Whole Exome Sequencing
Gene package Vision disorders, version 8, 30-9-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Alissa Interpret 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	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ABCA4	601691	157	100	99	97
ABCB6	605452	217	100	100	100
ABCC6	603234	201	100	100	99
ABHD12	613599	133	100	99	93
ACBD5	616618	104	100	100	97
ACO2	100850	270	100	100	100
ACTB	102630	253	100	100	100
ACTG1	102560	233	100	100	100
ADAM9	602713	75	100	96	92
ADAMTS10	608990	267	100	100	100
ADAMTS17	607511	177	98	96	94
ADAMTS18	607512	136	100	100	99
ADAMTSL4	610113	244	100	100	100
ADGRV1	602851	98	99	94	89
ADIPOR1	607945	101	100	100	99
AGBL1	615496	134	100	100	98
AGBL5	615900	168	100	100	99
AGK	610345	97	100	97	97
AHI1	608894	65	96	86	77
AHR	600253	94	100	98	93
AIPL1	604392	227	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ALDH1A3	600463	129	100	100	99
ALMS1	606844	116	100	99	96
ALX3	606014	202	100	98	96
ANTXR1	606410	116	99	98	95
AP3B1	603401	58	93	79	66
AP3D1	607246	241	99	98	97
ARHGEF18	616432	191	100	100	99
ARL13B	608922	56	100	90	72
ARL2BP	615407	81	100	98	90
ARL3	604695	90	89	89	89
ARL6	608845	44	98	88	79
ARMC9	617612	134	96	93	90
ARR3	301770	91	100	100	100
ARSG	610008	167	100	100	100
ASB10	615054	345	100	100	100
ASPH	600582	86	93	86	81
ASRGL1	609212	115	100	100	98
ATF6	605537	70	99	93	85
ATOH7	609875	231	100	100	100
ATP1A3	182350	221	100	100	100
B3GLCT	610308	64	97	84	73
B9D1	614144	285	100	100	100
B9D2	611951	336	100	100	100
BBIP1	613605	79	100	99	92
BBS1	209901	225	100	100	100
BBS10	610148	100	100	100	99
BBS12	610683	118	100	100	98
BBS2	606151	117	100	99	94
BBS4	600374	100	100	100	93
BBS5	603650	52	91	66	47
BBS7	607590	68	95	82	75
BBS9	607968	73	99	94	88
BCOR	300485	120	100	100	98
BEST1	607854	214	100	100	100
BFSP1	603307	138	100	100	100
BFSP2	603212	268	100	100	100
BLOC1S3	609762	141	100	100	100
BLOC1S6	604310	65	100	78	57
BMP4	112262	224	100	100	100
BMP7	112267	279	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
BSG	109480	228	100	100	100
C12orf57	615140	308	100	100	100
C19orf12	614297	235	100	100	100
C1QTNF5	608752	352	100	100	100
C3	120700	210	100	100	100
C8orf37	614477	124	100	93	85
CA4	114760	236	100	100	100
CABP4	608965	258	100	100	100
CACNA1F	300110	121	100	100	99
CACNA2D4	608171	207	100	100	100
CAPN5	602537	227	100	100	100
CC2D2A	612013	88	98	93	86
CCT2	605139	71	98	86	84
CDH23	605516	257	100	100	100
CDH3	114021	173	100	100	100
CDHR1	609502	176	100	100	100
CEP104	616690	94	100	99	95
CEP120	613446	84	99	94	89
CEP164	614848	149	100	100	100
CEP250	609689	157	100	100	99
CEP290	610142	35	74	58	45
CEP41	610523	83	100	94	91
CEP78	617110	67	98	95	88
CEP83	615847	35	83	64	46
CERKL	608381	82	100	94	81
CFAP410	603191	250	100	100	100
CFB	138470	158	100	100	100
CFH	134370	78	95	89	81
CFI	217030	78	96	92	89
CHD7	608892	167	100	99	97
CHM	300390	47	87	79	69
CHMP4B	610897	116	100	100	100
CHRDL1	300350	73	100	98	90
CHST6	605294	533	100	100	100
CIB2	605564	185	100	100	100
CISD2	611507	97	76	76	76
CLCC1	617539	82	97	89	83
CLDN19	610036	369	100	100	100
CLN3	607042	211	100	100	100
CLN5	608102	113	100	93	89

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
CLN6	606725	264	100	100	100
CLN8	607837	195	100	100	100
CLRN1	606397	92	100	96	92
CLUAP1	616787	73	100	91	79
CNGA1	123825	76	96	89	81
CNGA3	600053	183	100	100	100
CNGB1	600724	133	100	100	98
CNGB3	605080	83	97	91	86
CNNM4	607805	260	100	100	99
COL11A1	120280	86	99	95	89
COL11A2	120290	186	100	100	99
COL18A1	120328	247	100	100	100
COL25A1	610004	105	99	97	94
COL2A1	120140	203	100	100	99
COL8A2	120252	153	100	100	100
COL9A1	120210	99	100	98	96
COL9A2	120260	153	100	100	100
COL9A3	120270	185	100	100	100
CPAMD8	608841	180	100	99	98
CPLANE1	614571	75	97	93	86
CPSF1	606027	282	100	99	99
CRB1	604210	214	100	100	100
CRX	602225	158	100	100	100
CRYAA	123580	511	100	100	100
CRYAB	123590	160	100	100	100
CRYBA1	123610	123	100	100	100
CRYBA2	600836	158	100	100	100
CRYBA4	123631	170	100	100	100
CRYBB1	600929	315	100	100	100
CRYBB2	123620	282	100	100	100
CRYBB3	123630	274	100	100	100
CRYGB	123670	138	100	100	100
CRYGC	123680	201	100	100	100
CRYGD	123690	258	100	100	100
CRYGS	123730	304	100	100	100
CSPP1	611654	68	99	91	80
CTDP1	604927	173	100	100	100
CTNNA1	116805	109	100	100	97
CTNND1	601045	149	100	99	95
CTSD	116840	278	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
CTSH	116820	149	100	100	96
CWC27	617170	46	83	76	59
CYP1B1	601771	364	100	100	100
CYP4V2	608614	105	98	93	88
DCN	125255	113	100	100	100
DHDDS	608172	146	100	100	100
DHX38	605584	194	100	100	99
DKC1	300126	73	100	93	88
DNM1L	603850	61	99	92	82
DNMBP	611282	152	100	100	99
DPYD	612779	73	94	92	86
DRAM2	613360	61	100	100	96
DTHD1	616979	80	100	97	93
DTNBP1	607145	130	100	91	86
EFEMP1	601548	125	100	100	98
ELOVL1	611813	118	100	100	100
ELOVL4	605512	98	98	89	81
ELP4	606985	87	93	80	69
EMC1	616846	147	100	100	99
EPG5	615068	106	100	99	98
EPHA2	176946	284	100	100	100
EXOSC2	602238	107	100	100	100
EYA1	601653	111	95	95	89
EYS	612424	89	100	96	91
FA2H	611026	182	100	100	100
FAM161A	613596	73	98	90	84
FBN1	134797	141	100	100	99
FBN2	612570	139	100	100	99
FLVCR1	609144	132	100	95	85
FOXC1	601090	137	100	96	93
FOXE3	601094	100	93	83	82
FOXL2	605597	274	100	100	100
FRAS1	607830	141	100	99	99
FREM1	608944	123	100	99	96
FREM2	608945	189	100	100	99
FRMD7	300628	80	100	99	93
FSCN2	607643	333	100	100	100
FTL	134790	188	100	100	100
FYCO1	607182	166	100	100	100
FZD4	604579	175	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
GALK1	604313	319	100	100	100
GALM	137030	134	100	100	98
GALT	606999	231	100	100	100
GCNT2	600429	149	100	100	100
GDF3	606522	173	100	100	100
GDF6	601147	147	100	100	100
GFER	600924	168	100	100	100
GJA1	121014	143	100	100	100
GJA3	121015	267	100	100	100
GJA8	600897	207	100	100	100
GLI2	165230	260	100	100	100
GLIS2	608539	245	100	100	100
GNAT1	139330	336	100	100	100
GNAT2	139340	125	100	100	100
GNB3	139130	199	100	100	100
GNPTG	607838	302	100	100	100
GPR143	300808	111	100	100	99
GPR179	614515	401	100	100	100
GRASP	612027	135	98	95	92
GRHL2	608576	120	100	100	98
GRIP1	604597	117	100	100	98
GRK1	180381	270	100	100	100
GRM6	604096	285	100	98	96
GRN	138945	336	100	100	100
GSN	137350	201	100	100	100
GUCA1A	600364	216	100	100	100
GUCA1B	602275	122	100	100	99
GUCY2D	600179	218	100	100	100
HARS1	142810	163	100	100	100
HCCS	300056	64	100	98	90
HGSNAT	610453	93	99	94	92
HK1	142600	158	100	100	99
HMGB3	300193	47	100	100	89
HMX1	142992	94	100	100	100
HPS1	604982	250	100	100	100
HPS3	606118	92	97	91	85
HPS4	606682	365	100	99	98
HPS5	607521	85	100	97	91
HPS6	607522	246	100	100	100
HSF4	602438	224	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
IDH3A	601149	117	99	95	91
IDH3B	604526	244	100	100	100
IFT140	614620	199	100	100	100
IFT172	607386	143	100	100	98
IFT27	615870	101	100	100	99
IFT43	614068	133	100	100	100
IFT74	608040	31	84	52	36
IFT81	605489	37	96	78	54
IGBP1	300139	86	100	98	89
IMPDH1	146690	255	100	100	100
IMPG1	602870	92	93	85	83
IMPG2	607056	106	99	97	94
INPP5E	613037	259	100	100	100
INVS	243305	123	100	99	97
IPO13	610411	200	100	100	98
IQCB1	609237	75	100	99	88
JAM3	606871	129	100	100	100
KCNJ13	603208	143	100	100	100
KCNV2	607604	268	100	100	100
KERA	603288	118	100	100	100
KIAA0556	616650	181	100	100	100
KIAA0586	610178	62	96	86	75
KIAA1549	613344	173	98	97	96
KIF11	148760	46	95	80	64
KIF21A	608283	69	97	91	83
KIF7	611254	224	100	100	98
KIZ	615757	79	98	91	85
KLHL7	611119	89	100	97	88
KRT12	601687	143	100	100	100
KRT3	148043	154	100	100	99
LAMA1	150320	140	100	99	98
LAMB2	150325	282	100	100	100
LCA5	611408	64	95	94	88
LCAT	606967	392	100	100	100
LEMD2	616312	134	100	100	99
LIM2	154045	298	100	100	100
LOXL3	607163	234	100	100	100
LRAT	604863	260	100	100	100
LRIT3	615004	173	100	100	100
LRMDA	614537	148	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
LRP2	600073	118	100	99	97
LRP5	603506	325	98	98	98
LRPAP1	104225	173	100	100	99
LSS	600909	205	100	100	100
LTBP2	602091	241	100	100	100
LYST	606897	87	97	92	89
LZTFL1	606568	43	84	76	70
MAB21L2	604357	511	100	100	100
MAF	177075	206	89	85	82
MAK	154235	81	94	92	88
MAPKAPK3	602130	174	100	100	100
MERTK	604705	123	100	100	96
MFN2	608507	182	100	100	100
MFRP	606227	212	100	100	100
MFSD8	611124	82	99	93	83
MIP	154050	203	100	100	100
MIR184	613146	No coverage data			
MIR204	610942	No coverage data			
MITF	156845	119	100	100	93
MKKS	604896	251	100	100	100
MKS1	609883	157	100	100	100
MLPH	606526	150	100	100	98
MTTP	157147	89	100	100	98
MVK	251170	207	100	100	100
MYO5A	160777	106	96	93	90
MYO7A	276903	214	100	100	100
MYOC	601652	195	100	100	100
NAA10	300013	98	100	100	100
NBAS	608025	103	100	98	94
NDP	300658	155	100	100	100
NEK2	604043	79	100	92	80
NEK8	609799	274	100	100	100
NEUROD1	601724	236	100	100	100
NHS	300457	96	100	99	97
NMNAT1	608700	118	100	100	96
NPHP1	607100	89	97	96	89
NPHP3	608002	89	94	89	87
NPHP4	607215	258	100	100	100
NR2E3	604485	217	100	100	100
NR2F1	132890	208	100	99	96

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
NRL	162080	186	100	100	100
NYX	300278	184	100	100	100
OAT	613349	75	89	83	73
OCA2	611409	152	100	97	96
OCRL	300535	75	100	98	91
OFD1	300170	51	97	80	64
OPA1	605290	55	90	79	67
OPA3	606580	317	100	100	100
OPN1LW	300822	105	89	87	81
OPN1MW	300821	24	53	35	31
OPN1SW	613522	133	100	100	100
OPTN	602432	81	99	90	77
OR2W3	616729	203	100	100	99
OTX2	600037	208	100	100	100
OVOL2	616441	146	100	100	100
P3H2	610341	102	100	100	96
P4HA2	600608	152	100	100	100
PANK2	606157	117	100	99	97
PAX2	167409	211	100	100	100
PAX6	607108	267	100	100	100
PCARE	613425	861	100	100	100
PCDH15	605514	152	99	97	95
PCYT1A	123695	126	100	100	99
PDE6A	180071	174	100	99	95
PDE6B	180072	237	100	100	100
PDE6C	600827	82	96	84	76
PDE6D	602676	118	100	97	84
PDE6G	180073	229	100	100	100
PDE6H	601190	93	100	100	100
PDZD7	612971	247	100	100	100
PEX1	602136	80	95	94	90
PEX2	170993	93	100	100	100
PEX7	601757	94	100	100	94
PGK1	311800	89	100	100	95
PHOX2A	602753	163	100	100	100
PHYH	602026	99	100	97	87
PIGL	605947	159	100	100	100
PIK3R1	171833	65	100	93	82
PIKFYVE	609414	93	99	96	92
PITPNM3	608921	213	100	99	98

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
PITX2	601542	197	100	100	97
PITX3	602669	225	100	100	100
PLA2G5	601192	212	100	100	100
PLK4	605031	77	98	90	83
PNPLA6	603197	269	100	100	100
POC1B	614784	68	100	93	88
POC5	617880	71	96	85	78
POMGNT1	606822	228	100	100	100
PPT1	600722	145	100	100	100
PRCD	610598	137	100	100	100
PRDM13	616741	269	100	100	100
PRDM5	614161	96	95	90	85
PRIMPOL	615421	41	92	76	60
PROM1	604365	67	95	88	82
PRPF3	607301	74	100	100	96
PRPF31	606419	249	100	100	100
PRPF4	607795	121	100	97	93
PRPF6	613979	186	100	100	100
PRPF8	607300	159	100	100	100
PRPH2	179605	418	100	100	100
PRSS56	613858	203	100	100	100
PXDN	605158	267	99	96	96
RAB28	612994	53	85	72	58
RAB3GAP2	609275	96	97	91	81
RARB	180220	98	100	98	92
RAX	601881	198	100	100	100
RAX2	610362	192	100	100	97
RBP3	180290	455	100	100	100
RBP4	180250	281	100	100	100
RCBTB1	607867	129	100	100	96
RD3	180040	148	100	100	100
RDH11	607849	132	100	100	99
RDH12	608830	147	100	100	100
RDH5	601617	283	100	100	100
REEP6	609346	245	100	100	100
RGR	600342	138	100	100	100
RGS9	604067	162	100	100	98
RGS9BP	607814	268	100	100	100
RHO	180380	270	100	100	100
RIMS1	606629	110	100	97	92

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
RLBP1	180090	205	100	100	100
ROM1	180721	171	100	100	100
RP1	603937	109	100	99	95
RP1L1	608581	188	100	99	96
RP2	300757	71	100	100	88
RP9	607331	95	100	100	100
RPE65	180069	92	100	100	96
RPGR	312610	35	70	62	51
RPGRIP1	605446	119	100	99	97
RPGRIP1L	610937	80	95	91	83
RS1	300839	110	100	100	94
RTN4IP1	610502	107	100	100	100
SAG	181031	133	100	100	100
SALL2	602219	212	100	100	100
SALL4	607343	229	100	100	100
SAMD11	616765	192	100	100	100
SC5D	602286	79	100	98	90
SCAPER	611611	80	97	93	86
SCO2	604272	377	100	100	100
SDCCAG8	613524	66	100	91	78
SEMA4A	607292	197	100	100	100
SGSH	605270	272	99	94	94
SHH	600725	259	100	100	100
SIPA1L3	616655	257	100	100	100
SIX3	603714	279	100	100	100
SIX6	606326	241	100	100	100
SLC16A12	611910	97	100	100	98
SLC24A1	603617	139	100	100	99
SLC24A5	609802	76	96	84	73
SLC25A46	610826	103	96	90	82
SLC33A1	603690	147	98	93	85
SLC38A8	615585	213	100	100	100
SLC39A5	608730	180	100	100	100
SLC45A2	606202	127	100	100	97
SLC4A11	610206	304	100	100	100
SLC52A2	607882	210	100	100	100
SLC7A14	615720	166	100	100	100
SMOC1	608488	133	100	99	97
SNRNP200	601664	164	100	100	100
SOX2	184429	257	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
SOX5	604975	117	100	100	98
SPATA7	609868	114	88	81	77
SPP2	602637	124	100	100	100
SRD5A3	611715	138	100	100	100
STRA6	610745	229	100	100	100
SUFU	607035	206	100	100	100
TACSTD2	137290	617	100	100	100
TCF4	602272	145	100	99	97
TCOF1	606847	201	100	100	98
TCTN1	609863	117	98	97	95
TCTN2	613846	108	100	95	93
TCTN3	613847	126	100	100	98
TDRD7	611258	94	97	92	87
TEAD1	189967	116	100	100	100
TEK	600221	110	100	99	96
TENM3	610083	151	100	100	99
TFAP2A	107580	236	100	100	100
TGFBI	601692	153	100	100	100
TGIF1	602630	133	100	100	100
TIMM8A	300356	98	100	100	100
TIMP3	188826	203	100	100	100
TMEM107	616183	136	100	100	100
TMEM126A	612988	66	100	93	77
TMEM138	614459	110	100	100	100
TMEM216	613277	142	100	100	100
TMEM231	614949	271	100	100	98
TMEM237	614423	77	98	98	95
TMEM67	609884	61	90	74	62
TMEM98	615949	161	100	100	100
TOPORS	609507	125	100	100	100
TPP1	607998	223	100	100	100
TRAF3IP1	607380	80	99	90	81
TREX1	606609	313	100	100	100
TRIM32	602290	222	100	100	100
TRNT1	612907	47	90	86	74
TRPM1	603576	140	100	99	95
TSPAN12	613138	96	100	98	90
TTC8	608132	104	96	91	86
TTLL5	612268	92	100	100	97
TUB	601197	182	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
TUBA3D	617878	273	100	100	100
TUBB3	602661	330	100	100	97
TUBB4B	602660	252	100	100	100
TUBGCP4	609610	122	100	100	98
TUBGCP6	610053	356	100	100	100
TULP1	602280	172	100	97	92
TYR	606933	147	100	99	98
TYRP1	115501	137	100	100	100
UBIAD1	611632	203	100	100	100
UNC119	604011	211	100	100	100
UNC45B	611220	203	100	100	100
USH1C	605242	209	100	100	99
USH1G	607696	427	100	100	100
USH2A	608400	116	100	99	97
USP45	618439	56	92	76	66
VAX1	604294	125	97	92	89
VCAN	118661	128	100	100	99
VIM	193060	138	100	99	96
VSX1	605020	136	100	99	98
VSX2	142993	227	100	100	100
WASHC5	610657	82	100	94	87
WDPCP	613580	87	98	91	82
WDR19	608151	75	100	96	92
WDR36	609669	83	98	91	84
WFS1	606201	268	100	100	100
WHRN	607928	288	100	100	100
WRN	604611	62	95	87	77
YAP1	606608	104	100	100	97
YME1L1	607472	68	93	88	80
ZEB1	189909	99	99	94	86
ZNF408	616454	276	100	100	100
ZNF423	604557	316	100	100	100
ZNF469	612078	331	100	100	100
ZNF513	613598	344	100	100	100
ZNF644	614159	86	100	99	97

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