Whole Exome Sequencing
Gene package Congenital Heart Defects (CHD), version 6.3, 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ACTC1	102540	100	100	100	98.96
ACVR1	102576	100	100	100	98.54
ACVR2B	602730	100	98.69	95.90	95.90
ADAMTS19	607513	100	99.54	95.24	88.22
ANKRD1	609599	100	100	97.58	92.57
CFAP53	614759	99.38	96.74	95.12	87.46
CFC1	605194	33.46	33.46	32.70	27.88
CHD7	608892	100	100	99.89	98.69
CITED2	602937	100	100	99.48	87.23
CRELD1	607170	99.87	96.60	89.99	70.83
DCHS1	603057	100	100	100	99.67
DNAAF1	613190	100	100	100	96.17
ELN	130160	100	98.14	92.73	71.74
FLNA	300017	99.95	99.05	97.51	95.12
FLT4	136352	100	98.74	98.12	92.43
FOXH1	603621	100	100	100	100
GATA4	600576	100	100	100	94.74
GATA5	611496	100	99.62	96.45	79.18
GATA6	601656	100	94.72	89.71	78.74

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GDF1	602880	100	87.20	74.01	59.86
GJA1	121014	100	100	100	100
HAND1	602406	100	100	91.53	76.57
HAND2	602407	100	100	99.57	89.15
HEY2	604674	100	100	100	99.19
IRX4	606199	100	100	96.30	88.29
JAG1	601920	100	98.67	97.58	97.41
LEFTY2	601877	100	100	100	97.54
MATR3	164015	100	100	100	97.96
MED13L	608771	100	100	99.35	96.30
MMP21	608416	100	97.78	93.99	84.87
MYH11	160745	100	99.67	98.35	91.31
MYH6	160710	100	99.84	98.83	93.63
MYH7	160760	100	100	99.54	93.45
MYOCD	606127	100	100	99.47	97.17
NFATC1	600489	100	100	99.37	95.15
NKX2-5	600584	100	99.19	91.99	52.74
NKX2-6	611770	100	100	100	100
NODAL	601265	100	100	100	100
NOTCH1	190198	99.03	98.46	97.39	92.00
NOTCH2	600275	99.86	98.85	98.02	95.09
NR2F2	107773	100	100	100	98.23
PKD1L1	609721	100	99.88	99.24	94.71
PRDM6	616982	100	97.51	87.01	71.06
PTPN11	176876	98.37	98.37	98.37	98.37
ROBO1	602430	100	100	99.91	98.15
ROBO4	607528	100	100	99.44	93.12
SMAD2	601366	100	100	100	98.09
SMAD6	602931	100	87.44	66.07	48.41
SMO	601500	100	97.31	95.12	89.20
SOS1	182530	100	99.96	97.71	96.03
TAB2	605101	100	100	100	99.73
TBX1	602054	91.65	78.18	67.92	47.40
TBX20	606061	100	100	100	96.27
TBX5	601620	100	100	100	97.78
TDGF1	187395	100	100	100	100
TFAP2B	601601	100	99.21	97.47	90.92
TLL1	606742	100	100	100	97.70
ZFPM2	603693	100	100	99.70	95.45
ZIC3	300265	100	100	100	98.56

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

- OMIM release used: 18-2-2021
- 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 (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x