Whole Exome Sequencing
Gene package Vision disorders, version 4, 1-2-2018

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).

Erasmus MC
University Medical Center Rotterdam

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	65	100	98	93
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	64	100	100	97
ABCC6	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850	603234	79	100	100	98
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	613599	42	100	96	69
ACBD5	No OMIM phenotype	616618	49	100	94	76
ACO2	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289	100850	93	100	100	100
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	135	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
ACTG1	Baraitser-Winter syndrome 2, 614583 Deafness 20/26, 604717	102560	121	100	100	100
ADAM9	Cone-rod dystrophy 9, 612775	602713	40	100	94	70
ADAMTS10	Weill-Marchesani syndrome 1, recessive, 277600	608990	74	100	100	98
ADAMTS17	Weill-Marchesani-like syndrome, 613195	607511	66	97	94	87
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458	607512	53	100	98	90
ADAMTSL4	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, 225100	610113	70	100	99	98
ADGRV1	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472	602851	60	100	98	90
ADIPOR1	No OMIM phenotype	607945	71	100	100	100
AGBL1	Corneal dystrophy, Fuchs endothelial, 8, 615523	615496	49	98	98	91
AGBL5	Retinitis pigmentosa 75, 617023	615900	59	100	98	92
AGK	Cataract 38, 614691 Sengers syndrome, 212350	610345	44	100	98	83
AHI1	Joubert syndrome 3, 608629	608894	51	100	94	73
AIPL1	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393	604392	81	100	100	100
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	60	100	98	89
ALMS1	Alstrom syndrome, 203800	606844	82	100	99	96
ALX3	Frontonasal dysplasia 1, 136760	606014	87	100	94	90
ANTXR1	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089	606410	38	98	92	68
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	50	100	95	71
ARHGEF18	Retinitis pigmentosa 78, 617433	616432	60	100	98	92
ARL13B	Joubert syndrome 8, 612291	608922	50	100	99	86
ARL2BP	Retinitis pigmentosa with or without situs inversus, 615434	615407	49	100	100	87
ARL3	No OMIM phenotype	604695	78	100	100	90
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	32	97	88	57
ASPH	Traboulsi syndrome, 601552	600582	42	100	94	70
ASRGL1	No OMIM phenotype	609212	45	100	96	84
ATF6	Achromatopsia 7, 616517	605537	45	100	97	80
ATOH7	Persistent hyperplastic primary vitreous, 221900	609875	91	91	86	82
B3GLCT	Peters-plus syndrome, 261540	610308	47	100	87	64

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
BBIP1	?Bardet-Biedl syndrome 18, 615995	613605	36	100	92	65
BBS1	Bardet-Biedl syndrome 1, 209900	209901	69	100	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	55	100	100	98
BBS12	Bardet-Biedl syndrome 12, 615989	610683	51	100	100	96
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	58	100	99	87
BBS4	Bardet-Biedl syndrome 4, 615982	600374	53	100	96	81
BBS5	Bardet-Biedl syndrome 5, 615983	603650	51	100	99	82
BBS7	Bardet-Biedl syndrome 7, 615984	607590	51	100	98	87
BBS9	Bardet-Biedl syndrome 9, 615986	607968	42	96	94	79
BCOR	Microphthalmia, syndromic 2, 300166	300485	77	100	97	92
BEST1	Bestrophinopathy, 611809 Macular dystrophy, vitelliform, 2, 153700 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa, concentric, 613194 Retinitis pigmentosa-50, 613194 Vitreoretinochoroidopathy, 193220	607854	63	100	99	96
BFSP1	Cataract 33, multiple types, 611391	603307	69	100	100	94
BFSP2	Cataract 12, multiple types, 611597	603212	61	100	98	87
BLOC1S3	Hermansky-Pudlak syndrome 8, 614077	609762	85	100	100	100
BLOC1S6	Hermansky-pudlak syndrome 9, 614171	604310	36	100	92	61
BMP4	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625	112262	72	100	94	84
BMP7	No OMIM phenotype	112267	81	100	100	98
C12orf57	Temtamy syndrome, 218340	615140	103	100	100	100
C19orf12	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, 615043	614297	120	100	100	100
C1QTNF5	Retinal degeneration, late-onset, 605670	608752	87	100	100	94
C21orf2	No OMIM phenotype	603191	86	100	100	99
C2orf71	Retinitis pigmentosa 54, 613428	613425	91	100	100	100
C5orf42	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	54	100	98	88
C8orf37	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500	614477	52	100	100	87
CA4	Retinitis pigmentosa 17, 600852	114760	67	100	100	100
CABP4	Cone-rod synaptic disorder, congenital nonprogressive, 610427	608965	88	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
CACNA1F	Aland Island eye disease, 300600 Cone-rod dystrophy, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, 300071	300110	63	100	99	91
CACNA2D4	Retinal cone dystrophy 4, 610478	608171	57	100	99	93
CAPN5	Vitreoretinopathy, neovascular inflammatory, 193235	602537	73	100	100	93
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	48	100	97	81
CDH23	Deafness 12, 601386 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067	605516	78	100	100	99
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553	114021	77	100	100	98
CDHR1	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660	609502	67	100	100	94
CEP164	Nephronophthisis 15, 614845	614848	63	100	96	88
CEP250	No OMIM phenotype	609689	55	100	98	89
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	57	100	98	84
CEP41	Joubert syndrome 15, 614464	610523	53	100	99	89
CEP78	Cone-rod dystrophy and hearing loss, 617236	617110	47	100	97	78
CERKL	Retinitis pigmentosa 26, 608380	608381	50	100	98	85
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	60	100	99	90
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	60	100	99	93
CHM	Choroideremia, 303100	300390	46	100	91	75
CHMP4B	Cataract 31, multiple types, 605387	610897	113	100	100	100
CHST6	Macular corneal dystrophy, 217800	605294	164	100	100	100
CIB2	Deafness 48, 609439 Usher syndrome, type IJ, 614869	605564	98	100	100	100
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	75	100	100	100
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	76	100	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	57	100	100	98
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	78	100	98	88
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	81	100	100	100
CLRN1	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902	606397	54	100	100	87
CLUAP1	No OMIM phenotype	616787	37	100	93	67
CNGA1	Retinitis pigmentosa 49, 613756	123825	50	96	90	80
CNGA3	Achromatopsia 2, 216900	600053	84	100	100	98
CNGB1	Retinitis pigmentosa 45, 613767	600724	63	100	99	95
CNGB3	Achromatopsia 3, 262300 Macular degeneration, juvenile, 248200	605080	46	100	97	82
CNNM4	Jalili syndrome, 217080	607805	90	100	99	96
COL11A1	Fibrochondrogenesis 1, 228520 {Lumbar disc herniation, susceptibility to}, 603932 Marshall syndrome, 154780 Stickler syndrome, type II, 604841	120280	47	100	96	79
COL11A2	Deafness 13, 601868 Deafness 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, 215150 Stickler syndrome, type III, 184840 Weissenbacher-Zweymuller syndrome, 277610	120290	70	100	100	97
COL18A1	Knobloch syndrome, type 1, 267750	120328	76	100	100	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	65	100	100	96
COL8A2	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140	120252	57	100	100	91
COL9A1	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134	120210	46	100	94	81
COL9A2	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284	120260	65	100	100	95
CRB1	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105	604210	54	100	100	96
CRX	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829	602225	93	100	100	100
CRYAA	Cataract 9, multiple types, 604219	123580	61	100	100	97
CRYAB	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869	123590	64	100	100	100
CRYBA1	Cataract 10, multiple types, 600881	123610	52	100	100	98
CRYBA2	?Cataract 42, 115900	600836	57	100	100	99
CRYBA4	Cataract 23, 610425	123631	65	100	100	100
CRYBB1	Cataract 17, multiple types, 611544	600929	67	100	100	100
CRYBB2	Cataract 3, multiple types, 601547	123620	83	100	100	100
CRYBB3	Cataract 22, 609741	123630	69	100	100	98
CRYGB	Cataract 39, multiple types, 615188	123670	61	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
CRYGC	Cataract 2, multiple types, 604307	123680	64	100	100	99
CRYGD	Cataract 4, multiple types, 115700	123690	77	100	100	100
CRYGS	Cataract 20, multiple types, 116100	123730	83	100	100	97
CSPP1	Joubert syndrome 21, 615636	611654	58	100	99	89
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	604927	78	100	91	86
CTNNA1	Macular dystrophy, patterned, 2, 608970	116805	65	100	99	93
CTNND1	No OMIM phenotype	601045	52	100	99	88
CWC27	Retinitis pigmentosa with or without skeletal anomalies, 250410	617170	38	100	93	62
CYP1B1	Anterior segment dysgenesis 6, multiple subtypes, 617315 Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300	601771	79	100	100	100
CYP4V2	Bietti crystalline corneoretinal dystrophy, 210370	608614	60	100	97	89
DCN	Corneal dystrophy, congenital stromal, 610048	125255	50	100	100	96
DHDDS	Retinitis pigmentosa 59, 613861	608172	47	100	100	92
DHX38	No OMIM phenotype	605584	73	100	100	99
DKC1	Dyskeratosis congenita, 305000	300126	45	100	96	83
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	46	100	99	85
DRAM2	Cone-rod dystrophy 21, 616502	613360	37	100	95	73
DTHD1	No OMIM phenotype	616979	60	100	100	97
DTNBP1	Hermansky-Pudlak syndrome 7, 614076	607145	60	100	97	92
EFEMP1	Doyne honeycomb degeneration of retina, 126600	601548	70	100	100	96
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 ?Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	56	100	100	95
ELP4	?Aniridia 2, 617141	606985	51	98	89	67
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875	616846	80	100	99	92
EPG5	Vici syndrome, 242840	615068	49	100	97	84
EPHA2	Cataract 6, multiple types, 116600	176946	86	100	99	97
EXOSC2	No OMIM phenotype	602238	49	100	99	86
EYA1	Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 ?Otofaciocervical syndrome, 166780	601653	62	100	99	90
EYS	Retinitis pigmentosa 25, 602772	612424	56	100	98	91
FA2H	Spastic paraplegia 35, 612319	611026	56	100	100	92
FAM161A	Retinitis pigmentosa 28, 606068	613596	45	100	99	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	111	100	100	100
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	69	100	99	89
FOXC1	Anterior segment dysgenesis 3, multiple subtypes, 601631 Axenfeld-Rieger syndrome, type 3, 602482	601090	46	99	90	81
FOXE3	Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 Cataract 34, multiple types, 612968	601094	32	82	68	54
FOXL2	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996	605597	86	100	94	88
FRAS1	Fraser syndrome, 219000	607830	56	100	98	90
FREM1	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485	608944	58	100	99	92
FREM2	Fraser syndrome, 219000	608945	74	100	100	98
FRMD7	Nystagmus 1, congenital, 310700 Nystagmus, infantile periodic alternating, 310700	300628	47	100	94	79
FSCN2	Retinitis pigmentosa 30, 607921	607643	78	100	100	98
FTL	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159	134790	90	100	100	100
FYCO1	Cataract 18, 610019	607182	72	100	100	94
FZD4	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780	604579	64	100	100	96
GALK1	Galactokinase deficiency with cataracts, 230200	604313	93	100	100	100
GALT	Galactosemia, 230400	606999	96	100	100	100
GCNT2	Adult i phenotype without cataract, 110800 [Blood group, Ii], 110800 Cataract 13 with adult i phenotype, 116700	600429	62	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
GDF3	Klippel-Feil syndrome 3, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704	606522	93	100	100	100
GDF6	Klippel-Feil syndrome 1, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094	601147	86	100	100	99
GFER	Myopathy progressive, with congenital cataract, hearing loss, and developmental delay, 613076	600924	48	100	100	98
GJA1	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, 218400 Erythrokeratodermia variabilis et progressiva, 133200 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100	121014	79	100	100	100
GJA3	Cataract 14, multiple types, 601885	121015	104	100	100	100
GJA8	Cataract 1, multiple types, 116200	600897	92	100	100	100
GLI2	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829	165230	81	100	100	99
GLIS2	Nephronophthisis 7, 611498	608539	61	100	100	93
GNAT1	Night blindness, congenital stationary 3, 610444 ?Night blindness, congenital stationary, type 1G, 616389	139330	107	100	100	100
GNAT2	Achromatopsia 4, 613856	139340	51	100	100	92
GNB3	{Hypertension, essential, susceptibility to}, 145500 Night blindness, congenital stationary, type 1H, 617024	139130	75	100	100	93
GNPTG	Mucolipidosis III gamma, 252605	607838	103	100	98	89
GPR143	Nystagmus 6, congenital, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500	300808	38	100	89	67
GPR179	Night blindness, congenital stationary (complete), 1E, 614565	614515	91	100	100	100
GRK1	Oguchi disease-2, 613411	180381	75	100	100	97
GRM6	Night blindness, congenital stationary (complete), 1B, 257270	604096	80	100	92	82
GRN	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485	138945	101	100	100	100
GSN	Amyloidosis, Finnish type, 105120	137350	60	100	100	97
GUCA1A	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093	600364	66	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
GUCA1B	Retinitis pigmentosa 48, 613827	602275	96	100	100	100
GUCY2D	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000	600179	73	100	100	99
HARS	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504	142810	71	100	100	96
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801	300056	35	100	90	54
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	48	94	94	85
HK1	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460	142600	68	100	99	94
HMGB3	?Microphthalmia, syndromic 13, 300915	300193	41	90	90	76
HMX1	Oculoauricular syndrome, 612109	142992	34	93	73	62
HPS1	Hermansky-Pudlak syndrome 1, 203300	604982	70	100	100	99
HPS3	Hermansky-Pudlak syndrome 3, 614072	606118	51	100	98	86
HPS4	Hermansky-Pudlak syndrome 4, 614073	606682	78	100	99	93
HPS5	Hermansky-Pudlak syndrome 5, 614074	607521	50	100	99	85
HPS6	Hermansky-Pudlak syndrome 6, 614075	607522	90	100	100	100
HSF4	Cataract 5, multiple types, 116800	602438	78	100	100	98
IDH3A	No OMIM phenotype	601149	46	100	98	86
IDH3B	Retinitis pigmentosa 46, 612572	604526	74	100	100	100
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920	614620	70	100	97	91
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	52	100	98	82
IFT27	?Bardet-Biedl syndrome 19, 615996	615870	54	100	100	96
IFT74	?Bardet-Biedl syndrome 20, 617119	608040	36	100	93	65
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472	300139	59	100	99	84
IMPDH1	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105	146690	67	100	94	87
IMPG1	Macular dystrophy, vitelliform, 4, 616151	602870	47	100	97	87
IMPG2	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581	607056	50	100	98	86
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	80	100	100	96
INVS	Nephronophthisis 2, infantile, 602088	243305	56	100	98	89
IQCB1	Senior-Loken syndrome 5, 609254	609237	42	100	89	67
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	49	100	97	83

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
KCNJ13	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230	603208	57	100	100	97
KCNV2	Retinal cone dystrophy 3B, 610356	607604	93	100	100	100
KERA	Cornea plana 2, 217300	603288	49	100	100	97
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	58	100	99	87
KIF21A	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700	608283	51	100	96	80
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	66	99	95	89
KIZ	Retinitis pigmentosa 69, 615780	615757	49	100	98	86
KLHL7	Cold-induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943	611119	54	100	100	91
KRT12	Meesmann corneal dystrophy, 122100	601687	63	100	100	97
KRT3	Meesmann corneal dystrophy, 122100	148043	61	100	100	92
LAMA1	Poretti-Boltshauser syndrome, 615960	150320	57	100	98	89
LCA5	Leber congenital amaurosis 5, 604537	611408	63	100	100	97
LEMD2	Cataract 46, juvenile-onset, 212500	616312	59	100	96	88
LIM2	Cataract 19, multiple types, 615277	154045	74	100	100	99
LRAT	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341	604863	105	100	100	100
LRIT3	Night blindness, congenital stationary (complete), 1F, 615058	615004	61	100	100	98
LRMDA	Albinism, oculocutaneous, type VII, 615179	614537	56	100	94	83
LRMDA	Albinism, oculocutaneous, type VII, 615179	614537	56	100	94	83
LRP2	Donnai-Barrow syndrome, 222448	600073	47	100	97	84
LRP5	[Bone mineral density variability 1], 601884 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 {Osteoporosis}, 166710 Osteosclerosis, 144750 van Buchem disease, type 2, 607636	603506	81	100	98	96
LSS	Cataract 44, 616509	600909	47	100	94	82

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	70	100	100	99
LYST	Chediak-Higashi syndrome, 214500	606897	55	100	99	91
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	57	100	98	83
MAB21L2	Microphthalmia, syndromic 14, 615877	604357	118	100	100	100
MAF	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202	177075	52	85	80	75
MAK	Retinitis pigmentosa 62, 614181	154235	67	100	99	92
MAPKAPK3	?Macular dystrophy, patterned, 3, 617111	602130	51	100	98	85
MERTK	Retinitis pigmentosa 38, 613862	604705	64	100	99	93
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	74	100	100	95
MFRP	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549	606227	76	100	100	100
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	50	100	98	81
MIP	Cataract 15, multiple types, 615274	154050	85	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			
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	58			
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	59	100	100	99
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	81	100	100	97
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	49	100	99	89
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	58	100	100	91

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MYO7A	Deafness 11, 601317 Deafness 2, 600060 Usher syndrome, type 1B, 276900	276903	68	100	98	95
MYOC	Glaucoma 1A, primary open angle, 137750	601652	105	100	100	96
NAA10	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855 dominant	300013	56	100	95	92
NDP	Exudative vitreoretinopathy 2, 305390 Norrie disease, 310600	300658	57	100	100	100
NEK2	?Retinitis pigmentosa 67, 615565	604043	61	100	97	84
NEK8	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415	609799	92	100	100	100
NEUROD1	{Diabetes mellitus, noninsulin-dependent}, 125853 Maturity-onset diabetes of the young 6, 606394	601724	102	100	100	100
NHS	Cataract 40, 302200 Nance-Horan syndrome, 302350	300457	44	100	92	84
NMNAT1	Leber congenital amaurosis 9, 608553	608700	62	100	100	94
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	39	100	92	67
NPHP3	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540	608002	47	100	97	82
NPHP4	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996	607215	67	100	100	98
NR2E3	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131	604485	75	100	100	100
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	82	100	98	91
NRL	Retinal degeneration, clumped pigment type Retinitis pigmentosa 27, 613750	162080	71	100	100	100
NYX	Night blindness, congenital stationary (complete), 1A, 310500	300278	72	100	100	100
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	613349	53	100	100	89
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	60	100	99	89
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	36	100	92	67

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	42	100	97	81
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	46	100	96	77
OPA3	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300	606580	66	100	100	100
OPN1LW	Blue cone monochromacy, 303700 Colorblindness, protan, 303900	300822	93	100	100	100
OPN1MW	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800	300821	103	100	100	99
OPN1SW	Colorblindness, tritan, 190900	613522	57	100	100	100
OPTN	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657	602432	56	100	98	87
OR2W3	No OMIM phenotype	616729	98	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	80	100	100	100
P3H2	Myopia, high, with cataract and vitreoretinal degeneration, 614292	610341	47	100	97	80
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	57	100	100	92
PAX2	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330	167409	78	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	50	100	98	84

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PCDH15	Deafness 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083	605514	55	100	98	87
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940	123695	57	100	90	74
PDE6A	Retinitis pigmentosa 43, 613810	180071	50	100	98	84
PDE6B	Night blindness, congenital stationary 2, 163500 Retinitis pigmentosa-40, 613801	180072	75	100	100	98
PDE6C	Cone dystrophy 4, 613093	600827	48	100	99	87
PDE6G	Retinitis pigmentosa 57, 613582	180073	91	100	100	100
PDE6H	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024	601190	39	100	100	74
PDZD7	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472	612971	67	100	100	97
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	49	100	98	84
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	52	100	100	99
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	43	100	100	87
PGK1	Phosphoglycerate kinase 1 deficiency, 300653	311800	48	100	100	91
PHYH	Refsum disease, 266500	602026	80	100	92	78
PIGL	CHIME syndrome, 280000	605947	62	100	100	81
PIK3R1	?Agammaglobulinemia 7, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880	171833	59	100	99	89
PIKFYVE	Corneal fleck dystrophy, 121850	609414	49	100	98	87
PITPNM3	Cone-rod dystrophy 5, 600977	608921	66	99	99	97
PITX2	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550	601542	98	100	100	100
PITX3	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, 610623	602669	97	100	100	100
PLA2G5	[Fleck retina, familial benign], 228980	601192	56	100	100	94
PLK4	Microcephaly and chorioretinopathy, 2, 616171	605031	49	100	96	81

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PNPLA6	Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, 612020	603197	78	100	100	98
POC1B	Cone-rod dystrophy 20, 615973	614784	46	100	93	66
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	63	100	100	95
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	51	100	100	87
PRCD	Retinitis pigmentosa 36, 610599	610598	50	100	100	98
PRDM13	No OMIM phenotype	616741	84	100	100	95
PRDM5	Brittle cornea syndrome 2, 614170	614161	54	100	99	87
PROM1	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786	604365	57	100	96	74
PRPF3	Retinitis pigmentosa 18, 601414	607301	58	100	97	90
PRPF31	Retinitis pigmentosa 11, 600138	606419	66	100	100	94
PRPF4	Retinitis pigmentosa 70, 615922	607795	59	100	96	79
PRPF6	Retinitis pigmentosa 60, 613983	613979	76	100	100	95
PRPF8	Retinitis pigmentosa 13, 600059	607300	71	100	99	94
PRPH2	Choriodal dystrophy, central areolar 2, 613105 Leber congenital amaurosis 18, 608133 Macular dystrophy, patterned, 1, 169150 Macular dystrophy, vitelliform, 3, 608161 Retinitis pigmentosa 7 and digenic, 608133 Retinitis punctata albescens, 136880	179605	94	100	100	100
PRSS56	Microphthalmia, isolated 6, 613517	613858	58	100	100	99
PXDN	Anterior segment dysgenesis 7, with sclerocornea, 269400	605158	82	100	99	94
RAB28	Cone-rod dystrophy 18, 615374	612994	41	100	100	82
RARB	Microphthalmia, syndromic 12, 615524	180220	54	100	100	92
RAX	Microphthalmia, isolated 3, 611038	601881	72	100	100	89
RAX2	Cone-rod dystrophy 11, 610381 ?Macular degeneration, age-related, 6, 613757	610362	63	100	100	100
RBP3	?Retinitis pigmentosa 66, 615233	180290	98	100	100	100
RBP4	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147	180250	71	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
RCBTB1	Retinal dystrophy with or without extraocular anomalies, 617175	607867	63	100	100	90
RD3	Leber congenital amaurosis 12, 610612	180040	82	100	100	100
RDH11	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108	607849	59	100	100	89
RDH12	Leber congenital amaurosis 13, 612712	608830	76	100	100	95
RDH5	Fundus albipunctatus, 136880	601617	81	100	100	100
REEP6	Retinitis pigmentosa 77, 617304	609346	103	100	98	93
RGR	Retinitis pigmentosa 44, 613769	600342	65	100	100	96
RGS9	Bradyopsia, 608415	604067	65	100	98	85
RGS9BP	Bradyopsia, 608415	607814	92	100	100	100
RHO	Night blindness, congenital stationary 1, 610445 Retinitis pigmentosa 4 or recessive, 613731 Retinitis punctata albescens, 136880	180380	78	100	100	100
RIMS1	Cone-rod dystrophy 7, 603649	606629	63	100	99	91
RLBP1	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880	180090	72	100	100	92
ROM1	Retinitis pigmentosa 7, digenic, 608133	180721	74	100	100	100
RP1	Retinitis pigmentosa 1, 180100	603937	60	100	100	96
RP1L1	Occult macular dystrophy, 613587	608581	98	100	100	100
RP2	Retinitis pigmentosa 2, 312600	300757	49	100	100	93
RP9	?Retinitis pigmentosa 9, 180104	607331	36	78	78	60
RPE65	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794	180069	52	100	100	94
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	37	82	70	59
RPGRIP1	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826	605446	59	100	98	86
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	47	97	94	77
RS1	Retinoschisis, 312700	300839	38	100	92	69
RTN4IP1	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732	610502	37	100	94	74
SAG	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758	181031	59	100	99	84
SALL2	?Coloboma, ocular, 216820	602219	94	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
SDCCAG8	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615	613524	66	100	99	87
SEMA4A	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282	607292	72	100	100	99
SHH	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250	600725	68	100	98	95
SIPA1L3	?Cataract 45, 616851	616655	76	100	96	90
SIX3	Holoprosencephaly 2, 157170 Schizencephaly, 269160	603714	99	100	100	100
SIX6	Optic disc anomalies with retinal and/or macular dystrophy, 212550	606326	131	100	100	100
SLC16A12	Cataract 47, juvenile, with microcornea, 612018	611910	63	100	100	96
SLC24A1	Night blindness, congenital stationary (complete), 1D, 613830	603617	68	100	100	97
SLC24A5	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750	609802	64	100	100	98
SLC25A46	Neuropathy, hereditary motor and sensory, type VIB, 616505	610826	52	100	100	91
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, 612539	603690	55	100	97	78
SLC38A8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218	615585	52	100	99	89
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	60	100	100	89
SLC4A11	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal endothelial dystrophy, 217700	610206	86	100	100	99
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707	607882	114	100	100	100
SLC7A14	Retinitis pigmentosa 68, 615725	615720	76	100	100	97
SMOC1	Microphthalmia with limb anomalies, 206920	608488	68	100	98	91
SNRNP200	Retinitis pigmentosa 33, 610359	601664	72	100	99	94
SOX2	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900	184429	100	100	100	100
SPATA7	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, 604232	609868	51	100	98	89
SPP2	No OMIM phenotype	602637	50	100	100	98
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713	611715	63	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
STRA6	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186	610745	69	100	100	99
TACSTD2	Corneal dystrophy, gelatinous drop-like, 204870	137290	162	100	100	100
TCOF1	Treacher Collins syndrome 1, 154500	606847	78	100	100	98
TCTN1	Joubert syndrome 13, 614173	609863	61	100	99	88
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	50	100	99	90
TDRD7	Cataract 36, 613887	611258	50	100	99	89
TEAD1	Sveinsson chorioretinal atrophy, 108985	189967	57	100	100	90
TENM3	Microphthalmia, isolated, with coloboma 9, 615145	610083	60	100	99	96
TFAP2A	Branchiooculofacial syndrome, 113620	107580	80	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	52	100	98	85
TGIF1	Holoprosencephaly 4, 142946	602630	94	100	100	100
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	102	100	100	100
TIMP3	Sorsby fundus dystrophy, 136900	188826	65	100	100	100
TMEM126A	Optic atrophy 7, 612989	612988	57	100	100	79
TMEM138	Joubert syndrome 16, 614465	614459	38	100	100	87
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	71	100	100	90
TMEM237	Joubert syndrome 14, 614424	614423	38	100	90	69
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550	609884	58	100	98	82
TOPORS	Retinitis pigmentosa 31, 609923	609507	57	100	100	100
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	70	100	100	99
TRAF3IP1	Senior-Loken syndrome 9, 616629	607380	42	100	95	75

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	116	100	100	100
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110	602290	86	100	100	100
TRNT1	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084	612907	55	100	97	83
TRPM1	Night blindness, congenital stationary (complete), 1C, 613216	603576	60	100	100	95
TSPAN12	Exudative vitreoretinopathy 5, 613310	613138	48	100	98	79
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	46	100	99	85
TTLL5	Cone-rod dystrophy 19, 615860	612268	53	100	99	87
TUB	?Retinal dystrophy and obesity, 616188	601197	79	100	100	100
TUBGCP4	Microcephaly and chorioretinopathy, 3, 616335	609610	51	100	96	80
TULP1	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132	602280	69	100	100	98
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	67	100	99	93
TYRP1	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271	115501	52	100	100	93
UBIAD1	Corneal dystrophy, Schnyder type, 121800	611632	93	100	100	100
UNC119	?Cone-rod dystrophy ?Immunodeficiency 13, 615518	604011	65	100	100	96
UNC45B	?Cataract 43, 616279	611220	65	100	100	99
USH1C	Deafness 18A, 602092 Usher syndrome, type 1C, 276904	605242	57	100	98	89
USH1G	Usher syndrome, type 1G, 606943	607696	111	100	100	100
USH2A	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901	608400	59	100	99	92
VAX1	?Microphthalmia, syndromic 11, 614402	604294	59	99	92	86
VCAN	Wagner syndrome 1, 143200	118661	53	100	100	96
VIM	Cataract 30, pulverulent, 116300	193060	64	100	99	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
VSX1	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300	605020	59	100	100	97
VSX2	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093	142993	60	100	100	97
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, 603563	610657	45	100	97	78
WDPCP	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	613580	53	100	98	84
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	50	100	98	85
WFS1	?Cataract 41, 116400 Deafness 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome, 222300 Wolfram-like syndrome, 614296	606201	106	100	100	100
WHRN	Deafness 31, 607084 Usher syndrome, type 2D, 611383	607928	87	100	100	100
WRN	Werner syndrome, 277700	604611	47	100	98	84
YAP1	Coloboma, ocular, 120433 Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433	606608	49	100	95	79
YME1L1	?Optic atrophy 11, 617302	607472	36	100	91	64
ZEB1	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141	189909	62	100	100	97
ZIC2	Holoprosencephaly 5, 609637	603073	82	95	90	87
ZNF408	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469	616454	79	100	100	100
ZNF423	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844	604557	121	100	100	99
ZNF469	Brittle cornea syndrome 1, 229200	612078	88	100	100	100
ZNF513	?Retinitis pigmentosa 58, 613617	613598	96	100	100	100
ZNF644	Myopia 21, 614167	614159	54	100	100	99

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x

- The statistics above are based on a set of 96 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