VISION DISORDERS GENE PANEL

Gene symbol	Depth (reads)	Coverage (avg %)	OMIM disease	Description
ABCA4	100	85	604116	Cone-rod dystrophy 3
ABCC6	79	82	614473	Arterial calcification generalized of infancy 2
ABHD12	72	98	612674	Polyneuropathy hearing loss ataxia retinitis pigmentosa and cataract
ACBD5	107	87	188000	Thrombocytopenia 2
ADAM9	114	95	612775	Cone-rod dystrophy 9
AHI1	117	97	608629	Joubert syndrome-3
AIPL1	75	91	604393	Cone-rod dystrophy
ALMS1	217	94	203800	Alstrom syndrome
ARL6	142	99	209900	Bardet-Biedl syndrome 3
BBS1	126	89	209900	Bardet-Biedl syndrome 1
BBS10	128	100	209900	Bardet-Biedl syndrome 10
BBS12	163	100	209900	Bardet-Biedl syndrome 12
BBS2	130	91	209900	Bardet-Biedl syndrome 2
BBS4	104	87	209900	Bardet-Biedl syndrome 4
BBS5	114	96	209900	Bardet-Biedl syndrome 5
BBS7	121	98	209900	Bardet-Biedl syndrome 7
BBS9	117	95	209900	Bardet-Biedl syndrome 9
BEST1	110	94	153700	Best macular dystrophy
C1QTNF5	80	69	605670	Retinal degeneration late-onset autosomal dominant
C2orf71	110	99	613428	Retinitis pigmentosa 54
C8orf37	94	86	614500	Cone-rod dystrophy 16
CA4	79	91	600852	Retinitis pigmentosa 17
CABP4	71	87	610427	Night blindness congenital stationary (incomplete) 2B autosomal recessive
CACNA1F	69	89	300600	Aland Island eye disease
CACNA2D4	91	91	610478	Retinal cone dystrophy 4
CAPN5	82	83	193235	Vitreoretinopathy, neovascular inflammatory
CC2D2A	101	90	216360	COACH syndrome
CDH23	92	88	601386	Deafness autosomal recessive 12

CDH3	104	86	225280	Ectodermal dysplasia ectrodactyly and macular dystrophy
CDHR1	108	86	613660	Cone-rod dystrophy 15
CEP164	89	87	614845	Nephronophthisis 15
CEP290	95	92	209900	Bardet-Biedl syndrome 14
CERKL	127	93	608380	Retinitis pigmentosa 26
CFH	112	70	610698	{Macular degeneration, age-related, 4, susceptibility to}
CFH	112	70	126700	Basal laminar drusen
CHM	79	94	303100	Choroideremia
CIB2	76	79	609439	Deafness autosomal recessive 48
CLN3	89	87	204200	Ceroid lipofuscinosis neuronal 3
CLRN1	172	98	614180	Retinitis pigmentosa 61
CNGA1	128	98	613756	Retinitis pigmentosa 49
CNGA3	142	97	216900	Achromatopsia-2
CNGB1	96	87	613767	Retinitis pigmentosa 45
CNGB3	111	85	262300	Achromatopsia-3
CNNM4	162	94	217080	Jalili syndrome
COL11A1	98	96	228520	Fibrochondrogenesis
COL11A2	15	85	601868	Deafness autosomal dominant 13
COL2A1	95	86	200610	Achondrogenesis type II or hypochondrogenesis
COL9A1	110	91	614135	Epiphyseal dysplasia multiple 6
CRB1	175	93	613835	Leber congenital amaurosis 8
CRX	149	91	120970	Cone-rod retinal dystrophy-2
CYP4V2	113	89	210370	Bietti crystalline corneoretinal dystrophy
DFNB31	79	87	611383	Usher syndrome, type 2D
DHDDS	105	85	613861	Retinitis pigmentosa 59
EFEMP1	123	95	126600	Doyne honeycomb degeneration of retina
EIF4G1	113	88	614251	Parkinson disease 18
ELOVL4	111	92	614457	Ichthyosis spastic quadriplegia and mental retardation
EMC1	118	84	247100	Urbach-Wiethe disease
EYS	128	95	602772	Retinitis pigmentosa 25
FAM161A	133	97	606068	Retinitis pigmentosa 28
FLVCR1	95	96	609033	Ataxia posterior column with retinitis pigmentosa
FSCN2	84	94	607921	Retinitis pigmentosa 30

FZD4	144	100	133780	Exudative vitreoretinopathy
GLIS2	95	93	611498	Nephronophthisis 7
GNAT1	83	88	610444	Night blindness congenital stationary autosomal dominant 3
GNAT2	129	83	613856	Achromatopsia-4
GNPTG	69	100	252605	Mucolipidosis III gamma
GPR179	150	98	614565	Night blindness congenital stationary (complete) 1E autosomal recessive
GPR98	120	93	604352	Febrile seizures familial 4
GRK1	96	89	613411	Oguchi disease-2
GRM6	79	91	257270	Night blindness congenital stationary (complete) 1B autosomal recessive
GUCA1A	80	81	602093	Cone dystrophy-3
GUCA1B	118	100	613827	Retinitis pigmentosa 48
GUCY2D	77	90	200	Cone-rod dystrophy 6
HARS	139	89	614504	Usher syndrome type 3B
HMCN1	122	93	603075	{Macular degeneration, age-related, 1}
IDH3B	141	92	612572	Retinitis pigmentosa 46
IMPDH1	54	35	613837	Leber congenital amaurosis 11
IMPG2	136	93	613581	Maculopathy IMPG2-related
INPP5E	67	95	213300	Joubert syndrome 1
INVS	128	90	602088	Nephronophthisis 2 infantile
IQCB1	94	73	609254	Senior-Loken syndrome 5
KCNJ13	195	99	614186	Leber congenital amaurosis 16
KCNV2	75	95	610356	Retinal cone dystrophy 3B
KLHL7	123	94	612943	Retinitis pigmentosa 42
LCA5	157	98	604537	Leber congenital amaurosis 5
LRAT	182	100	613341	Leber congenital amaurosis 14
LRIT3	161	97	615058	Night blindness congenital stationary (complete) 1F autosomal recessive
LRP5	88	84	601813	Exudative vitreoretinopathy 4
LZTFL1	99	95	209900	Bardet-Biedl syndrome
MAK	94	85	614181	REtinitis pigmentosa 62
MERTK	120	90	613862	Retinitis pigmentosa 38
MFN2	112	88	609260	Charcot-Marie-Tooth disease type 2A2
MKKS	147	97	209900	Bardet-Biedl syndrome 6
MKS1	126	86	209900	Bardet-Biedl syndrome 13

MTTP	111	90	545000	Merff syndrome
MVK	101	78	260920	Hyper-IgD syndrome
MYO7A	82	87	601317	Deafness autosomal dominant 11
NDP	112	86	305390	Exudative vitreoretinopathy X-linked
NEK8	118	89	613824	Nephronophthisis 9
NMNAT1	144	52	608553	Leber congenital amaurosis 9
NPHP1	117	92	609583	Joubert syndrome 4
NPHP3	112	95	267010	Meckel syndrome 7
NPHP4	94	82	606966	Nephronophthisis 4
NR2E3	85	91	268100	Enhanced S-cone syndrome
NRL	48	95	200	Retinal degeneration autosomal recessive
NYX	49	97	310500	Night blindness congenital stationary (complete) 1A X-linked
OAT	58	58	258870	Gyrate atrophy of choroid and retina with or without ornithinemia
OFD1	61	54	300804	Joubert syndrome 10
OPA1	121	99	165500	Optic atrophy 1
OPA3	85	87	258501	3-methylglutaconic aciduria type III
OPN1SW	108	81	190900	Colorblindness tritan
OTX2	152	95	610125	Microphthalmia syndromic 5
PANK2	113	97	607236	HARP syndrome
PCDH15	129	94	609533	Deafness autosomal recessive 23
PDE6A	109	90	613810	Retinitis pigmentosa 43
PDE6B	96	96	163500	Night blindness congenital stationary autosomal dominant 2
PDE6C	116	97	613093	Cone dystrophy 4
PDE6G	72	91	613582	Retinitis pigmentosa 57
PDE6H	49	84	610024	Achromatopsia 6
PDZD7	90	83	605472	Usher syndrome type IIC GPR98/PDZD7 digenic
PEX1	122	93	214100	Peroxisome biogenesis disorder 1A (Zellweger)
PEX2	142	76	614866	Peroxisome biogenesis disorder 5A (Zellweger)
PEX2	142	76	614867	Peroxisome biogenesis disorder 5B
PEX7	112	91	614879	Peroxisome biogenesis disorder 9B
PHYH	90	93	266500	Refsum disease
PITPNM3	73	85	600977	Cone-rod dystrophy 5
PLA2G5	123	74	228980	Fleck retina familial benign

PRCD	116	46	610599	Retinitis pigmentosa 36
PROM1	90	91	612657	Cone-rod dystrophy 12
PRPF3	116	78	601414	Retinitis pigmentosa 18
PRPF31	89	72	600138	Retinitis pigmentosa 11
PRPF6	96	82	613983	Retinitis pigmentosa 60
PRPF8	141	89	600059	Retinitis pigmentosa 13
PRPH2	152	93	613105	Choriodal dystrophy central areolar 2
RAB28	99	75	615374	Cone-rod dystrophy 18
RAX2	46	98	610381	Cone-rod dystrophy 11
RBP3	97	97	615233	Retinitis pigmentosa 66
RBP4	105	88	615147	Retinol dystrophy iris coloboma and comedogenic acne syndrome
RD3	43	96	610612	Leber congenital amaurosis 12
RDH12	83	78	612712	Leber congenital amaurosis 13
RDH5	115	83	136880	Fundus albipunctatus
RGR	94	81	613769	Retinitis pigmentosa 44
RGS9	122	88	608415	Bradyopsia
RGS9BP	34	99	608415	Bradyopsia
RHO	114	88	610445	Night blindness congenital stationary autosomal dominant 1
RIMS1	103	93	603649	Cone-rod dystrophy 7
RLBP1	104	75	607475	Bothnia retinal dystrophy
ROM1	89	100	608133	Retinitis pigmentosa 7 digenic
RP1	192	98	180100	Retinitis pigmentosa 1
RP1L1	151	25	613587	Occult macular dystrophy
RP2	93	99	312600	Retinitis pigmentosa 2
RP9	56	77	180104	Retinitis pigmentosa 9
RPE65	129	88	204100	Leber congenital amaurosis 2
RPGR	100	89	304020	Cone-rod dystrophy X-linked 1
RPGRIP1	133	85	608194	Cone-rod dystrophy 13
RPGRIP1L	100	92	216360	COACH syndrome
RS1	67	86	312700	Retinoschisis
SAG	120	89	258100	Oguchi disease-1
SDCCAG8	96	88	613615	Senior-Loken syndrome 7
SEMA4A	100	90	610283	Cone-rod dystrophy 10

SLC24A1	148	80	613830	Night blindness congenital stationary (complete) 1D autosomal recessive
SNRNP200	129	85	610359	Retinitis pigmentosa 33
SPATA7	131	98	604232	Leber congenital amaurosis 3
TEAD1	97	86	108985	Sveinsson choreoretinal atrophy
TIMM8A	56	60	200	Deafness X-linked 1
TIMP3	133	89	136900	Sorsby fundus dystrophy
TMEM126A	76	92	612989	Optic atrophy-7
TMEM237	99	91	614424	Joubert syndrome 14
TMEM67	107	96	216360	COACH syndrome
TOPORS	163	99	609923	Retinitis pigmentosa 31
TRIM32	110	100	209900	Bardet-Biedl syndrome 11
TRPM1	141	88	613216	Night blindness congenital stationary (complete) 1C autosomal recessive
TSPAN12	127	100	613310	Exudative vitreoretinopathy 5
TTC8	107	91	209900	Bardet-Biedl syndrome 8
TTPA	97	97	277460	Ataxia with isolated vitamin E deficiency
TULP1	92	87	613843	Leber congenital amaurosis 15
USH1C	89	90	602092	Deafness autosomal recessive 18A
USH1G	103	93	606943	Usher syndrome, type 1G
USH2A	121	91	613809	Retinitis pigmentosa 39
VCAN	167	99	143200	Wagner syndrome 1
WDPCP	90	93	209900	Bardet-Biedl syndrome 15
WDR19	122	94	614376	Asphyxiating thoracic dystrophy 5
WFS1	141	92	600965	Deafness autosomal dominant 6/14/38
ZNF408	88	94	222300	Wolfram syndrome
ZNF423	124	98	614844	Joubert syndrome 19
ZNF513	93	96	613617	Retinitis pigmentosa 58

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Depth describes the average number of reads seen across 50 exomes
Coverage describes the average coverage of a gene across 50 exomes in percentiles
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013

Ad 1. OMIM identifier 200 signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors