without situs inversus, 615434	615407	53	100	100	95
ARL3	No OMIM phenotype	604695	104	100	100	97
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	35	96	93	71
ARR3	Myopia 26, X-linked, female-limited, 301010, X-linked	301770	51	100	99	90
ASPH	Traboulsi syndrome, 601552	600582	54	100	98	86
ASRGL1	No OMIM phenotype	609212	62	100	97	90
ATF6	Achromatopsia 7, 616517	605537	56	100	99	92
ATOH7	Persistent hyperplastic primary vitreous, autosomal recessive, 221900	609875	130	99	89	86

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B3GLCT	Peters-plus syndrome, 261540	610308	60	100	95	79
BBIP1	?Bardet-Biedl syndrome 18, 615995	613605	38	100	90	77
BBS1	Bardet-Biedl syndrome 1, 209900	209901	109	100	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	57	100	100	99
BBS12	Bardet-Biedl syndrome 12, 615989	610683	51	100	100	98
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	68	100	100	93
BBS4	Bardet-Biedl syndrome 4, 615982	600374	68	100	99	91
BBS5	Bardet-Biedl syndrome 5, 615983	603650	57	100	97	83
BBS7	Bardet-Biedl syndrome 7, 615984	607590	56	100	99	92
BBS9	Bardet-Biedl syndrome 9, 615986	607968	49	96	93	85
BCOR	Microphthalmia, syndromic 2, 300166	300485	69	100	98	91
BEST1	Bestrophinopathy, autosomal recessive, 611809 Macular dystrophy, vitelliform, 2, 153700 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa, concentric, 613194 Retinitis pigmentosa-50, 613194 Vitreoretinochoroidopathy, 193220	607854	82	100	100	98
BFSP1	Cataract 33, multiple types, 611391	603307	95	100	100	95
BFSP2	Cataract 12, multiple types, 611597	603212	72	100	98	88
BLOC1S3	Hermansky-Pudlak syndrome 8, 614077	609762	113	100	100	100
BLOC1S6	?Hermansky-pudlak syndrome 9, 614171	604310	41	100	98	75
BMP4	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625	112262	95	100	95	89
BMP7	No OMIM phenotype	112267	120	100	100	100
BSG	[Blood group, OK], 111380	109480	108	100	100	99
C12orf57	Temtamy syndrome, 218340	615140	140	100	100	100
C19orf12	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043	614297	166	100	100	100
C1QTNF5	Retinal degeneration, late-onset, autosomal dominant, 605670	608752	93	100	100	95
C21orf2	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271	603191	137	100	100	100
C3	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378	120700	117	100	100	99
C8orf37	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500	614477	71	100	100	91

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CA4	Retinitis pigmentosa 17, 600852	114760	96	100	100	100
CABP4	Cone-rod synaptic disorder, congenital nonprogressive, 610427	608965	144	100	100	100
CACNA1F	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071	300110	66	100	99	91
CACNA2D4	Retinal cone dystrophy 4, 610478	608171	80	100	100	97
CAPN5	Vitreoretinopathy, neovascular inflammatory, 193235	602537	129	100	100	96
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	56	100	99	90
CDH23	Deafness, autosomal recessive 12, 601386 {Pituitary adenoma 5, multiple types}, 617540 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067	605516	123	100	100	100
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553	114021	115	100	100	99
CDHR1	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660	609502	100	100	100	98
CEP164	Nephronophthisis 15, 614845	614848	85	100	100	96
CEP250	No OMIM phenotype	609689	65	100	98	93
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	59	100	96	81
CEP41	Joubert syndrome 15, 614464	610523	61	100	99	92
CEP78	Cone-rod dystrophy and hearing loss, 617236	617110	65	100	98	85
CERKL	Retinitis pigmentosa 26, 608380	608381	63	100	99	89
CFB	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489	138470	94	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	65	100	99	91
CFI	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439	217030	59	100	98	89

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CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	74	100	99	94
CHM	Choroideremia, 303100	300390	44	100	92	73
CHMP4B	Cataract 31, multiple types, 605387	610897	154	100	100	100
CHST6	Macular corneal dystrophy, 217800	605294	240	100	100	100
CIB2	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869	605564	175	100	100	100
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	143	100	100	100
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	101	100	100	99
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	62	100	100	99
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	117	100	100	99
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	123	100	100	100
CLRN1	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902	606397	77	100	100	93
CLUAP1	No OMIM phenotype	616787	51	100	96	84
CNGA1	Retinitis pigmentosa 49, 613756	123825	54	97	93	85
CNGA3	Achromatopsia 2, 216900	600053	101	100	100	100
CNGB1	Retinitis pigmentosa 45, 613767	600724	99	100	100	98
CNGB3	Achromatopsia 3, 262300 Macular degeneration, juvenile, 248200	605080	51	100	99	88
CNNM4	Jalili syndrome, 217080	607805	124	100	99	98
COL11A1	Fibrochondrogenesis 1, 228520 {Lumbar disc herniation, susceptibility to}, 603932 Marshall syndrome, 154780 Stickler syndrome, type II, 604841	120280	53	100	98	84
COL11A2	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150	120290	96	100	100	98
COL18A1	Knobloch syndrome, type 1, 267750	120328	138	100	100	98

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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 sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia	120140	92	100	100	98
COL8A2	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140	120252	91	100	100	100
COL9A1	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134	120210	57	100	97	87
COL9A2	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284	120260	99	100	100	96
CPLANE1	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	59	100	98	90
CRB1	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, autosomal recessive, 600105	604210	62	100	100	98
CRX	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829	602225	169	100	100	100
CRYAA	Cataract 9, multiple types, 604219	123580	130	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	71	100	100	100
CRYBA1	Cataract 10, multiple types, 600881	123610	62	100	100	99
CRYBA2	?Cataract 42, 115900	600836	100	100	100	100
CRYBA4	Cataract 23, 610425	123631	93	100	100	100
CRYBB1	Cataract 17, multiple types, 611544	600929	93	100	100	100
CRYBB2	Cataract 3, multiple types, 601547	123620	160	100	100	100

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CRYBB3	Cataract 22, 609741	123630	99	100	100	100
CRYGB	Cataract 39, multiple types, autosomal dominant, 615188	123670	76	100	100	99
CRYGC	Cataract 2, multiple types, 604307	123680	76	100	100	98
CRYGD	Cataract 4, multiple types, 115700	123690	100	100	100	100
CRYGS	Cataract 20, multiple types, 116100	123730	95	100	100	96
CSPP1	Joubert syndrome 21, 615636	611654	73	100	99	93
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	119	99	91	88
CTNNA1	Macular dystrophy, patterned, 2, 608970	116805	86	100	98	94
CTNND1	Blepharocheilodontic syndrome 2, 617681	601045	61	100	99	93
CTSH	No OMIM phenotype	116820	80	100	100	94
CWC27	Retinitis pigmentosa with or without skeletal anomalies, 250410	617170	43	100	94	75
CYP1B1	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300	601771	94	100	100	100
CYP4V2	Bietti crystalline corneoretinal dystrophy, 210370	608614	73	100	99	95
DCN	Corneal dystrophy, congenital stromal, 610048	125255	58	100	100	98
DHDDS	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861	608172	61	100	100	99
DHX38	No OMIM phenotype	605584	88	100	100	99
DKC1	Dyskeratosis congenita, X-linked, 305000	300126	43	100	97	79
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	54	100	99	90
DRAM2	Cone-rod dystrophy 21, 616502	613360	46	100	97	83
DTHD1	No OMIM phenotype	616979	71	100	100	97
DTNBP1	Hermansky-Pudlak syndrome 7, 614076	607145	85	100	100	94
EFEMP1	Doyne honeycomb degeneration of retina, 126600	601548	89	100	100	98
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	61	100	100	93
ELP4	?Aniridia 2, 617141	606985	58	95	88	74
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875	616846	110	100	100	97
EPG5	Vici syndrome, 242840	615068	61	100	99	93
EPHA2	Cataract 6, multiple types, 116600	176946	128	100	100	99
EXOSC2	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763	602238	61	100	100	95
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	80	100	100	92

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EYS	Retinitis pigmentosa 25, 602772	612424	62	100	99	94
FA2H	Spastic paraplegia 35, autosomal recessive, 612319	611026	73	100	100	95
FAM161A	Retinitis pigmentosa 28, 606068	613596	56	100	99	92
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	175	100	100	100
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	87	100	99	93
FOXC1	Anterior segment dysgenesis 3, multiple subtypes, 601631 Axenfeld-Rieger syndrome, type 3, 602482	601090	72	100	96	90
FOXE3	Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 Cataract 34, multiple types, 612968	601094	60	88	78	70
FOXL2	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996	605597	112	100	95	89
FRAS1	Fraser syndrome 1, 219000	607830	70	100	99	94
FREM1	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485	608944	72	100	100	96
FREM2	Fraser syndrome 2, 617666	608945	93	100	100	98
FRMD7	Nystagmus 1, congenital, X-linked, 310700, X-linked Nystagmus, infantile periodic alternating, X-linked, 310700	300628	42	100	96	81
FSCN2	Retinitis pigmentosa 30, 607921	607643	133	100	100	100
FTL	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159	134790	137	100	100	100
FYCO1	Cataract 18, autosomal recessive, 610019	607182	113	100	100	98
FZD4	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780	604579	74	100	100	97
GALK1	Galactokinase deficiency with cataracts, 230200	604313	133	100	100	98
GALT	Galactosemia, 230400	606999	146	100	100	100

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GCNT2	Adult i phenotype without cataract, 110800 [Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 116700	600429	69	100	99	96
GDF3	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704	606522	105	100	100	100
GDF6	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Multiple synostoses syndrome 4, 617898	601147	127	100	100	100
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076	600924	88	100	100	100
GJA1	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100	121014	82	100	100	97
GJA3	Cataract 14, multiple types, 601885	121015	135	100	100	100
GJA8	Cataract 1, multiple types, 116200	600897	131	100	100	100
GLI2	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829	165230	144	100	100	98
GLIS2	Nephronophthisis 7, 611498	608539	106	100	100	100
GNAT1	Night blindness, congenital stationary, autosomal dominant 3, 610444 Night blindness, congenital stationary, type 1G, 616389	139330	152	100	100	100
GNAT2	Achromatopsia 4, 613856	139340	61	100	100	96
GNB3	{Hypertension, essential, susceptibility to}, 145500 Night blindness, congenital stationary, type 1H, 617024	139130	108	100	100	100
GNPTG	Mucolipidosis III gamma, 252605	607838	149	100	100	95
GPR143	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500	300808	38	100	89	66
GPR179	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565	614515	125	100	100	100
GRASP	No OMIM phenotype	612027	58	94	87	77
GRK1	Oguchi disease-2, 613411	180381	112	100	100	99
GRM6	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270	604096	118	100	95	90

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GRN	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485	138945	148	100	100	100
GSN	Amyloidosis, Finnish type, 105120	137350	92	100	100	97
GUCA1A	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093	600364	99	100	100	100
GUCA1B	Retinitis pigmentosa 48, 613827	602275	142	100	100	100
GUCY2D	?Central areolar choroidal dystrophy 1, 215500 Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000	600179	110	100	100	99
HARS	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504	142810	90	100	100	98
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801	300056	36	100	95	75
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	60	94	94	88
HK1	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460	142600	93	100	99	98
HMGB3	?Microphthalmia, syndromic 13, 300915	300193	33	90	86	66
HMX1	Oculoauricular syndrome, 612109	142992	38	93	73	52
HPS1	Hermansky-Pudlak syndrome 1, 203300	604982	97	100	100	97
HPS3	Hermansky-Pudlak syndrome 3, 614072	606118	58	100	99	90
HPS4	Hermansky-Pudlak syndrome 4, 614073	606682	99	100	100	97
HPS5	Hermansky-Pudlak syndrome 5, 614074	607521	58	100	100	93
HPS6	Hermansky-Pudlak syndrome 6, 614075	607522	127	100	100	99
HSF4	Cataract 5, multiple types, 116800	602438	110	100	100	98
IDH3A	No OMIM phenotype	601149	57	100	99	93
IDH3B	Retinitis pigmentosa 46, 612572	604526	97	100	100	100
IFT140	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920	614620	110	100	99	97
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	65	100	99	92
IFT27	?Bardet-Biedl syndrome 19, 615996	615870	75	100	100	99
IFT74	?Bardet-Biedl syndrome 20, 617119	608040	40	100	94	75
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472	300139	59	100	99	79
IMPDH1	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105	146690	99	100	99	94
IMPG1	Macular dystrophy, vitelliform, 4, 616151	602870	54	100	98	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
IMPG2	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581	607056	57	100	99	93
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	115	100	100	98
INVS	Nephronophthisis 2, infantile, 602088	243305	74	100	99	94
IPO13	No OMIM phenotype	610411	95	100	100	97
IQCB1	Senior-Loken syndrome 5, 609254	609237	48	100	92	74
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	61	100	100	93
KCNJ13	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230	603208	63	100	100	97
KCNV2	Retinal cone dystrophy 3B, 610356	607604	119	100	100	100
KERA	Cornea plana 2, autosomal recessive, 217300	603288	51	100	100	97
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	63	100	98	86
KIF21A	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700	608283	57	100	98	88
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	104	99	96	93
KIZ	Retinitis pigmentosa 69, 615780	615757	59	100	99	91
KLHL7	Cold-induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943	611119	60	100	100	93
KRT12	Meesmann corneal dystrophy, 122100	601687	78	100	100	98
KRT3	Meesmann corneal dystrophy, 122100	148043	106	100	100	98
LAMA1	Poretti-Boltshauser syndrome, 615960	150320	76	100	99	94
LCA5	Leber congenital amaurosis 5, 604537	611408	63	100	100	97
LEMD2	Cataract 46, juvenile-onset, 212500	616312	90	100	98	94
LIM2	Cataract 19, multiple types, 615277	154045	106	100	100	100
LOXL3	No OMIM phenotype	607163	121	100	100	100
LRAT	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341	604863	147	100	100	100
LRIT3	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058	615004	69	100	100	99
LRMDA	Albinism, oculocutaneous, type VII, 615179	614537	88	100	100	91
LRP2	Donnai-Barrow syndrome, 222448	600073	58	100	99	91

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LRP5	[Bone mineral density variability 1], 601884 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 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	137	100	99	97
LRPAP1	Myopia 23, autosomal recessive, 615431	104225	131	100	100	100
LSS	Cataract 44, 616509	600909	82	100	100	95
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	105	100	100	99
LYST	Chediak-Higashi syndrome, 214500	606897	62	100	98	91
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	66	100	100	89
MAB21L2	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877	604357	162	100	100	100
MAF	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202	177075	84	85	80	77
MAK	Retinitis pigmentosa 62, 614181	154235	71	100	99	94
MAPKAPK3	?Macular dystrophy, patterned, 3, 617111	602130	65	100	100	97
MERTK	Retinitis pigmentosa 38, 613862	604705	85	100	99	95
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	107	100	100	98
MFRP	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549	606227	90	100	100	99
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	56	100	99	89
MIP	Cataract 15, multiple types, 615274	154050	115	100	100	100
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	72	100	100	97

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MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	67	100	100	98
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	112	100	100	98
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	55	100	99	92
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	96	100	100	98
MYO7A	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900	276903	105	100	100	99
MYOC	Glaucoma 1A, primary open angle, 137750	601652	130	100	100	98
NAA10	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855	300013	82	100	100	96
NDP	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600	300658	73	100	100	100
NEK2	?Retinitis pigmentosa 67, 615565	604043	74	100	95	81
NEK8	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415	609799	122	100	100	100
NEUROD1	{Diabetes mellitus, noninsulin-dependent}, 125853 Maturity-onset diabetes of the young 6, 606394	601724	104	100	100	100
NHS	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350	300457	46	100	95	83
NMNAT1	Leber congenital amaurosis 9, 608553	608700	75	100	100	95
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	50	100	96	82
NPHP3	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540	608002	57	100	98	88
NPHP4	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996	607215	105	100	100	99
NR2E3	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131	604485	110	100	100	98
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	169	100	100	100

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NRL	Retinal degeneration, autosomal recessive, clumped pigment type Retinitis pigmentosa 27, 613750	162080	107	100	100	100
NYX	Night blindness, congenital stationary (complete), 1A, X-linked, 310500	300278	105	100	100	100
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	54	100	94	80
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	86	100	99	95
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	35	100	91	67
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	37	100	89	63
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	53	100	96	83
OPA3	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300	606580	124	100	100	100
OPN1LW	Blue cone monochromacy, 303700 Colorblindness, protan, 303900	300822	145	100	100	100
OPN1MW	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800	300821	163	100	100	100
OPN1SW	Colorblindness, tritan, 190900	613522	62	100	100	98
OPTN	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657	602432	71	100	99	93
OR2W3	No OMIM phenotype	616729	122	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	86	100	100	100
P3H2	Myopia, high, with cataract and vitreoretinal degeneration, 614292	610341	64	100	99	91
P4HA2	Myopia 25, autosomal dominant, 617238	600608	91	100	100	97
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	72	100	100	96

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PAX2	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330	167409	133	100	100	97
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	66	100	99	94
PCARE	Retinitis pigmentosa 54, 613428	613425	98	100	100	97
PCDH15	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083	605514	60	100	98	89
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940	123695	67	100	97	75
PDE6A	Retinitis pigmentosa 43, 613810	180071	67	100	99	93
PDE6B	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801	180072	111	100	100	100
PDE6C	Cone dystrophy 4, 613093	600827	56	100	99	89
PDE6G	Retinitis pigmentosa 57, 613582	180073	149	100	100	100
PDE6H	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024	601190	40	100	100	80
PDZD7	Deafness, autosomal recessive 57, 618003 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472	612971	82	100	100	98
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	52	100	98	90
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	57	100	100	100
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	49	100	100	95
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	37	100	99	81
PHYH	Refsum disease, 266500	602026	109	100	98	87
PIGL	CHIME syndrome, 280000	605947	87	100	100	97

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PIK3R1	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880	171833	72	100	100	94
PIKFYVE	Corneal fleck dystrophy, 121850	609414	57	100	98	90
PITPNM3	Cone-rod dystrophy 5, 600977	608921	93	100	99	97
PITX2	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550	601542	139	100	100	100
PITX3	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623	602669	108	100	100	100
PLA2G5	[Fleck retina, familial benign], 228980	601192	80	100	100	100
PLK4	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171	605031	53	100	98	87
PNPLA6	Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020	603197	113	100	100	98
POC1B	Cone-rod dystrophy 20, 615973	614784	56	100	95	77
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	80	100	100	98
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	75	100	100	97
PRCD	Retinitis pigmentosa 36, 610599	610598	82	100	100	100
PRDM13	No OMIM phenotype	616741	137	100	100	96
PRDM5	Brittle cornea syndrome 2, 614170	614161	67	100	99	92
PRIMPOL	Myopia 22, autosomal dominant, 615420	615421	41	94	86	69
PROM1	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786	604365	79	100	98	83
PRPF3	Retinitis pigmentosa 18, 601414	607301	64	100	97	93
PRPF31	Retinitis pigmentosa 11, 600138	606419	92	100	100	99
PRPF4	Retinitis pigmentosa 70, 615922	607795	75	100	98	88
PRPF6	Retinitis pigmentosa 60, 613983	613979	105	100	100	97
PRPF8	Retinitis pigmentosa 13, 600059	607300	89	100	100	96

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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, 608133 Retinitis punctata albescens, 136880	179605	143	100	100	100
PRSS56	Microphthalmia, isolated 6, 613517	613858	93	100	100	100
PXDN	Anterior segment dysgenesis 7, with sclerocornea, 269400	605158	99	100	100	98
RAB28	Cone-rod dystrophy 18, 615374	612994	51	100	100	87
RARB	Microphthalmia, syndromic 12, 615524	180220	70	100	100	95
RAX	Microphthalmia, isolated 3, 611038	601881	124	100	100	92
RAX2	Cone-rod dystrophy 11, 610381 ?Macular degeneration, age-related, 6, 613757	610362	85	100	100	100
RBP3	?Retinitis pigmentosa 66, 615233	180290	137	100	100	100
RBP4	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147	180250	121	100	100	100
RD3	Leber congenital amaurosis 12, 610612	180040	163	100	100	100
RDH11	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108	607849	72	100	100	97
RDH12	Leber congenital amaurosis 13, 612712	608830	107	100	100	100
RDH5	Fundus albipunctatus, 136880	601617	132	100	100	100
REEP6	Retinitis pigmentosa 77, 617304	609346	169	100	100	99
RGR	Retinitis pigmentosa 44, 613769	600342	84	100	98	91
RGS9	Bradyopsia, 608415	604067	102	100	100	95
RGS9BP	Bradyopsia, 608415	607814	151	100	100	100
RHO	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880	180380	118	100	100	100
RIMS1	Cone-rod dystrophy 7, 603649	606629	82	100	100	96
RLBP1	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880	180090	102	100	100	99
ROM1	Retinitis pigmentosa 7, digenic, 608133	180721	101	100	100	100
RP1	Retinitis pigmentosa 1, 180100	603937	62	100	99	95
RP1L1	Occult macular dystrophy, 613587	608581	161	100	100	100
RP2	Retinitis pigmentosa 2, 312600	300757	59	100	100	95
RP9	?Retinitis pigmentosa 9, 180104	607331	42	92	78	69

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RPE65	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794	180069	63	100	100	96
RPGR	Cone-rod dystrophy, X-linked, 1, 304020, X-linked Macular degeneration, X-linked atrophic, 300834 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455	312610	33	79	67	53
RPGRIP1	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826	605446	76	100	99	93
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	52	98	94	81
RS1	Retinoschisis, 312700	300839	38	100	96	65
RTN4IP1	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732	610502	43	99	95	82
SAG	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758	181031	87	100	100	93
SALL2	?Coloboma, ocular, autosomal recessive, 216820	602219	113	100	100	100
SALL4	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750	607343	149	100	100	97
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908	604272	126	100	100	100
SDCCAG8	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615	613524	78	100	99	90
SEMA4A	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282	607292	102	100	100	100
SHH	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250	600725	116	100	100	99
SIPA1L3	?Cataract 45, 616851	616655	127	100	99	96
SIX3	Holoprosencephaly 2, 157170 Schizencephaly, 269160	603714	149	100	99	95
SIX6	Optic disc anomalies with retinal and/or macular dystrophy, 212550	606326	210	100	100	100
SLC16A12	Cataract 47, juvenile, with microcornea, 612018	611910	77	100	100	99
SLC24A1	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830	603617	87	100	100	98
SLC24A5	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750	609802	65	100	100	98
SLC25A46	Neuropathy, hereditary motor and sensory, type VIB, 616505	610826	60	100	100	91

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SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539	603690	60	100	96	81
SLC38A8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218	615585	80	100	98	91
SLC39A5	Myopia 24, autosomal dominant, 615946	608730	136	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	83	100	100	96
SLC4A11	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal endothelial dystrophy, autosomal recessive, 217700	610206	114	100	100	99
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707	607882	166	100	100	100
SLC7A14	Retinitis pigmentosa 68, 615725	615720	97	100	100	99
SMOC1	Microphthalmia with limb anomalies, 206920	608488	99	100	100	95
SNRNP200	Retinitis pigmentosa 33, 610359	601664	94	100	100	97
SOX2	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900	184429	161	100	100	100
SPATA7	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232	609868	55	100	98	90
SPP2	No OMIM phenotype	602637	62	100	100	98
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	107	100	99	95
STRA6	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186	610745	93	100	100	100
TACSTD2	Corneal dystrophy, gelatinous drop-like, 204870	137290	223	100	100	100
TCOF1	Treacher Collins syndrome 1, 154500	606847	108	100	100	99
TCTN1	Joubert syndrome 13, 614173	609863	87	100	100	94
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	59	100	100	94
TDRD7	Cataract 36, 613887	611258	57	100	99	94
TEAD1	Sveinsson chorioretinal atrophy, 108985	189967	71	100	100	96
TENM3	Microphthalmia, isolated, with coloboma 9, 615145	610083	71	100	100	97
TFAP2A	Branchiooculofacial syndrome, 113620	107580	93	100	100	100

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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	72	100	100	97
TGIF1	Holoprosencephaly 4, 142946	602630	138	100	100	100
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	131	100	100	100
TIMP3	Sorsby fundus dystrophy, 136900	188826	94	100	100	100
TMEM126A	Optic atrophy 7, 612989	612988	61	100	100	87
TMEM138	Joubert syndrome 16, 614465	614459	46	100	100	95
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	101	100	100	99
TMEM237	Joubert syndrome 14, 614424	614423	48	100	96	83
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550	609884	68	100	99	87
TOPORS	Retinitis pigmentosa 31, 609923	609507	65	100	100	98
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270	607998	87	100	100	99
TRAF3IP1	Senior-Loken syndrome 9, 616629	607380	48	100	94	77
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	217	100	100	100
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110	602290	96	100	100	100
TRNT1	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084	612907	58	100	98	87
TRPM1	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216	603576	76	100	100	97
TSPAN12	Exudative vitreoretinopathy 5, 613310	613138	60	100	100	88
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	57	100	99	87
TTLL5	Cone-rod dystrophy 19, 615860	612268	65	100	100	96
TUB	?Retinal dystrophy and obesity, 616188	601197	112	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
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335	609610	65	100	98	90
TULP1	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132	602280	103	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	77	100	100	96
TYRP1	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271	115501	65	100	100	96
UBIAD1	Corneal dystrophy, Schnyder type, 121800	611632	144	100	100	100
UNC119	?Cone-rod dystrophy ?Immunodeficiency 13, 615518	604011	93	100	100	100
UNC45B	?Cataract 43, 616279	611220	87	100	100	100
USH1C	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904	605242	71	100	95	86
USH1G	Usher syndrome, type 1G, 606943	607696	181	100	100	100
USH2A	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901	608400	70	100	99	96
VAX1	?Microphthalmia, syndromic 11, 614402	604294	96	96	90	86
VCAN	Wagner syndrome 1, 143200	118661	56	100	100	97
VIM	Cataract 30, pulverulent, 116300	193060	105	100	100	98
VSX1	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300	605020	86	100	100	100
VSX2	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093	142993	93	100	100	100
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563	610657	52	100	98	86
WDPCP	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	613580	65	100	98	88
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	62	100	99	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
WFS1	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296	606201	175	100	100	100
WHRN	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383	607928	113	100	100	99
WRN	Werner syndrome, 277700	604611	53	100	98	88
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433	606608	63	100	99	87
YME1L1	?Optic atrophy 11, 617302	607472	41	99	93	75
ZEB1	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141	189909	69	100	99	97
ZIC2	Holoprosencephaly 5, 609637	603073	140	96	94	92
ZNF408	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469	616454	121	100	100	100
ZNF423	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844	604557	161	100	100	100
ZNF469	Brittle cornea syndrome 1, 229200	612078	149	100	100	100
ZNF513	?Retinitis pigmentosa 58, 613617	613598	106	100	100	100
ZNF644	Myopia 21, autosomal dominant, 614167	614159	53	100	100	98

- Gene symbols according HGCN
- OMIM release used: 4-7-2018
- "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 95 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x
- % Covered 30x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 30x