# Whole Exome Sequencing
# Gene package Congenital Heart Defects (CHD), version 6.2, 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 |
|---|---|---|---|---|---|
| ACTC1 | 102540 | 100 | 100 | 100 | 97.48 |
| ACVR1 | 102576 | 100 | 100 | 100 | 99.36 |
| ACVR2B | 602730 | 100 | 96.13 | 95.90 | 95.90 |
| ADAMTS19 | 607513 | 100 | 99.44 | 94.94 | 87.44 |
| ANKRD1 | 609599 | 100 | 100 | 96.57 | 88.04 |
| CFAP53 | 614759 | 98.88 | 95.77 | 94.24 | 87.49 |
| CFC1 | 605194 | 33.46 | 33.46 | 32.95 | 28.01 |
| CHD7 | 608892 | 100 | 100 | 99.90 | 98.10 |
| CITED2 | 602937 | 100 | 100 | 100 | 88.27 |
| CRELD1 | 607170 | 100 | 96.73 | 91.96 | 72.70 |
| DCHS1 | 603057 | 100 | 100 | 100 | 99.83 |
| DNAAF1 | 613190 | 100 | 100 | 100 | 94.45 |
| ELN | 130160 | 100 | 99.13 | 95.08 | 75.43 |
| FLNA | 300017 | 99.98 | 98.83 | 97.47 | 92.13 |
| FLT4 | 136352 | 100 | 98.34 | 98.09 | 93.67 |
| FOXH1 | 603621 | 100 | 100 | 100 | 100 |
| GATA4 | 600576 | 100 | 100 | 100 | 94.62 |
| GATA5 | 611496 | 100 | 99.39 | 96.00 | 78.11 |
| GATA6 | 601656 | 100 | 97.11 | 90.97 | 76.17 |

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|---|---|---|---|---|---|
| GDF1 | 602880 | 100 | 92.13 | 78.94 | 60.08 |
| GJA1 | 121014 | 100 | 100 | 100 | 100 |
| HAND1 | 602406 | 100 | 100 | 88.03 | 77.52 |
| HAND2 | 602407 | 100 | 100 | 99.28 | 76.99 |
| HEY2 | 604674 | 100 | 100 | 100 | 98.47 |
| IRX4 | 606199 | 100 | 99.74 | 96.30 | 84.81 |
| JAG1 | 601920 | 100 | 98.32 | 97.58 | 97.57 |
| LEFTY2 | 601877 | 100 | 100 | 100 | 97.71 |
| MATR3 | 164015 | 100 | 100 | 100 | 97.93 |
| MED13L | 608771 | 100 | 99.89 | 98.72 | 96.49 |
| MMP21 | 608416 | 100 | 97.13 | 93.58 | 86.65 |
| MYH11 | 160745 | 100 | 99.84 | 98.72 | 92.21 |
| MYH6 | 160710 | 100 | 99.86 | 99.33 | 95.20 |
| MYH7 | 160760 | 100 | 100 | 99.94 | 94.93 |
| MYOCD | 606127 | 100 | 100 | 99.94 | 97.73 |
| NFATC1 | 600489 | 100 | 99.83 | 98.40 | 95.15 |
| NKX2-5 | 600584 | 100 | 98.78 | 90.26 | 42.09 |
| NKX2-6 | 611770 | 100 | 100 | 100 | 100 |
| NODAL | 601265 | 100 | 100 | 100 | 100 |
| NOTCH1 | 190198 | 99.03 | 98.64 | 97.87 | 94.58 |
| NOTCH2 | 600275 | 99.88 | 98.85 | 98.47 | 95.83 |
| NR2F2 | 107773 | 100 | 100 | 100 | 98.56 |
| PKD1L1 | 609721 | 100 | 99.98 | 99.19 | 94.93 |
| PRDM6 | 616982 | 100 | 93.25 | 81.58 | 68.18 |
| PTPN11 | 176876 | 98.37 | 98.37 | 98.37 | 98.37 |
| ROBO1 | 602430 | 100 | 100 | 100 | 98.66 |
| ROBO4 | 607528 | 100 | 100 | 99.88 | 96.30 |
| SMAD2 | 601366 | 100 | 100 | 100 | 98.69 |
| SMAD6 | 602931 | 100 | 85.24 | 60.94 | 46.59 |
| SMO | 601500 | 100 | 96.71 | 94.39 | 89.16 |
| SOS1 | 182530 | 100 | 98.47 | 97.56 | 94.16 |
| TAB2 | 605101 | 100 | 100 | 100 | 99.55 |
| TBX1 | 602054 | 89.95 | 75.58 | 63.04 | 42.43 |
| TBX20 | 606061 | 100 | 100 | 100 | 96.34 |
| TBX5 | 601620 | 100 | 100 | 100 | 97.99 |
| TDGF1 | 187395 | 100 | 100 | 100 | 100 |
| TFAP2B | 601601 | 100 | 99.47 | 97.43 | 91.74 |
| TLL1 | 606742 | 100 | 100 | 100 | 96.82 |
| ZFPM2 | 603693 | 100 | 100 | 99.07 | 94.84 |
| ZIC3 | 300265 | 100 | 100 | 100 | 92.40 |

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|---|---|---|---|---|---|

- OMIM release used: 18-2-2021
- 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