Whole Exome Sequencing
Gene package Cardiomyopathy, version 9, 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
ACAD9	611103	186	100	100	100
ACTC1	102540	173	100	100	100
ACTN2	102573	154	100	100	100
ALPK3	617608	243	100	98	97
ANO5	608662	61	94	87	75
BAG3	603883	212	100	100	100
CASQ2	114251	102	100	98	96
CRYAB	123590	160	100	100	100
CSRP3	600824	176	100	100	100
DES	125660	178	100	100	100
DSC2	125645	84	99	93	89
DSG2	125671	104	96	93	86
DSP	125647	411	100	100	99
DTNA	601239	109	100	99	96
EMD	300384	184	100	98	95
EYA4	603550	93	100	95	90
FHL1	300163	133	100	100	99
FHOD3	609691	141	100	98	94
FKTN	607440	104	90	81	68
FLNC	102565	290	100	100	100
GATAD1	614518	162	100	100	99
GLA	300644	87	100	100	98
HCN4	605206	306	100	100	100
ILK	602366	178	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
JPH2	605267	287	100	100	100
JUP	173325	266	100	100	100
KIF20A	605664	143	100	100	100
LAMP2	309060	54	100	89	76
LMNA	150330	238	100	100	100
MIB1	608677	96	100	98	94
MYBPC3	600958	272	100	100	100
MYH6	160710	225	100	100	100
MYH7	160760	221	100	100	100
MYL2	160781	172	100	100	100
MYL3	160790	219	100	100	100
MYLK2	606566	193	100	100	100
MYOZ1	605603	151	100	100	100
MYOZ2	605602	71	100	96	85
MYPN	608517	119	100	99	97
NEBL	605491	67	99	93	83
NEXN	613121	30	92	66	41
NKX2-5	600584	223	100	100	100
PDLIM3	605889	183	100	100	100
PKP2	602861	150	100	100	100
PLN	172405	43	100	99	85
PPA2	609988	58	89	72	58
PPCS	609853	109	100	99	96
PPP1R13L	607463	219	100	100	100
PRDM16	605557	245	100	100	100
PRKAG2	602743	154	93	90	86
PTPN11	176876	80	98	94	89
RAF1	164760	124	100	98	97
RBM20	613171	248	100	100	98
RYR2	180902	133	99	94	90
SCN5A	600163	208	100	100	99
SGCD	601411	80	100	99	94
SOS1	182530	74	94	89	79
TAZ	300394	145	100	100	100
TBX20	606061	158	100	100	100
TCAP	604488	300	100	100	100
TGFB3	190230	182	100	100	100
TMEM43	612048	199	100	100	99
TNNC1	191040	309	100	100	100
TNNI3	191044	218	100	100	100
TNNI3K	613932	97	100	95	91
TNNT2	191045	141	100	100	100
TPM1	191010	128	100	97	87

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
TRIM63	606131	141	100	100	100
TTN	188840	136	99	98	96
TTR	176300	168	100	100	100
TXNRD2	606448	216	100	100	99
VCL	193065	137	100	99	96

- 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