Whole Exome Sequencing
Gene package Cardiomyopathy, version 11.1, 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
ACAD9	611103	100	100	100	99.64
ACTC1	102540	100	100	100	98.96
ACTN2	102573	100	100	100	97.87
ALPK3	617608	97.19	96.93	96.57	92.04
ANO5	608662	100	99.81	98.88	93.14
BAG3	603883	100	100	99.25	95.93
CDH2	114020	99.13	97.37	96.07	90.44
CRYAB	123590	100	100	100	100
CSRP3	600824	100	100	100	100
DES	125660	100	100	99.87	95.38
DSC2	125645	100	99.62	97.06	95.20
DSG2	125671	100	98.22	97.26	94.16
DSP	125647	100	100	100	99.90
DTNA	601239	100	100	100	100
EMD	300384	99.38	95.35	85.49	64.29
EYA4	603550	100	100	100	100
FHL1	300163	100	100	100	97.34
FHOD3	609691	100	99.60	97.62	91.04

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FKTN	607440	100	100	99.84	97.44
FLNC	102565	100	100	99.67	95.43
GATAD1	614518	100	100	100	96.09
GLA	300644	100	100	100	98.28
HCN4	605206	100	99.75	97.81	88.26
ILK	602366	99.94	99.94	99.94	99.94
JPH2	605267	100	100	100	100
JUP	173325	100	100	99.94	97.11
KIF20A	605664	100	100	100	99.83
LAMP2	309060	100	100	100	91.30
LMNA	150330	100	100	98.66	80.67
MIB1	608677	100	100	99.14	94.68
MYBPC3	600958	100	99.36	96.98	93.53
MYH6	160710	100	99.84	98.83	93.63
MYH7	160760	100	100	99.54	93.45
MYL2	160781	100	100	100	100
MYL3	160790	100	100	100	83.55
MYLK2	606566	100	100	100	98.55
MYOZ1	605603	100	100	100	97.49
MYOZ2	605602	100	100	100	97.87
MYPN	608517	100	100	99.56	97.87
NEBL	605491	100	100	100	100
NEXN	613121	100	98.72	90.71	80.57
NKX2-5	600584	100	99.19	91.99	52.74
PDLIM3	605889	100	100	100	100
PKP2	602861	100	100	97.81	91.33
PLN	172405	100	100	100	100
PPA2	609988	100	99.65	94.74	78.80
PPCS	609853	100	100	100	90.74
PPP1R13L	607463	100	97.54	89.90	68.04
PRDM16	605557	100	99.20	97.04	92.43
PRKAG2	602743	100	97.14	97.14	91.05
PTPN11	176876	98.37	98.37	98.37	98.37
RAF1	164760	100	100	100	96.29
RBM20	613171	100	100	100	94.23
RYR2	180902	100	99.94	99.21	96.74
SCN5A	600163	100	100	99.74	97.78
SGCD	601411	100	100	100	98.57
SOS1	182530	100	99.96	97.71	96.03
TAFAZZIN	300394	No coverage data			

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TBX20	606061	100	100	100	96.27
TCAP	604488	100	100	100	95.38
TGFB3	190230	100	100	100	96.88
TMEM43	612048	100	100	100	97.95
TNNC1	191040	100	100	100	98.18
TNNI3	191044	100	100	100	88.10
TNNI3K	613932	100	100	100	99.23
TNNT2	191045	100	100	99.67	89.58
TPM1	191010	100	95.83	87.72	82.68
TRIM63	606131	100	100	100	97.62
TTN	188840	100	100	100.00	99.80
TTR	176300	100	100	100	100
TXNRD2	606448	98.95	94.69	93.57	89.70
VCL	193065	100	100	99.87	97.51

- OMIM release used: 23-9-2021
- 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