

Whole Exome Sequencing

Gene package Vision disorders, version 11, 25-2-2022



Technical information

DNA was enriched using the Agilent SureSelectXT Human All Exon V7 capture kit and paired-end sequenced on the Illumina platform (outsourced). Sequencing data are demultiplexed with bcl2fastq2 Conversion Software from Illumina. Illumina DRAGEN Bio-IT Platform is used for read mapping to the hg19 genome and sequence variant detection. The detected sequence variants are annotated and filtered with Alissa Interpret software and classified with Alamut Visual. Copy number variant detection is performed using the BAM multiscale reference method using depth of coverage analysis and dynamical bins in NexusClinical. The detected copy number variants are annotated and filtered with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). The sensitivity to detect variants using this technology is not 100%; pathogenic variants could be missed. 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).



Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
ABCA4	601691	100	100	99.28	94.51
ABCB6	605452	100	100	100	98.31
ABCC6	603234	99.07	97.66	95.46	80.54
ABHD12	613599	100	96.57	91.81	82.57
ACBD5	616618	100	99.32	97.67	95.81
ACO2	100850	100	100	100	100
ACTB	102630	100	100	100	99.18
ACTG1	102560	100	100	100	100
ADAM9	602713	100	100	98.72	94.46
ADAMTS10	608990	100	100	99.84	93.42
ADAMTS17	607511	95.41	90.63	86.38	82.36
ADAMTS18	607512	100	98.91	97.67	94.94
ADAMTSL4	610113	100	99.01	98.00	91.41
ADGRV1	602851	100	100	99.73	97.56
ADIPOR1	607945	100	100	98.26	86.28
AFG3L2	604581	95.13	91.50	91.50	90.50
AGBL1	615496	100	100	99.75	97.16
AGBL5	615900	100	99.64	98.49	95.93
AGK	610345	100	100	100	100
AHI1	608894	100	100	100	99.41
AHR	600253	100	100	100	99.53
AIPL1	604392	100	100	100	97.40
ALDH18A1	138250	100	100	100	98.06
ALDH1A3	600463	100	99.78	95.41	85.77

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ALMS1	606844	99.89	99.40	98.78	96.05
ALX3	606014	92.43	80.12	73.22	73.13
ANTXR1	606410	98.59	97.32	96.05	88.40
AP3B1	603401	99.05	96.39	94.98	89.23
AP3D1	607246	97.71	97.64	95.96	89.40
ARHGEF18	616432	100	98.26	94.91	87.02
ARL13B	608922	100	99.46	98.38	87.50
ARL2	601175	100	100	95.55	81.60
ARL2BP	615407	100	100	100	100
ARL3	604695	100	99.40	89.34	85.89
ARL6	608845	100	100	100	95.81
ARMC9	617612	100	99.93	98.64	90.94
ARR3	301770	100	100	100	93.80
ARSG	610008	100	100	99.03	94.60
ASB10	615054	100	100	100	98.90
ASPH	600582	100	100	100	97.55
ASRGL1	609212	100	100	96.70	85.73
ATF6	605537	100	100	98.45	91.63
ATOH7	609875	100	99.79	90.02	80.46
ATP1A3	182350	100	100	99.45	94.42
B3GLCT	610308	100	99.36	94.98	94.20
B9D1	614144	100	100	100	97.19
B9D2	611951	100	100	100	100
BBIP1	613605	100	100	100	100
BBS1	209901	100	100	100	98.56
BBS10	610148	100	100	99.41	97.96
BBS12	610683	100	100	100	100
BBS2	606151	100	100	100	100
BBS4	600374	100	100	100	96.00
BBS5	603650	100	100	100	99.01
BBS7	607590	100	100	99.68	97.21
BBS9	607968	99.97	99.87	99.08	94.25
BCOR	300485	100	100	99.96	98.12
BEST1	607854	100	100	100	98.33
BFSP1	603307	100	100	99.91	91.51
BFSP2	603212	100	100	100	100
BLOC1S3	609762	100	100	91.37	52.16
BLOC1S5	607289	100	100	98.18	77.46
BLOC1S6	604310	100	100	96.91	78.29
BMP4	112262	100	100	99.10	95.74
BMP7	112267	100	100	100	98.01
BSG	109480	100	98.20	92.50	85.96
C12orf57	615140	100	100	100	100
C19ORF12	614297	No coverage data			

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C1QTNF5	608752	100	89.27	79.65	68.14
C3	120700	100	100	99.30	96.05
CA4	114760	100	100	99.64	96.85
CABP4	608965	100	100	98.62	89.31
CACNA1F	300110	100	99.89	97.66	89.82
CACNA2D4	608171	100	100	99.89	95.40
CAPN5	602537	100	100	99.63	93.36
CBS	613381	100	100	100	100
CC2D2A	612013	100	99.84	98.65	91.30
CCT2	605139	100	100	99.64	93.97
CDH15	114019	100	98.70	94.82	79.13
CDH2	114020	99.13	97.37	96.07	90.44
CDH23	605516	100	100	100	89.21
CDHR1	609502	98.66	97.43	97.43	94.26
CEP104	603464	100	100	99.53	96.98
CDK10	616690	100	100	99.55	92.43
CEP120	613446	100	100	100	97.66
CEP164	614848	100	99.52	97.04	86.58
CEP250	609689	100	99.89	98.59	90.04
CEP290	610142	95.56	90.87	87.25	80.13
CEP41	610523	100	100	100	99.55
CEP78	617110	100	99.96	99.16	94.59
CEP83	615847	100	97.51	90.22	80.44
CERKL	608381	100	100	100	98.82
CFAP410	603191	100	100	99.67	86.95
CFAP418	614477	No coverage data			
CFB	138470	100	100	100	99.47
CFH	134370	100	99.10	96.82	92.91
CFI	217030	86.45	84.36	82.41	78.74
CHD7	608892	100	100	99.89	98.69
CHM	300390	91.63	91.63	91.63	91.63
CHMP4B	610897	100	100	100	89.38
CHRDL1	300350	100	100	100	95.95
CHST6	605294	100	100	100	100
CIB2	605564	100	100	100	98.38
CISD2	611507	88.49	76.99	76.99	76.99
CLCC1	617539	100	100	99.41	95.90
CLDN19	610036	100	100	100	100
CLN3	607042	100	98.64	91.76	88.17
CLN5	608102	93.14	93.14	88.00	73.62
CLN6	606725	100	100	100	96.55
CLN8	607837	100	100	100	100
CLRN1	606397	100	100	100	100
CLUAP1	616787	100	100	100	91.03

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CNGA1	123825	100	100	100	98.74
CNGA3	600053	100	100	100	99.14
CNGB1	600724	99.99	97.15	91.54	74.41
CNGB3	605080	100	100	100	95.66
CNNM4	607805	99.72	98.54	97.83	95.82
COA8	616003	100	100	100	100
COL11A1	120280	100	99.21	97.82	88.98
COL11A2	120290	100	99.90	95.62	84.23
COL18A1	120328	100	99.84	98.50	87.55
COL25A1	610004	100	100	99.96	94.54
COL2A1	120140	100	100	99.42	93.30
COL4A1	120130	99.39	97.25	93.74	85.37
COL8A2	120252	100	99.49	96.93	74.45
COL9A1	120210	100	100	98.08	86.28
COL9A2	120260	100	96.25	87.33	63.80
COL9A3	120270	100	100	96.30	79.94
CPAMD8	608841	99.82	97.84	94.26	82.89
CPLANE1	614571	100	99.80	98.38	95.82
CPSF1	606027	99.09	98.55	97.89	92.60
CRB1	604210	100	100	100	100
CRX	602225	100	100	99.48	93.73
CRYAA	123580	100	100	100	100
CRYAB	123590	100	100	100	100
CRYBA1	123610	100	100	100	100
CRYBA2	600836	100	100	98.14	78.99
CRYBA4	123631	100	100	99.71	88.37
CRYBB1	600929	100	100	100	97.96
CRYBB2	123620	100	100	100	100
CRYBB3	123630	100	100	100	100
CRYGB	123670	100	99.15	95.21	88.55
CRYGC	123680	100	100	100	100
CRYGD	123690	100	100	100	97.42
CRYGS	123730	100	100	100	100
CSPP1	611654	100	99.29	96.27	91.41
CTDP1	604927	100	97.04	89.34	81.24
CTNNA1	116805	100	99.92	98.92	94.80
CTNND1	601045	100	100	100	98.14
CTSD	116840	100	93.79	93.79	90.47
CTSH	116820	100	100	100	96.54
CWC27	617170	97.94	89.68	89.56	79.69
CYP1B1	601771	100	100	100	100
CYP4V2	608614	100	100	100	97.99
DCN	125255	100	100	100	99.65
DCT	191275	100	100	100	100

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DDHD1	614603	99.93	98.53	96.74	94.76
DHDDS	608172	100	100	100	93.45
DHX38	605584	100	99.73	98.16	90.60
DKC1	300126	100	100	98.05	93.43
DNAJC30	618202	100	100	100	100
DNM1L	603850	100	100	99.23	93.91
DNMBP	611282	100	100	100	99.60
DPYD	612779	100	100	100	100
DRAM2	613360	100	100	100	99.89
DTHD1	616979	100	98.44	96.57	94.95
DTNBP1	607145	100	100	100	96.72
DYNC2I2	613363	100	100	98.01	84.76
EFEMP1	601548	100	100	100	99.82
ELOVL1	611813	100	100	100	97.85
ELOVL4	605512	100	99.81	98.78	92.80
ELP4	606985	95.23	92.17	90.41	87.29
EMC1	616846	99.16	99.16	99.16	97.72
EPG5	615068	100	99.90	99.27	95.85
EPHA2	176946	100	100	99.82	94.68
EXOSC2	602238	100	100	100	98.44
EYA1	601653	97.62	97.62	97.62	95.91
EYS	612424	100	100	99.71	94.99
FA2H	611026	100	96.58	89.53	72.53
FAM161A	613596	100	99.96	98.27	93.49
FBN2	134797	100	100	100	99.22
FBN1	612570	100	100	100	99.38
FLVCR1	609144	100	100	99.46	94.13
FOXC1	601090	96.31	91.19	86.78	75.61
FOXE3	601094	82.50	73.08	63.20	37.82
FOXL2	605597	100	94.43	90.64	80.31
FRAS1	607830	100	99.87	99.03	94.06
FREM1	608944	100	100	100	98.80
FREM2	608945	100	100	99.91	98.87
FSCN2	300628	100	100	100	98.85
FRMD7	607643	100	100	100	100
FTL	134790	100	100	95.12	84.71
FYCO1	607182	100	100	100	97.98
FZD4	604579	100	100	100	99.61
GALK1	604313	100	100	100	96.78
GALM	137030	100	100	100	99.74
GALT	606999	100	100	100	97.01
GCNT2	600429	100	100	100	100
GDF3	606522	100	100	100	97.75
GDF6	601147	100	100	98.68	75.80

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GDPD1	616317	100	100	100	99.82
GFER	600924	100	100	100	91.63
GJA1	121014	100	100	100	100
GJA3	121015	100	100	100	85.81
GJA8	600897	100	100	100	96.29
GLI2	165230	99.88	98.56	96.02	88.28
GLIS2	608539	100	100	99.38	92.32
GNAT1	139330	100	100	100	100
GNAT2	139340	100	100	100	100
GNB3	139130	100	100	100	97.21
GNPTG	607838	100	97.58	94.41	93.66
GPR143	300808	99.89	82.33	80.60	76.22
GPR179	614515	100	99.27	97.72	94.77
GRHL2	608576	100	100	99.84	98.09
GRIP1	604597	100	100	100	97.30
GRK1	180381	100	100	100	98.72
GRM6	604096	97.56	95.76	94.33	89.99
GRN	138945	100	100	100	97.18
GSN	137350	100	98.34	94.75	86.19
GUCA1A	600364	100	100	100	100
GUCA1B	602275	100	100	100	94.34
GUCY2D	600179	100	100	98.69	86.94
HARS1	142810	100	100	98.38	93.84
HCCS	300056	100	100	100	98.97
HGSNAT	610453	94.18	93.42	91.23	83.05
HK1	142600	100	99.94	98.93	93.60
HMGB3	300193	100	100	100	98.03
HMX1	142992	100	86.07	67.70	20.90
HPS1	604982	100	100	99.68	90.74
HPS3	606118	100	100	98.87	91.36
HPS4	606682	100	100	100	96.66
HPS5	607521	100	100	99.35	95.43
HPS6	607522	100	100	98.59	84.20
HRAS	190020	100	100	100	100
HSF4	602438	100	99.57	97.58	88.57
HSPG2	142461	99.45	99.11	97.49	90.45
IDH3A	601149	100	96.43	96.43	92.72
IDH3B	604526	100	100	100	98.45
IFT140	614620	100	100	99.07	94.18
IFT172	607386	100	100	99.58	97.91
IFT27	615870	100	100	100	98.35
IFT43	614068	100	100	100	98.69
IFT52	617094	100	100	100	100
IFT74	608040	100	100	95.28	79.01

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IFT81	605489	100	100	99.87	94.26
IGBP1	300139	100	100	99.15	93.30
IMPDH1	146690	100	98.62	92.40	89.49
IMPG1	602870	100	100	99.96	93.46
IMPG2	607056	100	99.88	98.61	96.28
INVS	243305	100	99.60	98.89	96.61
IPO13	610411	100	100	99.45	95.70
IQCB1	609237	100	100	100	99.98
JAM3	606197	100	100	100	98.29
IRX1	147265	90.86	88.22	86.09	80.43
ITPR1	601920	100	100	99.70	97.45
JAG1	606871	100	98.67	97.58	97.41
KATNIP	616650	100	99.80	98.31	93.37
KCNJ13	603208	100	100	100	100
KCNV2	607604	100	100	100	98.69
KERA	603288	100	100	100	100
KIAA0586	610178	100	100	98.83	91.18
KIAA1549	613344	97.04	96.69	96.47	93.45
KIF11	148760	100	99.75	97.56	90.98
KIF21A	608283	100	99.88	98.88	96.55
KIF3B	603754	100	100	100	99.02
KIF7	611254	99.38	95.78	90.74	79.67
KIZ	615757	100	100	100	96.10
KLHL7	611119	100	100	100	100
KRT12	601687	100	100	100	100
KRT3	148043	100	100	99.64	91.38
LAMA1	150320	100	100	99.91	98.34
LAMC1	150290	100	99.78	98.79	95.80
LCA5	309060	100	100	100	99.33
LAMP2	611408	100	100	100	91.30
LCAT	606967	100	100	98.65	87.92
LEMD2	616312	100	100	100	100
LIM2	154045	100	100	100	99.86
LOXL3	607163	100	100	98.91	97.02
LRAT	604863	100	100	100	100
LRIT3	615004	100	100	100	100
LRMDA	614537	100	100	96.94	93.89
LRP2	600073	100	99.96	99.64	97.62
LRP5	603506	97.91	97.91	97.26	93.39
LRPAP1	104225	99.27	94.39	89.44	80.99
LSS	600909	100	99.26	98.12	90.59
LTBP3	602090	99.59	98.30	93.77	80.79
LYST	606897	100	99.99	99.68	96.53
LZTFL1	606568	100	100	92.66	80.73

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MAB21L2	604357	100	100	100	99.36
MAF	177075	83.71	78.54	75.42	70.82
MAK	154235	100	100	98.90	93.62
MAPKAPK3	602130	100	100	100	96.73
MERTK	604705	100	100	100	99.16
MFN2	608507	100	100	99.87	96.69
MFRP	606227	100	100	100	95.77
MFSD8	611124	100	100	100	97.84
MIP	154050	100	100	100	99.19
MIR184	613146	No coverage data			
MIR204	610942	No coverage data			
MITF	156845	100	100	100	100
MKKS	604896	100	100	100	100
MKS1	609883	100	99.95	98.94	93.72
MLPH	606526	100	96.07	91.36	87.88
MT-TP	590075	No coverage data			
MVK	251170	100	100	100	96.15
MYO5A	160777	98.18	96.92	95.18	92.18
MYO7A	276903	100	99.89	98.51	93.74
MYOC	601652	100	100	100	97.68
NAA10	300013	100	100	94.22	61.79
NAGLU	609701	100	94.32	91.15	85.27
NBAS	608025	100	100	100	98.26
NDP	300658	100	100	100	100
NEK2	602985	100	100	100	97.93
NDUFS2	604043	100	100	100	93.32
NEK8	609799	100	100	100	94.78
NEUROD1	601724	100	100	100	100
NHS	300457	100	98.47	93.63	90.79
NMNAT1	608700	100	100	100	98.58
NPHP1	607100	99.79	96.88	96.88	95.81
NPHP3	608002	100	100	99.82	95.51
NPHP4	607215	100	100	99.14	92.28
NR2E3	604485	99.86	99.86	98.94	90.67
NR2F1	132890	100	97.29	93.34	85.21
NRL	162080	100	100	98.87	90.21
NYX	300278	100	100	97.37	88.00
OAT	613349	100	96.42	93.55	89.43
OCA2	611409	100	100	99.28	94.33
OCRL	300535	98.15	98.15	98.15	97.22
OFD1	300170	100	99.97	99.28	94.92
OPA1	605290	100	100	98.77	92.07
OPA3	606580	100	100	100	100
OPN1LW	300822	89.44	86.10	79.29	68.61

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OPN1MW	300821	52.60	39.03	30.16	21.45
OPN1SW	613522	100	100	100	99.65
OPTN	602432	100	100	97.51	79.26
OTX2	616729	100	100	100	100
OR2W3	600037	100	100	98.96	94.75
OVOL2	616441	100	95.52	84.48	68.29
P3H2	610341	100	100	100	91.30
P4HA2	600608	100	100	100	100
PANK2	606157	100	100	100	99.69
PAX2	167409	48.09	48.09	35.79	35.79
PAX6	607108	100	100	100	100
PCARE	613425	100	100	99.85	98.53
PCDH15	605514	100	100	99.92	98.82
PCYT1A	123695	100	100	100	99.80
PDE6A	180071	100	100	100	99.74
PDE6B	180072	100	100	100	98.77
PDE6C	600827	100	100	100	99.30
PDE6D	602676	100	100	100	100
PDE6G	180073	100	100	100	94.70
PDE6H	601190	100	100	94.50	51.94
PDZD7	612971	100	100	99.71	95.14
PEX1	602136	100	99.82	97.75	93.52
PEX2	170993	100	100	100	100
PEX26	608666	100	100	100	95.81
PEX6	601498	100	99.12	95.44	87.85
PEX7	601757	100	100	99.06	87.85
PGK1	311800	100	100	100	95.92
PHOX2A	602753	100	100	91.39	66.23
PHYH	602026	100	89.33	89.33	83.03
PIGL	605947	100	100	100	100
PIK3R1	171833	100	100	100	98.75
PIKFYVE	609414	100	100	100	98.74
PITPNM3	608921	98.74	95.89	92.41	85.51
PITX2	601542	100	96.62	93.01	82.88
PITX3	602669	100	100	100	88.30
PLA2G5	601192	100	100	100	100
PLK4	605031	100	100	100	98.37
PMPCA	613036	100	100	99.24	93.13
PNPLA6	603197	100	99.11	95.89	89.98
POC1B	614784	100	99.64	97.10	90.50
POC5	617880	100	100	100	98.77
POMGNT1	606822	100	100	100	98.99
PPT1	600722	100	100	100	96.71
PRCD	610598	100	88.96	59.68	18.69

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
PRDM13	616741	100	96.34	91.84	85.17
PRDM5	614161	100	100	98.97	93.00
PRIMPOL	615421	100	100	100	94.92
PROM1	604365	100	97.56	96.66	93.07
PRPF3	607301	100	100	99.96	97.57
PRPF31	606419	100	100	100	99.49
PRPF4	607795	100	100	100	100
PRPF6	613979	100	100	100	97.30
PRPF8	607300	100	100	100	98.94
PRPH2	179605	100	100	97.95	94.35
PRSS56	613858	100	97.90	91.58	70.58
PXDN	605158	100	99.87	99.20	96.52
RAB28	612994	100	99.75	95.64	84.56
RAB3GAP2	609275	100	100	98.75	97.08
RARB	180220	100	100	100	97.21
RAX	601881	100	99.59	90.48	78.83
RAX2	610362	100	100	100	93.92
RBP3	180290	100	100	100	100
RBP4	180250	100	100	100	100
RCBTB1	607867	100	100	100	99.78
RD3	180040	100	96.64	87.36	59.20
RDH11	607849	100	100	99.42	95.37
RDH12	608830	100	100	99.26	94.12
RDH5	601617	100	100	100	99.52
REEP6	609346	100	98.47	94.82	84.00
RGR	600342	100	100	98.29	79.98
RGS9	604067	100	100	100	95.42
RGS9BP	607814	100	100	100	97.66
RHO	180380	100	100	100	100
RIMS1	606629	100	100	99.34	87.65
RIMS2	606630	100	100	99.18	88.26
RLBP1	180090	100	100	100	94.96
ROM1	180721	100	99.10	96.45	74.39
RP1	603937	100	100	100	99.62
RP1L1	608581	100	99.98	96.41	88.24
RP2	300757	100	100	100	97.65
RP9	607331	100	99.62	97.96	92.98
RPE65	180069	100	100	100	100
RPGR	312610	77.06	72.91	70.98	65.44
RPGRIP1	605446	100	100	99.78	98.61
RPGRIP1L	610937	99.73	98.44	96.86	92.30
RS1	300839	100	100	100	100
RTN4IP1	610502	100	100	100	100
SAG	181031	100	100	100	100

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SALL2	602219	100	100	100	96.49
SALL4	607343	100	99.35	95.66	95.37
SAMD11	616765	80.78	74.51	66.36	55.72
SC5D	602286	100	100	100	100
SCAPER	611611	100	100	99.60	95.61
SCO2	604272	100	100	100	100
SDCCAG8	613524	100	100	99.08	92.68
SEMA4A	607292	100	100	99.22	94.09
SGSH	605270	93.52	93.52	93.52	93.25
SHH	600725	100	100	100	100
SIPA1L3	616655	100	99.24	97.28	91.43
SIX3	603714	100	100	98.12	93.58
SIX6	606326	100	100	99.36	95.89
SLC16A12	611910	100	100	100	98.98
SLC24A1	603617	100	100	99.11	92.09
SLC24A5	609802	100	100	100	97.32
SLC25A46	610826	100	100	99.15	92.40
SLC33A1	603690	100	100	100	99.18
SLC38A8	615585	100	100	98.84	87.87
SLC39A5	608730	100	99.45	95.11	75.30
SLC45A2	606202	100	100	100	99.88
SLC4A11	610206	100	100	100	98.46
SLC52A2	607882	100	100	100	100
SLC7A14	615720	100	100	100	98.10
SMOC1	608488	100	98.93	95.24	88.83
SNRNP200	601664	100	100	100	99.14
SOX2	184429	100	100	99.49	93.56
SOX5	604975	100	100	100	97.30
SPATA7	609868	100	99.90	95.24	86.60
SPP2	602637	100	100	100	100
SRD5A3	611715	100	100	98.58	94.73
SSBP1	600439	100	100	100	99.11
STRA6	610745	100	100	100	96.01
SUFU	607035	100	100	99.70	96.51
TACSTD2	137290	100	100	100	97.67
TAMALIN	612027	89.95	77.80	65.54	44.45
TCF7L2	602228	100	100	98.54	89.94
TCOF1	606847	100	99.96	96.81	86.51
TCTN1	609863	100	100	100	100
TCTN2	613846	100	100	99.43	95.27
TCTN3	613847	100	100	100	98.55
TDRD7	611258	100	99.09	98.60	96.38
TEAD1	189967	100	100	100	99.67
TEK	600221	100	100	100	99.73

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TENM3	610083	100	100	99.90	98.59
TFAP2A	107580	100	100	100	93.34
TGFBI	601692	100	99.67	97.74	92.51
TGIF1	602630	100	100	100	100
TIMM8A	300356	100	100	100	93.05
TIMP3	188826	100	100	96.79	85.95
TMCO3	617134	100	99.19	97.18	89.72
TMEM107	616183	100	100	100	95.82
TMEM126A	612988	100	100	100	100
TMEM138	614459	100	100	100	100
TMEM216	613277	100	100	100	99.45
TMEM231	614949	100	100	100	95.56
TMEM237	614423	100	100	99.26	94.88
TMEM67	609884	100	100	99.87	95.43
TMEM98	615949	100	100	100	99.00
TOGARAM1	617618	100	100	99.65	96.38
TOPORS	609507	100	100	100	99.00
TPP1	607998	100	98.87	96.77	93.28
TRAF3IP1	607380	100	99.52	95.15	79.98
TREX1	606609	100	100	100	100
TRIM32	602290	100	100	100	100
TRNT1	612907	100	97.75	89.81	86.79
TRPM1	603576	100	99.44	98.82	97.16
TSPAN12	613138	100	100	100	99.15
TTC8	608132	100	100	100	100
TLL10	No OMIM ID	100	100	98.22	85.30
TUB	601197	100	100	98.07	89.04
TUBA3D	617878	100	100	100	100
TUBB3	602661	100	100	94.62	94.62
TUBB4B	602660	100	100	100	98.30
TUBGCP4	609610	100	100	99.79	97.90
TUBGCP6	610053	100	100	100	98.11
TULP1	602280	100	96.21	86.42	61.71
TYR	606933	100	100	100	99.76
TYRP1	115501	100	100	100	100
UBIAD1	611632	100	100	100	98.77
UNC119	604011	100	100	97.74	91.34
UNC45B	611220	100	100	100	98.64
USH1C	605242	100	100	99.91	95.61
USH1G	607696	100	100	100	100
USH2A	608400	100	100	100	99.34
USP45	618439	100	99.91	96.51	89.40
VAX1	604294	100	99.92	95.53	71.39
VCAN	118661	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
VPS13B	193060	99.28	98.29	97.00	91.79
VIM	607817	100	100	100	100
VSX1	605020	100	96.83	82.87	72.46
VSX2	142993	100	100	99.54	90.66
WASHC5	610657	100	100	100	99.12
WDPCP	613580	100	100	99.54	95.61
WDR19	608151	100	100	99.75	97.05
WDR36	609669	100	100	100	98.98
WFS1	606201	100	100	99.11	96.60
WHRN	607928	100	100	99.70	95.64
WRN	604611	100	100	99.77	94.20
YAP1	606608	100	99.30	96.13	89.73
- OMIM release used: 23-9-2021				98.20	88.89
- The statistics above are based on a set of 104 samples				98.85	92.88
- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x				100	98.32
ZNF423	604557	98.51	98.51	98.51	98.09