Whole Exome Sequencing
Gene package Cardiomyopathy, version 8.1, 31-1-2020

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	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ABCC9	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850	601439	65	100	100	95
ACAD9	Mitochondrial complex I deficiency, nuclear type 20, 611126	611103	95	100	100	100
ACTC1	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424	102540	194	100	100	100
ACTN2	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158	102573	94	100	100	99
ALPK3	Cardiomyopathy, familial hypertrophic 27, 618052	617608	117	100	97	96
ANKRD1	No OMIM phenotype	609599	91	100	100	97
ANO5	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle 12, 611307	608662	77	100	100	96

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BAG3	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954	603883	196	100	100	100
CALR3	No OMIM phenotype	611414	67	100	100	97
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938	114251	65	100	100	97
CAV3	Cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease 2, 606072	601253	150	100	100	100
CRYAB	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869	123590	79	100	100	100
CSRP3	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124	600824	102	100	100	100
CTNNA3	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616	607667	61	100	100	96
DES	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400	125660	106	100	100	100
DSC2	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476	125645	184	100	100	100
DSG2	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877	125671	158	100	100	99
DSP	Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655	125647	156	100	100	100
DTNA	Left ventricular noncompaction 1, with or without congenital heart defects, 604169	601239	84	100	100	100
EMD	Emery-Dreifuss muscular dystrophy 1, 310300	300384	105	100	100	100
EYA4	?Cardiomyopathy, dilated, 1J, 605362 Deafness 10, 601316	603550	62	100	100	94

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FHL1	Emery-Dreifuss muscular dystrophy 6, 300696 Myopathy, with postural muscle atrophy, 300696 Reducing body myopathy 1a, severe, infantile or early childhood onset, 300717 Reducing body myopathy 1b, with late childhood or adult onset, 300718 Scapuloperoneal myopathy dominant, 300695 ?Uruguay faciocardiomusculoskeletal syndrome, 300280	300163	92	100	100	94
FHOD3	No OMIM phenotype	609691	89	100	99	95
FKTN	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588	607440	85	100	100	100
FLNC	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524	102565	133	100	100	100
GATAD1	?Cardiomyopathy, dilated, 2B, 614672	614518	73	100	100	97
GLA	Fabry disease, 301500 Fabry disease, cardiac variant, 301500	300644	101	100	100	100
HCN4	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800	605206	103	100	100	99
ILK	No OMIM phenotype	602366	103	100	100	99
JPH2	Cardiomyopathy, hypertrophic, 17, 613873	605267	109	100	100	98
JUP	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214	173325	106	100	100	98
KIF20A	No OMIM phenotype	605664	74	100	99	95
LAMA4	Cardiomyopathy, dilated, 1JJ, 615235	600133	71	100	100	97
LAMP2	Danon disease, 300257	309060	44	100	95	77
LDB3	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452	605906	123	100	100	100

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LMNA	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, 181350 Emery-Dreifuss muscular dystrophy 3, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Restrictive dermopathy, lethal, 275210	150330	143	100	100	100
MIB1	Left ventricular noncompaction 7, 615092	608677	69	100	100	97
MYBPC3	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396	600958	169	100	100	98
MYH6	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090	160710	145	100	99	98
MYH7	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Laing distal myopathy, 160500 Left ventricular noncompaction 5, 613426 Myopathy, myosin storage, 608358 Myopathy, myosin storage, 255160 Scapuloperoneal syndrome, myopathic type, 181430	160760	176	100	100	100
MYL2	Cardiomyopathy, hypertrophic, 10, 608758	160781	170	100	100	100
MYL3	Cardiomyopathy, hypertrophic, 8, 608751	160790	118	100	100	100
MYLK2	Cardiomyopathy, hypertrophic, 1, digenic, 192600	606566	111	100	100	100
MYOZ1	No OMIM phenotype	605603	83	100	100	96
MYOZ2	Cardiomyopathy, hypertrophic, 16, 613838	605602	78	100	100	100
MYPN	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248 Nemaline myopathy 11, 617336	608517	80	100	100	97

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NEBL	No OMIM phenotype	605491	57	100	99	91
NEXN	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876	613121	85	100	100	99
NKX2-5	Atrial septal defect 7, with or without AV conduction defects, 108900 Conotruncal heart malformations, variable, 217095 Hypoplastic left heart syndrome 2, 614435 Hypothyroidism, congenital nongoitrous, 5, 225250 Tetralogy of Fallot, 187500 Ventricular septal defect 3, 614432	600584	101	100	100	97
PDLIM3	No OMIM phenotype	605889	130	100	100	100
PKP2	Arrhythmogenic right ventricular dysplasia 9, 609040	602861	147	100	98	96
PLN	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874	172405	112	100	100	100
PPA2	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222	609988	70	100	94	76
PPCS	Cardiomyopathy, dilated, 2C, 618189	609853	121	100	100	96
PRDM16	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373	605557	191	100	100	99
PRKAG2	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200	602743	123	100	100	97
PTPN11	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950	176876	77	100	99	90
RAF1	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553	164760	78	100	99	95
RBM20	Cardiomyopathy, dilated, 1DD, 613172	613171	136	100	100	100
RYR2	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772	180902	128	100	100	97

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SCN5A	Atrial fibrillation, familial, 10, 614022 Brugada syndrome 1, 601144 Cardiomyopathy, dilated, 1E, 601154 Heart block, nonprogressive, 113900 Heart block, progressive, type IA, 113900 Long QT syndrome-3, 603830 Sick sinus syndrome 1, 608567 {Sudden infant death syndrome, susceptibility to}, 272120 Ventricular fibrillation, familial, 1, 603829	600163	192	100	100	100
SGCD	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle 6, 601287	601411	60	100	100	97
SOS1	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733	182530	71	100	100	95
TAZ	Barth syndrome, 302060	300394	97	100	98	90
TBX20	Atrial septal defect 4, 611363	606061	80	100	100	97
TCAP	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle 7, 601954	604488	102	100	100	100
TGFB3	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582	190230	104	100	100	100
TMEM43	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302	612048	133	100	100	100
TNNC1	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243	191040	127	100	100	100
TNNI3	Cardiomyopathy, dilated, 1FF, 613286 ?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690	191044	165	100	100	97
TNNI3K	Cardiac conduction disease with or without dilated cardiomyopathy, 616117	613932	60	100	99	92
TNNT2	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494	191045	149	100	100	100
TPM1	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878	191010	145	100	100	100

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TRIM63	No OMIM phenotype	606131	94	100	100	97
TTN	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle 10, 608807 Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Salih myopathy, 611705 Tibial muscular dystrophy, tardive, 600334	188840	68	100	99	97
TTR	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680	176300	93	100	100	100
TXNRD2	?Glucocorticoid deficiency 5, 617825	606448	100	100	96	93
VCL	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255	193065	89	100	99	93

- Gene symbols according HGCN
- OMIM release used: 8-9-2019
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- 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