

Whole Exome Sequencing

Gene package Congenital Heart Defects (CHD), version 6.1, 26-2-2021



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).



Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
ACTC1	102540	100	100	100	95.28
ACVR1	102576	100	100	100	96.28
ACVR2B	602730	100	97.35	95.90	95.64
ADAMTS19	607513	100	96.71	92.74	84.25
ANKRD1	609599	100	97.45	93.93	84.48
CFAP53	614759	98.91	95.59	93.89	84.72
CFC1	605194	33.46	33.46	30.16	24.59
CHD7	608892	100	99.87	98.61	96.80
CITED2	602937	100	100	98.44	85.43
CRELD1	607170	100	96.40	88.73	71.53
DCHS1	603057	100	100	100	98.84
DNAAF1	613190	100	100	98.43	90.48
ELN	130160	100	97.40	90.82	66.16
FLNA	300017	100	99.43	98.63	95.60
FLT4	136352	100	98.57	97.41	90.26
FOXH1	603621	100	100	100	99.61
GATA4	600576	100	100	100	90.59
GATA5	611496	100	99.01	94.01	67.43
GATA6	601656	100	94.46	89.58	77.53

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
GDF1	602880	100	84.78	73.92	57.87
GJA1	121014	100	100	100	100
HAND1	602406	100	98.32	85.26	73.07
HAND2	602407	100	100	98.77	70.19
HEY2	604674	100	100	99.86	97.66
IRX4	606199	100	100	96.30	84.99
JAG1	601920	100	98.20	97.58	96.60
LEFTY2	601877	100	100	99.53	96.86
MATR3	164015	100	100	99.50	94.28
MED13L	608771	100	99.87	98.57	94.09
MMP21	608416	100	97.08	92.61	82.27
MYH11	160745	100	99.60	98.37	92.07
MYH6	160710	100	99.80	98.65	93.05
MYH7	160760	100	100	99.37	93.53
MYOCD	606127	100	100	99.72	96.57
NFATC1	600489	100	99.92	99.01	95.08
NKX2-5	600584	100	97.26	86.00	31.85
NKX2-6	611770	100	100	100	100
NODAL	601265	100	100	100	100
NOTCH1	190198	99.03	98.04	96.96	89.26
NOTCH2	600275	99.78	98.49	97.37	93.59
NR2F2	107773	100	100	100	97.12
PKD1L1	609721	100	99.43	97.87	90.64
PRDM6	616982	100	95.79	85.71	67.45
PTPN11	176876	98.37	98.37	98.37	98.28
ROBO1	602430	100	100	99.69	97.20
ROBO4	607528	100	100	99.05	88.33
SMAD2	601366	100	100	100	96.91
SMAD6	602931	99.23	78.32	62.37	47.07
SMO	601500	100	96.46	94.16	85.62
SOS1	182530	100	97.71	97.35	91.97
TAB2	605101	100	100	100	98.86
TBX1	602054	91.70	82.58	69.57	45.38
TBX20	606061	100	100	100	94.64
TBX5	601620	100	100	99.24	92.94
TDGF1	187395	100	100	100	100
TFAP2B	601601	100	99.28	97.24	91.02
TLL1	606742	100	100	99.38	92.96
ZFPM2	603693	100	100	98.33	91.93
ZIC3	300265	100	100	100	94.18

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

- OMIM release used: 18-2-2021
- The statistics above are based on a set of 100 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