

Whole Exome Sequencing

Gene package Hearing impairment, 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).



HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ABCC1	158343	184	100	99	98
ABHD12	613599	133	100	99	93
ACTB	102630	253	100	100	100
ACTG1	102560	233	100	100	100
ADCY1	103072	177	98	97	96
ADGRV1	602851	98	99	94	89
AIFM1	300169	89	100	99	96
ALMS1	606844	116	100	99	96
AP1B1	600157	250	100	100	100
ARSG	610008	167	100	100	100
ATP1A3	182350	221	100	100	100
ATP2B2	108733	231	100	100	100
ATP6V0A4	605239	104	100	99	98
ATP6V1B1	192132	206	100	100	100
ATP6V1B2	606939	94	100	99	98
BCAP31	300398	82	100	100	95
BCS1L	603647	263	100	100	100
BDP1	607012	60	97	87	75
BSND	606412	267	100	100	100
CABP2	607314	162	100	100	100
CACNA1D	114206	148	100	99	96
CCDC50	611051	94	100	96	89
CD164	603356	109	100	98	88
CDC14A	603504	86	96	90	84

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CDH23	605516	257	100	100	100
CEACAM16	614591	211	100	100	100
CEP250	609689	157	100	100	99
CEP78	617110	67	98	95	88
CHD7	608892	167	100	99	97
CIB2	605564	185	100	100	100
CISD2	611507	97	76	76	76
CLDN14	605608	601	100	100	100
CLDN9	615799	303	100	100	100
CLIC5	607293	151	100	100	100
CLPP	601119	139	100	100	100
CLRN1	606397	92	100	96	92
COA8	616003	91	100	94	80
COCH	603196	137	100	100	99
COL11A1	120280	86	99	95	89
COL11A2	120290	186	100	100	99
COL2A1	120140	203	100	100	99
COL4A3	120070	132	100	99	97
COL4A4	120131	123	100	99	97
COL4A5	303630	84	99	97	92
COL4A6	303631	92	100	99	96
COL9A1	120210	99	100	98	96
COL9A2	120260	153	100	100	100
CRYM	123740	142	100	100	100
DCDC2	605755	144	95	84	77
DIABLO	605219	135	100	100	100
DIAPH1	602121	101	100	99	96
DIAPH3	614567	70	86	79	71
DMXL2	612186	87	99	95	89
DSPP	125485	32	54	49	45
EDN3	131242	187	100	100	100
EDNRB	131244	135	100	99	94
ELMOD3	615427	173	100	100	100
ELOVL1	611813	118	100	100	100
EPS8	600206	82	100	96	88
EPS8L2	614988	217	100	100	100
ERAL1	607435	224	100	100	100
ESPN	606351	187	99	99	98
ESRP1	612959	102	99	96	92
ESRRB	602167	283	100	100	100
EXOSC2	602238	107	100	100	100
EYA1	601653	111	95	95	89
EYA4	603550	93	100	95	90

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FGF3	164950	219	100	100	100
FOXF2	603250	244	92	89	88
FOX11	601093	172	100	100	98
GAB1	604439	86	100	99	93
GATA3	131320	199	100	100	100
GIPC3	608792	232	100	100	100
GJB2	121011	191	100	100	100
GJB3	603324	289	100	100	100
GJB6	604418	154	100	100	100
GPRASP2	300969	103	98	95	92
GPSM2	609245	54	100	96	84
GRAP	604330	162	68	68	68
GREB1L	617782	113	100	99	97
GRHL2	608576	120	100	100	98
GRXCR1	613283	100	100	100	100
GRXCR2	615762	149	100	99	96
GSDME	608798	125	100	99	97
HARS1	142810	163	100	100	100
HARS2	600783	138	100	100	100
HGF	142409	103	99	95	90
HOMER2	604799	170	100	98	98
HSD17B4	601860	78	93	91	84
IFNLR1	607404	144	100	100	100
ILDR1	609739	193	100	100	98
KARS1	601421	123	100	100	99
KCNE1	176261	657	100	100	100
KCNJ10	602208	271	100	100	100
KCNQ1	607542	235	100	97	94
KCNQ4	603537	245	98	97	96
KITLG	184745	83	100	100	92
LARS2	604544	126	100	100	99
LHFPL5	609427	263	100	100	100
LMX1A	600298	170	100	100	100
LOXHD1	613072	197	100	100	100
LOXL3	607163	234	100	100	100
LRTOMT	612414	174	100	100	100
MARVELD2	610572	142	90	85	84
MCM2	116945	211	100	100	100
MET	164860	111	100	99	97
MIR96	611606	No coverage data			
MITF	156845	119	100	100	93
MPZL2	604873	142	100	100	100
MSRB3	613719	64	92	79	78

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MYH14	608568	204	100	100	100
MYH9	160775	156	100	100	99
MYO15A	602666	285	100	100	100
MYO3A	606808	86	97	87	80
MYO6	600970	57	93	82	66
MYO7A	276903	214	100	100	100
NARS2	612803	79	97	93	84
NLRP3	606416	404	100	100	100
NOG	602991	322	100	100	100
OPA1	605290	55	90	79	67
OSBPL2	606731	196	100	100	97
OTOA	607038	120	81	78	75
OTOF	603681	267	100	100	99
OTOG	604487	246	100	100	100
OTOGL	614925	70	95	91	83
P2RX2	600844	216	100	100	100
PAX3	606597	178	100	100	100
PCDH15	605514	152	99	97	95
PDE1C	602987	96	99	96	96
PDZD7	612971	247	100	100	100
PET100	614770	178	100	100	100
PEX1	602136	80	95	94	90
PEX26	608666	151	100	100	100
PEX6	601498	253	100	100	99
PJK	610219	107	100	100	99
PLOD3	603066	212	100	100	100
PLS1	602734	66	100	97	89
PNPT1	610316	57	84	76	65
POLD1	174761	291	100	100	100
POLR1C	610060	136	100	100	100
POLR1D	613715	123	100	100	100
POU3F4	300039	397	100	100	100
POU4F3	602460	364	100	100	100
PPIP5K2	611648	51	94	83	68
PRKCB	176970	125	100	99	96
PRPS1	311850	70	100	98	92
PTPRQ	603317	57	96	88	75
RAI1	607642	405	100	100	100
RDX	179410	58	86	72	61
REST	600571	141	100	100	99
RIPOR2	611410	120	100	99	98
ROR1	602336	144	100	98	96
S1PR2	605111	491	100	100	100

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SERPINB6	173321	151	100	100	95
SIX1	601205	284	100	100	100
SIX5	600963	167	100	100	100
SLC17A8	607557	109	100	100	98
SLC19A2	603941	208	100	99	96
SLC22A4	604190	138	100	98	93
SLC25A2	608157	230	100	100	100
SLC26A4	605646	121	100	96	91
SLC26A5	604943	86	98	96	94
SLC29A3	612373	233	100	100	100
SLC33A1	603690	147	98	93	85
SLC44A4	606107	170	100	100	100
SLC52A2	607882	210	100	100	100
SLITRK6	609681	134	100	100	100
SMPX	300226	34	100	80	40
SNAI2	602150	151	100	100	100
SOX10	602229	381	100	100	100
SPATA5	613940	95	100	99	94
SPNS2	612584	192	97	96	95
STRC	606440	67	60	50	46
SYNE4	615535	173	100	100	97
TBC1D24	613577	390	100	100	100
TCOF1	606847	201	100	100	98
TECTA	602574	226	100	100	100
TIMM8A	300356	98	100	100	100
TJP2	607709	125	100	100	98
TMC1	606706	86	99	95	89
TMEM132E	616178	270	100	100	100
TMIE	607237	135	94	76	76
TMPRSS3	605511	207	100	100	100
TMTC2	615856	105	100	100	99
TNC	187380	225	100	100	100
TPRN	613354	185	93	88	83
TRIOBP	609761	259	100	100	100
TSPEAR	612920	207	100	100	100
TWNK	606075	231	100	100	100
TYR	606933	147	100	99	98
USH1C	605242	209	100	100	99
USH1G	607696	427	100	100	100
USH2A	608400	116	100	99	97
USP48	617445	63	98	96	89
WBP2	606962	155	100	100	100
WFS1	606201	268	100	100	100

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WHRN	607928	288	100	100	100
YAP1	606608	104	100	100	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 (± 10 bp flanking introns) of the longest transcript

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