

Whole Exome Sequencing

Gene package Hearing impairment, version 9, 26-2-2021



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/>). Sequence variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: <http://www.broadinstitute.org/gatk/>). The detected sequence variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. Copy 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 filtered and annotated with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). It is not excluded that pathogenic variants 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).



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 $\geq 50x$
ABCC1	158343	98.70	98.45	97.52	93.91
ABHD12	613599	99.76	95.79	90.25	80.60
ACTB	102630	100	100	100	98.33
ACTG1	102560	100	100	100	100
ADCY1	103072	95.29	93.41	91.51	87.10
ADGRV1	602851	100	99.85	99.13	95.06
AIFM1	300169	100	100	98.86	93.87
ALMS1	606844	99.78	99.08	97.76	93.88
AP1B1	600157	100	100	99.41	91.94
ARSG	610008	100	100	98.75	93.06
ATP1A3	182350	100	100	99.04	92.02
ATP2B2	108733	100	99.93	98.78	92.98
ATP6VOA4	605239	100	99.55	98.84	95.98
ATP6V1B1	192132	100	100	98.38	90.54
ATP6V1B2	606939	100	99.34	97.38	91.51
BCAP31	300398	100	100	97.49	82.40
BCS1L	603647	100	100	100	100
BDP1	607012	99.78	98.04	94.73	81.59
BSND	606412	100	100	100	98.17

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
CABP2	607314	100	99.63	92.18	67.73
CACNA1D	114206	100	100	99.51	96.28
CCDC50	611051	100	100	100	97.09
CD151	602243	100	100	99.44	87.26
CD164	603356	100	100	100	91.28
CDC14A	603504	100	100	100	94.44
CDH23	605516	100	100	99.82	84.11
CEACAM16	614591	100	100	99.53	87.49
CEP250	609689	100	99.34	96.41	82.99
CEP78	617110	100	99.50	97.67	88.86
CHD7	608892	100	99.87	98.61	96.80
CHSY1	608183	94.28	93.55	92.76	90.21
CIB2	605564	100	100	100	96.62
CISD2	611507	76.99	76.99	76.99	76.99
CLDN14	605608	100	100	100	100
CLDN9	615799	100	100	97.91	85.17
CLIC5	607293	100	100	100	99.77
CLPP	601119	100	92.90	81.44	62.09
CLRN1	606397	100	100	100	98.34
CLRN2	618988	100	99.27	95.24	82.54
COA8	616003	100	100	100	97.20
COCH	603196	100	100	97.38	90.02
COL11A1	120280	100	98.68	95.94	84.52
COL11A2	120290	100	99.07	94.23	80.93
COL2A1	120140	100	99.83	98.57	91.36
COL4A3	120070	99.92	98.23	97.80	91.93
COL4A4	120131	100	98.74	95.36	80.57
COL4A5	303630	99.68	97.90	95.61	83.91
COL4A6	303631	100	99.05	96.81	86.28
COL9A1	120210	100	99.65	96.88	80.71
COL9A2	120260	100	95.05	86.28	57.61
CRYM	123740	100	100	96.55	80.94
DCAF17	612515	100	100	98.51	91.39
DCDC2	605755	100	99.36	95.76	87.13
DIABLO	605219	100	100	100	100
DIAPH1	602121	100	97.68	91.70	86.14
DIAPH3	614567	99.79	96.82	93.01	84.74
DMXL2	612186	100	99.81	98.99	95.20
DSPP	125485	56.72	53.11	50.48	44.82

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
EDN3	131242	100	100	91.95	82.06
EDNRB	131244	100	99.35	97.81	92.14
ELMOD3	615427	100	100	100	100
ELOVL1	611813	100	100	100	94.06
EPS8	600206	100	98.92	97.75	91.49
EPS8L2	614988	100	95.13	85.48	72.12
ERAL1	607435	100	100	100	99.54
ESPN	606351	98.19	93.59	84.59	65.31
ESRP1	612959	100	100	99.19	93.36
ESRRB	602167	100	100	99.88	95.99
EXOSC2	602238	100	100	100	96.51
EYA1	601653	97.62	97.62	97.41	92.02
EYA4	603550	100	100	100	98.36
FDXR	103270	100	100	99.95	94.19
FGF3	164950	100	97.88	94.47	79.99
FITM2	612029	100	100	99.76	93.40
FOXF2	603250	89.87	87.83	85.79	82.18
FOXI1	601093	100	100	100	95.95
GAB1	604439	100	100	99.16	92.20
GATA3	131320	100	100	100	91.62
GIPC3	608792	100	96.97	94.13	85.56
GJB2	121011	100	100	100	96.78
GJB3	603324	100	100	100	100
GJB6	604418	100	100	100	100
GLA	300644	100	100	99.72	95.09
GPRASP2	300969	100	99.96	98.84	95.15
GPSM2	609245	100	100	100	96.66
GRAP	604330	67.91	67.91	65.91	52.20
GREB1L	617782	100	99.89	99.37	95.76
GRHL2	608576	100	100	99.09	92.21
GRXCR1	613283	100	100	100	100
GRXCR2	615762	100	100	100	98.01
GSDME	608798	100	99.64	97.90	92.03
HARS1	142810	100	100	99.05	93.84
HARS2	600783	100	100	100	99.26
HGF	142409	100	100	97.19	85.21
HOMER2	604799	97.99	97.99	97.99	97.99
HSD17B4	601860	98.96	97.05	94.86	87.75
IFNLR1	607404	100	100	96.95	75.41

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ILDR1	609739	100	100	99.83	94.91
KARS1	601421	100	100	100	98.00
KCNE1	176261	100	100	100	100
KCNJ10	602208	100	100	100	100
KCNQ1	607542	95.21	92.93	91.44	85.39
KCNQ4	603537	96.74	94.86	93.76	89.94
KITLG	184745	100	100	100	99.40
LARS2	604544	100	100	99.87	96.99
LHFPL5	609427	100	100	100	97.35
LMX1A	600298	100	98.85	95.14	82.35
LOXHD1	613072	100	100	100	99.22
LOXL3	607163	100	100	98.87	96.90
LRTOMT	612414	100	100	97.33	78.21
MARVELD2	610572	100	96.18	91.50	87.18
MCM2	116945	100	100	99.74	94.13
MET	164860	100	99.65	98.92	95.52
MGP	154870	100	91.01	89.05	87.60
MIR96	611606	No coverage data			
MITF	156845	100	100	100	100
MPZL2	604873	100	100	100	97.58
MSRB3	613719	100	88.00	83.19	77.23
MYH14	608568	100	98.43	94.74	76.63
MYH9	160775	100	98.52	93.72	79.57
MYO15A	602666	99.86	98.52	96.67	87.35
MYO3A	606808	100	100	100	100
MYO6	600970	99.21	97.84	96.44	88.51
MYO7A	276903	100	99.80	98.09	92.06
NARS2	612803	97.15	93.76	93.76	88.66
NCOA3	601937	100	100	99.57	96.44
NDP	300658	100	100	100	100
NLRP3	606416	100	100	99.92	98.10
NOG	602991	100	100	100	97.91
OPA1	605290	100	99.07	96.09	86.31
OSBPL2	606731	100	100	98.26	89
OTOA	607038	81.32	78.63	76.43	71.85
OTOF	603681	100	99.42	97.65	93.38
OTOG	604487	100	99.76	98.10	92.43
OTOGL	614925	100	99.29	97.83	93.89
P2RX2	600844	99.88	97.07	91.37	79.18

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
PAX3	606597	100	100	100	92.76
PCDH15	605514	100	100	99.56	97.34
PDE1C	602987	100	99.83	97.90	95.63
PDZD7	612971	100	100	99.24	91.63
PET100	614770	100	100	100	100
PEX1	602136	100	98.75	95.78	90.34
PEX26	608666	100	100	100	91.21
PEX6	601498	100	98.93	95.14	85.69
PJKV	610219	100	100	100	100
PLOD3	603066	100	95.66	91.42	75.96
PLS1	602734	100	100	99.29	96.18
PNPT1	610316	97.04	90.87	86.78	81.14
POLD1	174761	100	99.85	97.55	88.71
POLR1C	610060	100	100	100	92.16
POLR1D	613715	100	100	100	89.52
POU3F4	300039	100	100	100	100
POU4F3	602460	100	100	100	100
PPIP5K2	611648	99.68	98.49	95.90	88.69
PRKCB	176970	100	100	99.85	95.15
PRPS1	311850	100	100	100	92.79
PSIP1	603620	100	99.37	93.28	83.44
PTPRQ	603317	98.31	97.20	96.63	93.71
PTRH2	608625	100	100	100	100
RAI1	607642	100	100	99.76	96.85
RDX	179410	100	93.94	92.26	86.76
REST	600571	100	100	100	98.73
RIPOR2	611410	100	100	99.03	93.95
ROR1	602336	96.29	96.29	96.19	94.22
S1PR2	605111	100	100	100	100
SCD5	608370	100	100	100	98.00
SERPINB6	173321	100	100	100	97.32
SIX1	601205	100	100	100	95.07
SIX5	600963	99.93	90.89	72.31	46.95
SLC12A2	600840	98.54	95.71	93.24	87.10
SLC17A8	607557	100	100	99.88	97.33
SLC19A2	603941	100	100	98.38	84.41
SLC22A4	604190	100	100	99.30	93.36
SLC25A2	608157	100	100	100	100
SLC26A4	605646	100	100	100	97.88

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
SLC26A5	604943	100	100	99.85	95.43
SLC29A3	612373	100	97.60	97.60	97.14
SLC33A1	603690	100	100	99.77	98.02
SLC44A4	606107	100	99.22	95.24	79.20
SLC52A2	607882	100	100	100	95.83
SLITRK6	609681	100	100	100	100
SMPX	300226	100	100	100	71.60
SNAI2	602150	100	100	99.25	93.75
SOX10	602229	100	98.83	96.64	87.59
SPATA5	613940	100	100	99.70	96.35
SPNS2	612584	94.36	92.24	88.02	76.74
STRC	606440	51.96	43.36	40.76	36.02
SYNE4	615535	99.14	92.04	86.81	55.89
TBC1D24	613577	100	100	99.15	96.48
TBL1Y	400033	0	0	0	0
TCOF1	606847	100	99.17	94.44	81.90
TECTA	602574	100	100	99.60	96.70
THOC1	606930	99.77	93.64	93.64	92.24
TIMM8A	300356	100	100	100	92.75
TJP2	607709	100	100	99.54	95.69
TMC1	606706	100	98.00	93.47	87.43
TMEM132E	616178	100	100	98.59	91.77
TMIE	607237	96.26	72.99	56.30	56.20
TMPRSS3	605511	100	100	98.69	90.64
TMTC2	615856	100	99.64	96.67	96.03
TNC	187380	100	100	100	99.72
TPRN	613354	86.24	74.04	65.93	61.27
TRIOBP	609761	99.83	97.73	94.23	84.46
TSHZ1	614427	98.17	98.17	98.17	98.17
TSPEAR	612920	100	100	98.66	89.83
TWINK	606075	100	100	100	100
TYR	606933	100	100	100	99.47
USH1C	605242	100	100	99.27	93.22
USH1G	607696	100	100	100	99.51
USH2A	608400	100	100	100	97.52
USP48	617445	100	97.06	95.23	91.07
WBP2	606962	100	100	97.77	83.40
WFS1	606201	100	100	99.40	96.23
WHRN	607928	100	100	99.32	92.43

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
YAP1	606608	100	97.83	93.87	84.65

- OMIM release used: 18-2-2021

- The statistics above are based on a set of 100 samples

- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence ($\pm 10bp$ flanking introns) that is covered at least 10x, 20x, 30x or 50x