Whole Exome Sequencing
Gene package Vision disorders, version 7, 21-2-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

Erasmus MC
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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	94	100	100	97
ABCB6	605452	110	100	100	100
ABCC6	603234	118	100	100	99
ABHD12	613599	61	100	100	94
ACBD5	616618	85	100	99	89
ACO2	100850	146	100	98	94
ACTB	102630	192	100	100	100
ACTG1	102560	188	100	100	100
ADAM9	602713	57	100	99	92
ADAMTS10	608990	112	100	100	100
ADAMTS17	607511	100	97	95	92
ADAMTS18	607512	79	100	100	98
ADAMTSL4	610113	115	100	100	99
ADGRV1	602851	80	100	100	97
ADIPOR1	607945	100	100	100	100
AGBL1	615496	74	98	98	97
AGBL5	615900	90	100	99	96
AGK	610345	57	100	100	93
AHI1	608894	67	100	98	90
AHR	600253	63	100	100	95

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
AIPL1	604392	141	100	100	100
ALDH1A3	600463	88	100	100	97
ALMS1	606844	100	100	100	99
ALX3	606014	116	98	91	86
ANTXR1	606410	60	99	97	92
AP3B1	603401	66	100	98	87
AP3D1	607246	103	99	98	97
ARHGEF18	616432	123	100	99	93
ARL13B	608922	63	100	100	94
ARL2BP	615407	57	100	100	96
ARL3	604695	111	100	100	100
ARL6	608845	43	96	95	83
ARMC9	617612	76	100	98	93
ARR3	301770	62	100	100	96
ARSG	610008	88	100	100	97
ASPH	600582	63	100	99	92
ASRGL1	609212	72	100	98	93
ATF6	605537	65	100	100	97
ATOH7	609875	131	95	88	85
ATP1A3	182350	144	100	100	100
B3GLCT	610308	67	100	99	85
B9D1	614144	90	100	100	100
B9D2	611951	82	100	100	100
BBIP1	613605	45	100	96	86
BBS1	209901	126	100	100	100
BBS10	610148	66	100	100	100
BBS12	610683	59	100	100	99
BBS2	606151	77	100	100	97
BBS4	600374	76	100	100	95
BBS5	603650	65	100	98	89
BBS7	607590	64	100	99	95
BBS9	607968	59	96	95	90
BCOR	300485	78	100	99	94
BEST1	607854	94	100	99	98
BFSP1	603307	98	100	100	96
BFSP2	603212	73	100	99	92
BLOC1S3	609762	104	100	100	100
BLOC1S6	604310	46	100	100	84
BMP4	112262	103	100	96	93

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BMP7	112267	116	100	100	100
BSG	109480	107	100	100	99
C12orf57	615140	141	100	100	100
C19orf12	614297	186	100	100	100
C1QTNF5	608752	102	100	100	93
C3	120700	127	100	100	99
C8orf37	614477	78	100	100	96
CA4	114760	100	100	100	100
CABP4	608965	149	100	100	100
CACNA1F	300110	76	100	100	96
CACNA2D4	608171	86	100	100	98
CAPN5	602537	143	100	100	99
CC2D2A	612013	66	100	100	95
CDH23	605516	135	100	100	100
CDH3	114021	124	100	100	100
CDHR1	609502	116	100	100	98
CEP104	616690	71	100	98	93
CEP120	613446	75	100	100	96
CEP164	614848	93	100	100	98
CEP250	609689	73	100	99	95
CEP290	610142	71	100	98	89
CEP41	610523	69	100	100	94
CEP78	617110	69	100	100	94
CEP83	615847	60	100	96	81
CERKL	608381	73	100	99	94
CFAP410	603191	141	100	100	100
CFB	138470	106	100	100	100
CFH	134370	78	100	100	96
CFI	217030	67	100	99	93
CHD7	608892	87	100	100	97
CHM	300390	50	100	94	80
CHMP4B	610897	178	100	100	100
CHRDL1	300350	48	100	100	92
CHST6	605294	246	100	100	100
CIB2	605564	195	100	100	100
CISD2	611507	137	100	100	100
CLCC1	617539	55	100	96	80
CLDN19	610036	149	100	100	100
CLN3	607042	111	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
CLN5	608102	70	100	100	100
CLN6	606725	120	100	100	99
CLN8	607837	129	100	100	100
CLRN1	606397	83	100	100	97
CLUAP1	616787	58	100	99	89
CNGA1	123825	62	96	91	86
CNGA3	600053	118	100	100	100
CNGB1	600724	112	100	100	99
CNGB3	605080	62	100	100	95
CNNM4	607805	140	100	100	99
COL11A1	120280	62	100	99	93
COL11A2	120290	110	100	100	100
COL18A1	120328	143	100	100	97
COL25A1	610004	73	100	98	91
COL2A1	120140	97	100	100	99
COL8A2	120252	95	100	100	98
COL9A1	120210	65	100	99	91
COL9A2	120260	100	100	99	96
CPAMD8	608841	84	98	94	88
CPLANE1	614571	71	100	99	95
CRB1	604210	71	100	100	99
CRX	602225	196	100	100	100
CRYAA	123580	136	100	100	100
CRYAB	123590	79	100	100	100
CRYBA1	123610	74	100	100	100
CRYBA2	600836	113	100	100	100
CRYBA4	123631	106	100	100	100
CRYBB1	600929	97	100	100	100
CRYBB2	123620	181	100	100	100
CRYBB3	123630	105	100	100	100
CRYGB	123670	87	100	100	100
CRYGC	123680	86	100	100	100
CRYGD	123690	97	100	100	100
CRYGS	123730	104	100	100	100
CSPP1	611654	83	100	100	97
CTDP1	604927	125	98	89	87
CTNNA1	116805	93	100	100	96
CTNND1	601045	74	100	100	96
CTSD	116840	137	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	92	100	100	96
CWC27	617170	51	100	96	84
CYP1B1	601771	97	100	100	100
CYP4V2	608614	82	100	100	98
DCN	125255	69	100	100	100
DHDDS	608172	69	100	100	99
DHX38	605584	100	100	100	100
DKC1	300126	53	100	98	89
DNM1L	603850	70	100	99	94
DNMBP	611282	81	100	99	94
DPYD	612779	66	100	99	95
DRAM2	613360	53	100	99	92
DTHD1	616979	81	100	100	99
DTNBP1	607145	94	100	100	96
EFEMP1	601548	99	100	100	99
ELOVL1	611813	82	100	99	96
ELOVL4	605512	71	100	100	98
ELP4	606985	66	98	91	83
EMC1	616846	117	100	100	99
EPG5	615068	69	100	100	97
EPHA2	176946	141	100	100	99
EXOSC2	602238	69	100	100	97
EYA1	601653	86	100	100	98
EYS	612424	72	100	100	96
FA2H	611026	83	100	99	91
FAM161A	613596	64	100	100	96
FBN1	134797	186	100	100	100
FBN2	612570	74	100	100	97
FLVCR1	609144	98	100	100	96
FOXC1	601090	80	100	95	89
FOXE3	601094	74	87	79	74
FOXL2	605597	114	100	94	88
FRAS1	607830	79	100	100	97
FREM1	608944	83	100	100	98
FREM2	608945	104	100	100	100
FRMD7	300628	50	100	98	89
FSCN2	607643	139	100	100	100
FTL	134790	144	100	100	100
FYCO1	607182	115	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
FZD4	604579	90	100	100	99
GALK1	604313	133	100	100	99
GALT	606999	151	100	100	100
GCNT2	600429	81	100	100	98
GDF3	606522	120	100	100	100
GDF6	601147	124	100	100	100
GFER	600924	86	100	100	100
GJA1	121014	101	100	100	100
GJA3	121015	138	100	100	100
GJA8	600897	145	100	100	100
GLI2	165230	163	100	100	99
GLIS2	608539	117	100	100	100
GNAT1	139330	166	100	100	100
GNAT2	139340	72	100	100	98
GNB3	139130	118	100	100	100
GNPTG	607838	153	100	99	94
GPR143	300808	43	100	89	76
GPR179	614515	135	100	100	100
GRASP	612027	75	94	87	79
GRHL2	608576	81	100	100	99
GRIP1	604597	84	100	100	99
GRK1	180381	114	100	100	99
GRM6	604096	127	100	95	90
GRN	138945	155	100	100	100
GSN	137350	95	100	100	99
GUCA1A	600364	108	100	100	100
GUCA1B	602275	156	100	100	100
GUCY2D	600179	112	100	100	99
HARS1	142810	104	100	100	99
HCCS	300056	45	100	99	84
HGSNAT	610453	70	94	94	92
HK1	142600	102	100	100	99
HMGB3	300193	40	90	90	79
HMX1	142992	38	94	72	49
HPS1	604982	103	100	100	97
HPS3	606118	66	100	100	95
HPS4	606682	103	100	100	98
HPS5	607521	68	100	100	97
HPS6	607522	130	100	100	93

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
HSF4	602438	114	100	100	97
IDH3A	601149	67	100	99	95
IDH3B	604526	117	100	100	100
IFT140	614620	113	100	100	98
IFT172	607386	75	100	100	96
IFT27	615870	84	100	100	99
IFT43	614068	75	100	100	100
IFT74	608040	46	100	96	83
IFT81	605489	47	99	94	82
IGBP1	300139	69	100	100	90
IMPDH1	146690	99	100	96	91
IMPG1	602870	64	100	99	93
IMPG2	607056	68	100	100	97
INPP5E	613037	113	100	100	97
INVS	243305	81	100	100	97
IPO13	610411	108	100	100	98
IQCB1	609237	54	100	94	78
JAM3	606871	70	100	100	97
KCNJ13	603208	73	100	100	99
KCNV2	607604	130	100	100	100
KERA	603288	61	100	100	98
KIAA0556	616650	97	100	99	97
KIAA0586	610178	72	100	98	94
KIF11	148760	73	100	99	93
KIF21A	608283	68	100	99	93
KIF7	611254	107	98	96	93
KIZ	615757	69	100	100	96
KLHL7	611119	70	100	100	98
KRT12	601687	94	100	100	100
KRT3	148043	134	100	100	99
LAMA1	150320	87	100	100	97
LAMB2	150325	153	100	100	100
LCA5	611408	74	100	100	99
LEMD2	616312	99	100	99	95
LIM2	154045	120	100	100	100
LOXL3	607163	140	100	100	100
LRAT	604863	159	100	100	100
LRIT3	615004	78	100	100	100
LRMDA	614537	91	100	95	93

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
LRP2	600073	69	100	100	96
LRP5	603506	146	100	99	98
LRPAP1	104225	135	100	100	100
LSS	600909	91	100	100	96
LTBP2	602091	106	100	100	99
LYST	606897	72	100	99	95
LZTFL1	606568	79	100	100	93
MAB21L2	604357	170	100	100	100
MAF	177075	80	84	80	76
MAK	154235	82	100	100	97
MAPKAPK3	602130	73	100	100	97
MERTK	604705	97	100	100	96
MFN2	608507	114	100	100	98
MFRP	606227	100	100	100	100
MFSD8	611124	68	100	100	95
MIP	154050	123	100	100	100
MIR184	613146	No coverage data			
MIR204	610942	No coverage data			
MITF	156845	87	100	100	99
MKKS	604896	80	100	100	100
MKS1	609883	117	100	100	99
MLPH	606526	107	100	100	100
MTTP	157147	67	100	100	97
MVK	251170	100	100	100	100
MYO5A	160777	71	100	99	94
MYO7A	276903	109	100	100	99
MYOC	601652	140	100	100	100
NAA10	300013	89	100	100	94
NBAS	608025	66	100	99	94
NDP	300658	77	100	100	100
NEK2	604043	84	100	98	84
NEK8	609799	132	100	100	100
NEUROD1	601724	115	100	100	100
NHS	300457	57	100	96	90
NMNAT1	608700	90	100	100	98
NPHP1	607100	58	100	98	88
NPHP3	608002	66	100	99	94
NPHP4	607215	110	100	100	100
NR2E3	604485	123	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
NR2F1	132890	197	100	100	100
NRL	162080	108	100	100	100
NYX	300278	132	100	100	100
OAT	613349	64	100	96	85
OCA2	611409	93	100	100	97
OCRL	300535	43	100	95	80
OFD1	300170	43	100	95	75
OPA1	605290	59	100	98	89
OPA3	606580	130	100	100	100
OPN1LW	300822	121	89	89	88
OPN1MW	300821	47	54	45	42
OPN1SW	613522	78	100	100	100
OPTN	602432	81	100	100	96
OR2W3	616729	140	100	100	100
OTX2	600037	100	100	100	100
OVOL2	616441	107	100	99	94
P3H2	610341	65	100	100	95
P4HA2	600608	101	100	100	98
PANK2	606157	81	100	100	99
PAX2	167409	144	100	100	98
PAX6	607108	73	100	100	96
PCARE	613425	105	100	100	98
PCDH15	605514	72	100	100	95
PCYT1A	123695	74	100	99	85
PDE6A	180071	74	100	100	96
PDE6B	180072	125	100	100	100
PDE6C	600827	67	100	100	96
PDE6D	602676	86	100	100	100
PDE6G	180073	147	100	100	100
PDE6H	601190	47	100	100	96
PDZD7	612971	84	100	100	99
PEX1	602136	62	100	99	94
PEX2	170993	63	100	100	100
PEX7	601757	57	100	100	95
PGK1	311800	47	100	100	89
PHOX2A	602753	61	100	100	86
PHYH	602026	121	100	100	94
PIGL	605947	98	100	100	100
PIK3R1	171833	80	100	100	97

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
PIKFYVE	609414	67	100	99	95
PITPNM3	608921	99	100	99	97
PITX2	601542	149	100	100	100
PITX3	602669	96	100	100	100
PLA2G5	601192	89	100	100	100
PLK4	605031	62	100	99	91
PNPLA6	603197	123	100	100	99
POC1B	614784	64	100	98	83
POC5	617880	57	100	99	91
POMGNT1	606822	93	100	100	99
PPT1	600722	84	100	100	100
PRCD	610598	83	100	100	100
PRDM13	616741	143	100	100	95
PRDM5	614161	76	100	100	96
PRIMPOL	615421	49	95	89	78
PROM1	604365	84	100	99	93
PRPF3	607301	77	100	98	94
PRPF31	606419	101	100	100	99
PRPF4	607795	83	100	100	94
PRPF6	613979	108	100	100	99
PRPF8	607300	102	100	100	98
PRPH2	179605	162	100	100	100
PRSS56	613858	95	100	100	100
PXDN	605158	108	100	100	98
RAB28	612994	55	100	100	92
RAB3GAP2	609275	66	100	99	92
RARB	180220	85	100	100	99
RAX	601881	124	100	100	93
RAX2	610362	78	100	100	100
RBP3	180290	145	100	100	100
RBP4	180250	128	100	100	100
RCBTB1	607867	98	100	100	99
RD3	180040	153	100	100	100
RDH11	607849	82	100	100	99
RDH12	608830	104	100	100	100
RDH5	601617	149	100	100	100
REEP6	609346	174	100	100	99
RGR	600342	101	100	98	91
RGS9	604067	108	100	100	98

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RGS9BP	607814	153	100	100	100
RHO	180380	131	100	100	100
RIMS1	606629	94	100	100	98
RLBP1	180090	111	100	100	100
ROM1	180721	110	100	100	100
RP1	603937	73	100	100	97
RP1L1	608581	165	100	100	100
RP2	300757	67	100	100	98
RP9	607331	44	92	78	72
RPE65	180069	72	100	100	98
RPGR	312610	39	81	72	60
RPGRIP1	605446	88	100	100	96
RPGRIP1L	610937	60	98	96	89
RS1	300839	44	100	99	74
RTN4IP1	610502	50	100	96	89
SAG	181031	97	100	100	98
SALL2	602219	121	100	100	100
SALL4	607343	155	100	100	97
SAMD11	616765	92	94	87	79
SC5D	602286	88	100	100	100
SCAPER	611611	57	100	98	89
SCO2	604272	125	100	100	100
SDCCAG8	613524	92	100	100	96
SEMA4A	607292	108	100	100	100
SGSH	605270	103	100	94	90
SHH	600725	119	100	100	99
SIPA1L3	616655	149	100	99	97
SIX3	603714	158	100	100	96
SIX6	606326	233	100	100	100
SLC16A12	611910	89	100	100	100
SLC24A1	603617	105	100	100	99
SLC24A5	609802	77	100	100	100
SLC25A46	610826	70	100	100	94
SLC33A1	603690	68	100	98	86
SLC38A8	615585	83	100	99	94
SLC39A5	608730	131	100	100	100
SLC45A2	606202	93	100	100	97
SLC4A11	610206	125	100	100	99
SLC52A2	607882	173	100	100	100

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SLC7A14	615720	112	100	100	100
SMOC1	608488	106	100	100	97
SNRNP200	601664	102	100	100	98
SOX2	184429	183	100	100	100
SOX5	604975	78	100	100	96
SPATA7	609868	62	100	99	94
SPP2	602637	73	100	100	100
SRD5A3	611715	111	100	100	96
STRA6	610745	98	100	100	100
SUFU	607035	104	100	100	100
TACSTD2	137290	228	100	100	100
TCOF1	606847	115	100	100	99
TCTN1	609863	94	100	100	98
TCTN2	613846	84	100	100	97
TCTN3	613847	66	100	100	97
TDRD7	611258	70	100	100	98
TEAD1	189967	82	100	100	98
TENM3	610083	86	100	100	99
TFAP2A	107580	95	100	100	100
TGFBI	601692	82	100	100	98
TGIF1	602630	152	100	100	100
TIMM8A	300356	138	100	100	100
TIMP3	188826	104	100	100	100
TMEM107	616183	100	100	100	94
TMEM126A	612988	68	100	100	93
TMEM138	614459	54	100	100	100
TMEM216	613277	87	100	100	95
TMEM231	614949	96	100	95	90
TMEM237	614423	57	100	98	87
TMEM67	609884	77	100	100	94
TOPORS	609507	73	100	100	100
TPP1	607998	97	100	100	100
TRAF3IP1	607380	55	100	97	86
TREX1	606609	234	100	100	100
TRIM32	602290	107	100	100	100
TRNT1	612907	68	100	99	92
TRPM1	603576	85	100	100	99
TSPAN12	613138	67	100	100	92
TTC8	608132	66	100	100	93

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
TTLL5	612268	77	100	100	98
TUB	601197	123	100	100	100
TUBA3D	617878	198	100	100	100
TUBB3	602661	269	100	99	95
TUBB4B	602660	188	100	100	100
TUBGCP4	609610	71	100	98	94
TULP1	602280	108	100	100	100
TYR	606933	83	100	100	98
TYRP1	115501	73	100	100	98
UBIAD1	611632	173	100	100	100
UNC119	604011	97	100	100	100
UNC45B	611220	99	100	100	100
USH1C	605242	80	100	98	92
USH1G	607696	180	100	100	100
USH2A	608400	82	100	100	98
USP45	618439	48	100	94	80
VAX1	604294	104	97	91	87
VCAN	118661	67	100	100	99
VIM	193060	106	100	100	96
VSX1	605020	87	100	100	99
VSX2	142993	96	100	100	100
WASHC5	610657	62	100	100	94
WDPCP	613580	72	100	99	93
WDR19	608151	71	100	100	97
WFS1	606201	196	100	100	100
WHRN	607928	119	100	100	100
WRN	604611	63	100	100	95
YAP1	606608	73	100	99	94
YME1L1	607472	48	100	95	83
ZEB1	189909	84	100	100	98
ZNF408	616454	127	100	100	100
ZNF423	604557	166	100	100	100
ZNF469	612078	169	100	100	100
ZNF513	613598	121	100	100	100
ZNF644	614159	63	100	100	99

- 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