

Whole Exome Sequencing

Gene package Hearing impairment, 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 $\geq 50x$
ABCC1	158343	98.70	98.60	97.90	95.37
ABHD12	613599	100	96.57	91.81	82.57
ACTB	102630	100	100	100	99.18
ACTG1	102560	100	100	100	100
ADCY1	103072	95.82	93.94	92.19	87.13
ADGRV1	602851	100	100	99.73	97.56
AIFM1	300169	100	100	99.17	95.63
ALMS1	606844	99.89	99.40	98.78	96.05
AMMECR1	300195	100	100	100	95.24
AP1B1	600157	100	100	99.85	94.13
ARSG	610008	100	100	99.03	94.60
ATP1A3	182350	100	100	99.45	94.42
ATP2B4	108732	100	100	99.90	97.82
ATP6VOA4	605239	100	99.90	99.14	97.88
ATP6V1B1	192132	100	100	98.90	92.41
ATP6V1B2	606939	100	100	99.09	95.37
BCAP31	300398	100	100	99.09	85.82
BCS1L	603647	100	100	100	100
BDP1	607012	100	99.55	97.72	89.44

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BSND	606412	100	100	100	99.09
CABP2	607314	100	100	93.34	70.42
CACNA1D	114206	100	100	100	99.10
CCDC50	611051	100	100	100	99.56
CD151	602243	100	100	99.67	85.71
CD164	603356	100	100	100	99.37
CDC14A	603504	100	100	100	99.22
CDH23	605516	100	100	100	89.21
CEACAM16	614591	100	100	99.89	91.51
CEP250	609689	100	99.89	98.59	90.04
CEP78	617110	100	99.96	99.16	94.59
CHD7	608892	100	100	99.89	98.69
CHSY1	608183	94.20	93.39	92.74	90.55
CIB2	605564	100	100	100	98.38
CISD2	611507	88.49	76.99	76.99	76.99
CLDN14	605608	100	100	100	100
CLDN9	615799	100	100	98.36	89.57
CLIC5	607293	100	100	100	99.89
CLPP	601119	100	94.95	85.23	64.56
CLRN1	606397	100	100	100	100
CLRN2	618988	100	98.61	94.44	81.08
COA8	616003	100	100	100	100
COCH	603196	100	100	98.81	91.72
COL11A1	120280	100	99.21	97.82	88.98
COL11A2	120290	100	99.90	95.62	84.23
COL2A1	120140	100	100	99.42	93.30
COL4A3	120070	100	98.23	98.01	94.40
COL4A4	120131	100	98.86	95.50	81.24
COL4A5	303630	100	98.33	97.18	91.73
COL4A6	303631	100	99.10	96.48	88.09
COL9A1	120210	100	100	98.08	86.28
COL9A2	120260	100	96.25	87.33	63.80
CRYM	123740	100	100	97.19	80.40
DCAF17	612515	100	100	100	96.52
DCDC2	605755	100	100	97.97	88.76
DIABLO	605219	100	100	100	100
DIAPH1	602121	100	99.04	94.46	87.47
DIAPH3	614567	100	98.60	95.87	88.96
DMXL2	612186	100	100	99.75	97.48

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DSPP	125485	60.47	54.96	52.86	49.59
DTD1	610996	66.85	66.85	66.85	66.85
EDN3	131242	100	100	99.02	83.05
EDNRB	131244	100	99.83	98.58	95.24
ELMOD3	615427	100	100	100	100
ELOVL1	611813	100	100	100	97.85
EPS8	600206	100	99.41	98.64	95.32
EPS8L2	614988	100	97.21	88.21	76.29
ERAL1	607435	100	100	100	100
ESPN	606351	98.30	94.42	87.31	67.40
ESRP1	612959	100	100	99.91	96.50
ESRRB	602167	100	100	100	97.92
EXOSC2	602238	100	100	100	98.44
EYA1	601653	97.62	97.62	97.62	95.91
EYA4	603550	100	100	100	100
FDXR	103270	100	100	98.81	93.02
FGF3	164950	100	98.33	94.59	84.43
FITM2	612029	100	100	100	96.85
FOXF2	603250	89.50	87.39	85.60	82.51
FOX11	601093	100	100	100	94.66
GAB1	604439	100	100	100	97.09
GATA3	131320	100	100	100	94.52
GIPC3	608792	100	96.35	94.13	87.88
GJB2	121011	100	100	100	98.35
GJB3	603324	100	100	100	100
GJB6	604418	100	100	100	100
GLA	300644	100	100	100	98.28
GPRASP2	300969	100	100	100	98.90
GPSM2	609245	100	100	100	99.27
GRAP	604330	67.91	67.91	67.91	58.52
GREB1L	617782	100	100	99.83	98.44
GRHL2	608576	100	100	99.84	98.09
GRXCR1	613283	100	100	100	100
GRXCR2	615762	100	100	100	98.51
GSDME	608798	100	99.64	98.59	93.35
HARS1	142810	100	100	98.38	93.84
HGF	142409	100	100	100	96.16
HOMER2	604799	97.99	97.99	97.99	97.99
HSD17B4	601860	100	98.63	96.80	91.72

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IFNLR1	607404	100	100	98.47	81.16
ILDR1	609739	100	100	100	95.75
KARS1	601421	100	100	100	99.05
KCNE1	176261	100	100	100	100
KCNJ10	602208	100	100	100	100
KCNJ16	605722	100	100	100	100
KCNQ1	607542	94.04	92.61	91.25	85.86
KCNQ4	603537	96.66	94.82	93.72	90.08
KITLG	184745	100	100	100	100
LARS2	604544	100	100	100	98.83
LHFPL5	609427	100	100	100	97.91
LMX1A	600298	100	99.85	96.52	86.45
LOXHD1	613072	100	100	99.98	99.06
LOXL3	607163	100	100	98.91	97.02
LRTOMT	612414	100	100	96.92	77.03
MARVELD2	610572	100	99.58	94.73	88.13
MCM7	600592	100	100	100	97.23
MET	164860	100	100	99.48	97.53
MGP	154870	100	99.59	90.39	89.05
MIR96	611606	No coverage data		-	
MITF	156845	100	100	100	100
MPZL2	604873	100	100	100	100
MSRB3	613719	100	99.86	85.63	80.46
MYH14	608568	100	99.24	96.59	83.35
MYH9	160775	100	98.19	92.96	78.40
MYO15A	602666	99.72	98.59	97.20	89.86
MYO3A	606808	100	100	100	100
MYO6	600970	100	97.84	97.77	93.44
MYO7A	276903	100	99.89	98.51	93.74
NARS2	612803	99.89	93.87	93.76	92.42
NCOA3	601937	100	100	100	98.49
NDP	300658	100	100	100	100
NLRP3	606416	100	100	100	99.39
NOG	602991	100	100	100	99.16
OPA1	605290	100	100	98.77	92.07
OSBPL2	606731	100	100	100	95.53
OTOA	607038	82.19	79.18	77.13	74.34
OTOF	603681	100	99.51	97.54	93.94
OTOG	604487	100	99.75	98.30	93.75

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OTOGL	614925	100	99.85	98.56	95.54
P2RX2	600844	100	98.46	94.09	81.43
PAX3	606597	100	100	100	94
PCDH15	605514	100	100	99.92	98.82
PDE1C	602987	100	100	99.63	96.15
PDZD7	612971	100	100	99.71	95.14
PET100	614770	100	100	100	100
PEX1	602136	100	99.82	97.75	93.52
PEX26	608666	100	100	100	95.81
PEX6	601498	100	99.12	95.44	87.85
PISD	612770	100	100	99.78	86.73
PJKV	610219	100	100	100	100
PLOD3	603066	100	97.38	93.62	79.16
PLS1	602734	100	100	100	97.97
PNPT1	610316	98.54	93.92	89.45	85.32
POLD1	174761	100	99.85	98.25	90.01
POLR1C	610060	100	100	100	94.29
POLR1D	613715	100	100	100	90.09
POU3F4	300039	100	100	100	100
POU4F3	602460	100	100	100	100
PPIP5K2	611648	99.88	99.21	97.86	93.15
PRKCB	176970	100	100	100	98.94
PRPS1	311850	100	100	100	99.75
PSIP1	603620	100	100	98.97	89.18
PTPRR	602853	100	100	100	99.51
PTRH2	608625	100	100	100	100
RAI1	607642	100	100	99.95	98.22
RDX	179410	100	100	93.94	90.85
REST	600571	100	100	100	100
RIPOR2	611410	100	100	99.82	96.36
ROR1	602336	96.29	96.29	96.29	95.05
S1PR2	605111	100	100	100	100
SCD5	608370	100	100	100	99.82
SERPINB6	173321	100	100	100	100
SIX1	601205	100	100	100	96.52
SIX5	600963	100	93.35	79.31	56.68
SLC12A1	600839	100	100	99.40	96.06
SLC12A2	600840	98.93	96.68	95.06	89.88
SLC17A8	607557	100	100	100	99.15

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SLC19A2	603941	100	100	100	94.48
SLC22A4	604190	100	100	100	96.60
SLC26A4	605646	100	100	100	99.12
SLC26A5	604943	100	100	100	98.84
SLC29A3	612373	100	97.60	97.60	97.48
SLC33A1	603690	100	100	100	99.18
SLC44A4	606107	100	99.84	97.45	82.80
SLC52A2	607882	100	100	100	100
SLC52A3	613350	100	100	99.46	96.97
SLITRK6	609681	100	100	100	100
SMPX	300226	100	100	100	100
SNAI2	602150	100	100	100	95.14
SOX10	602229	100	99.66	97.70	90.57
SPATA5	613940	100	100	100	98.45
SPATA5L1	619578	99.92	98.02	96.13	86.94
SPNS2	612584	94.54	92.58	88.82	78.75
STRC	606440	52.29	44.27	41.46	36.79
SYNE4	615535	99.19	92.85	88.57	64.30
TBC1D24	613577	100	100	99.06	96.31
TBL1Y	400033	0	0	0	0
TCOF1	606847	100	99.96	96.81	86.51
TECTA	602574	100	100	99.94	97.96
TFAP2A	107580	100	100	100	93.34
THOC1	606930	100	99.37	93.66	93.64
TIMM8A	300356	100	100	100	93.05
TJP2	607709	100	100	100	98.63
TMC1	606706	100	99.48	98.28	93.36
TMEM132E	616178	100	100	99.41	94.03
TMIE	607237	99.45	89.60	63.87	56.20
TMPRSS3	605511	100	100	98.69	92.57
TMTC2	615856	100	100	97.87	96.25
TNC	187380	100	100	100	99.96
TPRN	613354	87.03	74.63	65.32	61.68
TRIOBP	609761	99.95	98.50	95.22	88.06
TRRAP	603015	99.84	99.71	99.47	96.97
TSHZ1	614427	98.17	98.17	98.17	98.17
TSPEAR	612920	100	100	99.40	93.37
TUBB4B	602660	100	100	100	98.30
TWNK	606075	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
TYR	606933	100	100	100	99.76
USH1C	605242	100	100	99.91	95.61
USH1G	607696	100	100	100	100
USH2A	608400	100	100	100	99.34
USP48	617445	100	98.92	95.68	93.80
USP53	617431	100	99.77	98.91	94.37
WBP2	606962	100	100	98.57	90.77
WFS1	606201	100	100	99.11	96.60
WHRN	607928	100	100	99.70	95.64
YAP1	606608	100	99.30	96.13	89.73

- OMIM release used: 23-9-2021

- The statistics above are based on a set of 104 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