Whole Exome Sequencing
Gene package Hearing impairment, 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ACTB	102630	192	100	100	100
ACTG1	102560	188	100	100	100
ADCY1	103072	109	97	95	93
ADGRV1	602851	80	100	100	97
AIFM1	300169	55	100	96	82
AP1B1	600157	94	100	99	96
ATP1A3	182350	144	100	100	100
ATP2B2	108733	139	100	100	100
ATP6V1B1	192132	136	100	100	100
BCS1L	603647	170	100	100	100
BDP1	607012	60	99	96	85
BSND	606412	115	100	100	100
CABP2	607314	94	100	100	96
CACNA1D	114206	102	100	100	98
CCDC50	611051	73	100	100	98
CD164	603356	71	100	100	93

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CDC14A	603504	79	100	100	94
CDH23	605516	135	100	100	100
CEACAM16	614591	94	100	100	100
CEP78	617110	69	100	100	94
CIB2	605564	195	100	100	100
CISD2	611507	137	100	100	100
CLDN14	605608	66	100	98	91
CLIC5	607293	76	100	100	100
CLPP	601119	125	100	100	100
CLRN1	606397	83	100	100	97
COA8	616003	69	100	100	92
COCH	603196	71	98	95	93
COL11A1	120280	62	100	99	93
COL11A2	120290	110	100	100	100
COL2A1	120140	97	100	100	99
COL4A3	120070	63	100	97	90
COL4A4	120131	70	100	100	96
COL4A5	303630	42	100	88	67
COL4A6	303631	62	100	98	90
COL9A1	120210	65	100	99	91
COL9A2	120260	100	100	99	96
CRYM	123740	76	100	100	99
DCDC2	605755	96	100	100	97
DIABLO	605219	105	100	100	99
DIAPH1	602121	84	100	99	93
DIAPH3	614567	61	100	99	93
DMXL2	612186	67	100	99	93
DSPP	125485	43	95	70	55
EDN3	131242	90	100	100	98
EDNRB	131244	96	100	100	100
ELMOD3	615427	96	100	100	97
ELOVL1	611813	82	100	99	96
EPS8	600206	68	100	100	94
EPS8L2	614988	132	100	97	95
ERAL1	607435	132	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ESPN	606351	101	97	88	81
ESRP1	612959	67	100	98	93
ESRRB	602167	142	100	100	100
EXOSC2	602238	69	100	100	97
EYA1	601653	86	100	100	98
EYA4	603550	62	100	100	94
FGF3	164950	122	100	100	100
FOXF2	603250	109	95	91	89
FOXI1	601093	161	100	100	100
GAB1	604439	72	100	99	97
GATA3	131320	203	100	100	100
GIPC3	608792	115	100	95	93
GJB2	121011	150	100	100	100
GJB3	603324	182	100	100	100
GJB6	604418	75	100	100	99
GPSM2	609245	93	100	100	96
GRAP	604330	52	68	68	65
GREB1L	617782	72	100	98	93
GRHL2	608576	81	100	100	99
GRXCR1	613283	100	100	100	99
GRXCR2	615762	109	100	100	100
GSDME	608798	94	100	100	100
HARS1	142810	104	100	100	99
HARS2	600783	76	100	100	99
HGF	142409	73	100	100	96
HOMER2	604799	121	99	98	98
HSD17B4	601860	62	100	98	93
IFNLR1	607404	92	100	97	95
ILDR1	609739	95	100	100	97
KARS1	601421	126	100	100	97
KCNE1	176261	252	100	100	100
KCNJ10	602208	171	100	100	100
KCNQ1	607542	160	97	94	93
KCNQ4	603537	147	98	95	91
KITLG	184745	78	100	100	97

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
LARS2	604544	79	100	100	99
LHFPL5	609427	175	100	100	100
LMX1A	600298	81	100	100	99
LOXHD1	613072	118	100	100	99
LRTOMT	612414	111	100	100	100
MARVELD2	610572	92	100	100	97
MCM2	116945	122	100	100	100
MET	164860	64	100	100	96
MIR96	611606	No coverage data			
MITF	156845	87	100	100	99
MPZL2	604873	64	100	100	98
MSRB3	613719	72	100	100	93
MYH14	608568	114	100	99	97
MYH9	160775	125	100	100	99
MYO15A	602666	134	100	99	98
MYO3A	606808	73	100	99	95
MYO6	600970	82	100	99	94
MYO7A	276903	109	100	100	99
NARS2	612803	55	99	97	92
NLRP3	606416	125	100	100	100
OPA1	605290	59	100	98	89
OSBPL2	606731	102	100	100	100
OTOA	607038	78	77	74	72
OTOF	603681	113	100	100	100
OTOG	604487	121	100	100	99
OTOGL	614925	76	100	100	95
P2RX2	600844	136	100	100	99
PAX3	606597	104	100	100	100
PCDH15	605514	72	100	100	95
PDE1C	602987	63	100	97	90
PDZD7	612971	84	100	100	99
PET100	614770	85	92	66	66
PEX1	602136	62	100	99	94
PEX26	608666	118	100	100	100
PEX6	601498	107	100	99	95

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
PJVK	610219	93	100	100	99
PNPT1	610316	57	100	97	83
POU3F4	300039	120	100	100	100
POU4F3	602460	247	100	100	100
PPIP5K2	611648	48	99	93	80
PRKCB	176970	73	100	100	99
PRPS1	311850	50	100	99	90
PTPRQ	603317	80	100	99	95
RAI1	607642	187	100	100	100
RDX	179410	54	100	93	76
REST	600571	69	100	100	97
RIPOR2	611410	77	100	100	97
ROR1	602336	87	97	96	96
S1PR2	605111	153	100	100	100
SERPINB6	173321	85	100	100	98
SIX1	601205	119	100	100	100
SIX5	600963	75	100	95	85
SLC17A8	607557	66	100	100	99
SLC22A4	604190	103	100	100	96
SLC25A2	608157	127	100	100	89
SLC26A4	605646	68	100	100	94
SLC26A5	604943	61	100	100	96
SLC29A3	612373	152	100	99	99
SLC33A1	603690	68	100	98	86
SLC44A4	606107	94	100	100	100
SLC52A2	607882	173	100	100	100
SLITRK6	609681	75	100	100	99
SMPX	300226	51	100	100	83
SNAI2	602150	66	100	100	100
SOX10	602229	72	100	97	89
SPATA5	613940	68	100	100	97
SPNS2	612584	123	96	93	91
STRC	606440	66	53	48	46
SYNE4	615535	107	100	100	100
TBC1D24	613577	158	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
TECTA	602574	123	100	100	98
TIMM8A	300356	138	100	100	100
TJP2	607709	83	100	100	99
TMC1	606706	61	100	100	94
TMEM132E	616178	117	100	99	96
TMIE	607237	85	100	100	99
TMPRSS3	605511	74	100	100	99
TMTC2	615856	79	100	100	98
TNC	187380	110	100	100	98
TPRN	613354	94	92	86	79
TRIOBP	609761	182	100	99	97
TSPEAR	612920	105	100	100	100
TWNK	606075	145	100	100	100
TYR	606933	83	100	100	98
USH1C	605242	80	100	98	92
USH1G	607696	180	100	100	100
USH2A	608400	82	100	100	98
WBP2	606962	60	100	96	89
WFS1	606201	196	100	100	100
WHRN	607928	119	100	100	100
YAP1	606608	73	100	99	94

- 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