Whole Exome Sequencing
Gene package Congenital Heart Defects (CHD), version 5, 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
ACTC1	102540	194	100	100	100
ACVR1	102576	73	100	100	99
ACVR2B	602730	105	100	100	98
ADAMTS19	607513	67	100	93	86
ANKRD1	609599	91	100	100	97
CFAP53	614759	98	100	100	98
CFC1	605194	70	41	34	28
CHD7	608892	87	100	100	97
CITED2	602937	93	100	100	100
CRELD1	607170	100	100	100	100
DCHS1	603057	128	100	100	100
DNAAF1	613190	131	100	100	99
ELN	130160	85	100	100	98
FLNA	300017	110	100	100	100
FLT4	136352	132	100	100	100
FOXH1	603621	78	100	100	97

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
GATA4	600576	81	100	87	80
GATA5	611496	78	100	99	95
GATA6	601656	128	100	95	91
GDF1	602880	69	100	93	81
GJA1	121014	101	100	100	100
HAND1	602406	146	100	100	98
HAND2	602407	99	100	100	100
HEY2	604674	160	100	99	96
IRX4	606199	149	98	96	96
JAG1	601920	96	100	100	96
LEFTY2	601877	144	100	100	99
MATR3	164015	63	100	100	95
MED13L	608771	73	100	100	97
MMP21	608416	81	100	96	88
MYH11	160745	136	100	100	99
MYH6	160710	145	100	99	98
MYH7	160760	176	100	100	100
MYOCD	606127	121	100	100	100
NKX2-5	600584	101	100	100	97
NKX2-6	611770	145	100	100	100
NODAL	601265	119	100	100	100
NOTCH1	190198	117	100	99	98
NOTCH2	600275	98	100	99	98
NR2F2	107773	232	100	100	100
PKD1L1	609721	69	100	98	91
PRDM6	616982	85	100	100	98
PTPN11	176876	77	100	99	90
ROBO1	602430	102	100	100	98
ROBO4	607528	91	100	100	98
SMAD2	601366	69	100	100	97
SMAD6	602931	165	100	97	87
SMO	601500	124	100	100	100
SOS1	182530	71	100	100	95
TAB2	605101	76	100	100	99
TBX1	602054	94	94	85	80

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
TBX20	606061	80	100	100	97
TBX5	601620	77	100	100	99
TDGF1	187395	127	100	100	100
TFAP2B	601601	142	100	100	100
TLL1	606742	67	100	100	96
ZFPM2	603693	82	100	100	100
ZIC3	300265	101	100	100	97

- 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