Whole Exome Sequencing
Gene package Congenital Heart Defects (CHD), version 6, 30-9-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + 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 Alissa Interpret 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	173	100	100	100
ACVR1	102576	104	100	100	99
ACVR2B	602730	272	100	100	100
ADAMTS19	607513	87	99	96	90
ANKRD1	609599	83	87	86	85
CFAP53	614759	78	89	80	74
CFC1	605194	59	33	33	30
CHD7	608892	167	100	99	97
CITED2	602937	154	100	100	100
CRELD1	607170	246	100	99	98
DCHS1	603057	292	100	100	100
DNAAF1	613190	146	100	100	97
ELN	130160	171	100	100	100
FLNA	300017	206	100	100	100
FLT4	136352	284	100	100	100
FOXH1	603621	355	100	100	100
GATA4	600576	164	100	100	100
GATA5	611496	213	100	100	100
GATA6	601656	183	100	96	92
GDF1	602880	150	100	100	99
GJA1	121014	143	100	100	100
HAND1	602406	246	100	100	100
HAND2	602407	217	100	100	100
HEY2	604674	246	100	98	94
IRX4	606199	206	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
JAG1	601920	213	100	100	100
LEFTY2	601877	342	100	100	100
MATR3	164015	86	98	92	85
MED13L	608771	124	100	100	100
MMP21	608416	151	100	100	99
MYH11	160745	171	100	100	99
MYH6	160710	225	100	100	100
MYH7	160760	221	100	100	100
MYOCD	606127	150	100	100	100
NFATC1	600489	265	100	100	100
NKX2-5	600584	223	100	100	100
NKX2-6	611770	265	100	100	100
NODAL	601265	262	100	100	100
NOTCH1	190198	318	100	99	99
NOTCH2	600275	188	100	98	97
NR2F2	107773	270	100	100	98
PKD1L1	609721	142	100	99	97
PRDM6	616982	152	100	100	98
PTPN11	176876	80	98	94	89
ROBO1	602430	133	100	100	99
ROBO4	607528	199	100	100	100
SMAD2	601366	87	100	100	94
SMAD6	602931	188	100	100	100
SMO	601500	213	100	100	100
SOS1	182530	74	94	89	79
TAB2	605101	120	100	99	93
TBX1	602054	131	94	90	87
TBX20	606061	158	100	100	100
TBX5	601620	233	100	100	98
TDGF1	187395	148	100	100	100
TFAP2B	601601	162	100	100	100
TLL1	606742	82	100	97	91
ZFPM2	603693	127	100	100	99
ZIC3	300265	137	100	100	99

- 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