Whole Exome Sequencing
Gene package Hereditary Congenital Defects, version 6, 30-9-2021

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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
Oesophagus atresia					
CHD7	608892	100	100	99.90	98.10
EFTUD2	603892	100	100	100	97.84
GLI3	165240	100	100	100	99.98
MID1	300552	100	100	100	95.23
MYCN	164840	100	100	97.68	91.49
SOX2	184429	100	100	99.23	93.41
Congenital Hernia Diaphragmatica					
GATA4	600576	100	100	100	94.62
GPC3	300037	100	100	98.72	90.20
LRP2	600073	100	99.89	99.21	97.47
MYRF	608329	99.66	96.86	93.60	87.34
NIPBL	608667	100	99.98	99.38	96.10
NR2F2	107773	100	100	100	98.56
SMC3	606062	100	99.68	96.35	87.44
STRA6	610745	100	100	100	97.57
WT1	607102	100	100	100	100

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ZFPM2	603693	100	100	99.07	94.84
Ano-rectal Marformation					
CASK	300172				
CCNB1	123836	100	100	100	100
CCNQ	300708	81.09	81.09	81.09	81.09
FLNA	300017	99.98	98.83	97.47	92.13
GLI3	165240	100	100	100	99.98
JAG1	601920	100	98.32	97.58	97.57
KDM6A	300128	100	99.78	97.98	86.21
KMT2D	602113	100	100	99.94	98.49
MED12	300188	100	99.82	97.42	89.88
MID1	300552	100	100	100	95.23
MNX1	142994	100	100	95.93	54.70
MYCN	164840	100	100	97.68	91.49
NOTCH2	600275	99.88	98.85	98.47	95.83
SALL1	602218	100	100	100	98.69
SALL4	607343	100	99.10	95.37	95.37
USP9X	300072	100	99.95	99.10	92.29
HSCR/CIPO/MMIHS					
AAAS	605378	100	100	100	100
ACTG2	102545	100	100	100	100
EDN3	131242	100	100	93.30	83.05
EDNRB	131244	100	99.80	98.80	95.27
FLNA	300017	99.98	98.83	97.47	92.13
KIFBP	609367	100	100	100	94.81
LMOD1	602715	100	100	100	97.28
MYH11	160745	100	99.84	98.72	92.21
MYL9	609905	100	100	100	99.13
MYLK	600922	100	99.97	98.92	95.89
NOS1	163731	100	99.82	98.97	94.76
PHOX2B	603851	93.51	81.44	70.46	55.14
RAD21	606462	100	100	99.58	96.96
RET	164761	100	99.44	97.03	91.74
SGO1	609168	100	91.81	91.81	91.81
SOX10	602229	100	100	98.32	91.53
VCL	193065	100	100	99.83	96.49
ZEB2	605802	100	100	100	100

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- OMIM release used: 23-9-2021
- The statistics above are based on a set of 150 samples
- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x