

# Whole Exome Sequencing

## Gene package Hereditary Congenital Defects, version 7, 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 ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
<i>Oesophagus atresia</i>					
CHD7	608892	100	100	99.89	98.69
EFTUD2	603892	100	100	100	97.47
GLI3	165240	100	100	100	99.89
MID1	300552	100	100	100	97.46
MYCN	164840	100	100	98.06	91.49
SOX2	184429	100	100	99.49	93.56
<i>Congenital Hernia Diaphragmatica</i>					
GATA4	600576	100	100	100	94.74
GPC3	300037	100	100	99.55	95.55
HLX	142995	100	100	98.54	91.32
LONP1	605490	100	99.29	97.44	91.37
LRP2	600073	100	99.96	99.64	97.62
MYRF	608329	99.79	97.35	93.93	87.17
NIPBL	608667	100	100	99.65	97.00
NR2F2	107773	100	100	100	98.23
SMC3	606062	100	99.98	97.35	89.41
SOX7	612202	100	100	99.75	94.56
STRA6	610745	100	100	100	96.01
WT1	607102	100	100	100	100
ZFPM2	603693	100	100	99.70	95.45

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
<i>Ano-rectal Malformation</i>					
CASK	300172	100	99.97	97.02	90.61
CCNB1	123836	100	100	100	100
CCNQ	300708	81.09	81.09	81.09	81.09
CDX1	600746	100	96.49	81.75	63.74
CDX2	600297	100	98.00	93.39	75.03
FLNA	300017	99.95	99.05	97.51	95.12
GLI3	165240	100	100	100	99.89
HOXD13	142989	100	99.11	93.92	85.31
JAG1	601920	100	98.67	97.58	97.41
KDM6A	300128	100	100	99.30	92.95
KMT2D	602113	100	100	99.75	97.30
MED12	300188	100	99.89	98.40	93.18
MID1	300552	100	100	100	97.46
MNX1	142994	100	100	97.05	67.62
MYCN	164840	100	100	98.06	91.49
NOTCH2	600275	99.86	98.85	98.02	95.09
SALL1	602218	100	100	100	99.07
SALL4	607343	100	99.35	95.66	95.37
USP9X	300072	100	100	99.83	96.43
<i>HSCR/CIPO/MMIHS</i>					
AAAS	605378	100	100	100	99.95
ACTG2	102545	100	100	100	100
EDN3	131242	100	100	99.02	83.05
EDNRB	131244	100	99.83	98.58	95.24
FLNA	300017	99.95	99.05	97.51	95.12
KIFBP	609367	100	100	100	93.41
LMOD1	602715	100	100	100	95.73
MYH11	160745	100	99.67	98.35	91.31
MYL9	609905	100	100	100	99.48
MYLK	600922	100	99.86	98.82	95.65
NOS1	163731	100	99.60	98.22	93.26
PHOX2B	603851	93.16	83.78	74.85	55.94
RAD21	606462	100	100	99.70	96.40
RET	164761	100	99.85	97.07	91.06
SGO1	609168	100	99.27	91.81	91.81
SOX10	602229	100	99.66	97.70	90.57
VCL	193065	100	100	99.87	97.51
ZEB2	605802	100	100	100	100

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
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- 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