

## Combined Cardiac Panel

**Panel Gene List:** *ABCC9, ACTC1, ACTN2, AKAP9, ALMS1, ALPK3, ANK2, ANKRD1, BAG3, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CHRM2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GAA, GATA4, GATA5, GATA6, GATAD1, GJA5, GLA, GNB5, GPD1L, HCN4, HFE, HRAS, ILK, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE1L (KCNE5), KCNH2 (HERG), KCNJ2, KCNJ5, KCNJ8, KCNQ1, KRAS, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MAP2K1, MAP2K2, MIB1, MTND1, MTND5, MTND6, MTTD, MTTG, MTTH, MTTI, MTTK, MTTL1, MTTL2, MTTM, MTTQ, MTTT1, MTTT2, MTTT3, MURC (CAVIN4), MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NRAS, PDLIM3, PKP2, PLN, PPA2, PRDM16, PRKAG2, PTPN11, RAF1, RANGRF, RBM20, RIT1, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SGCD, SHOC2, SNTA1, SOS1, TAZ, TBX20, TCAP, TECRL, TGFB3, TMEM43, TMPO, TNNC1, TNNT1, TNNT2, TOR1AIP1, TPM1, TRDN, TRPM4, TTN, TTR, TXNRD2, VCL*

*Additional genes from our cardiology test menu may be added to this panel by selecting test code 935C.*

### Clinical Features:

Cardiac arrhythmias occur due to disruption of the heart's natural rhythm and cardiomyopathy is defined as disease of the heart muscle. In some individuals or families, the clinical picture may be complex or features of both these conditions may be present, in which case a broader approach to genetic testing may be helpful. Both phenotypes are genetically heterogeneous and have many different clinical presentations.

Several risk factors can predispose an individual to develop an arrhythmia, including genetic disorders, trauma, electrolyte imbalance and structural abnormalities of the heart. There are several different genetic arrhythmia disorders. **Arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVC)** affects the cardiac desmosome, which is a protein complex that maintains cell-to-cell connections and provides mechanical attachments between adjacent cells. ARVC is characterized by myocyte death and replacement by fat and fibrous tissue in the right ventricle, predisposing to ventricular tachyarrhythmia and sudden cardiac death.<sup>1,2</sup> **Brugada syndrome (BrS)** is caused by abnormal ion channel function and is characterized by ST segment elevation on ECG (leads V1-3) in the absence of structural heart disease. BrS is associated with increased risk for syncope, entricular tachyarrhythmia and sudden cardiac death.<sup>3-5</sup> **Catecholaminergic polymorphic ventricular tachycardia (CPVT)** is characterized by cardiac calcium channel dysfunction that is precipitated by stress-induced release of catecholamines.<sup>6-8</sup> **Long QT syndrome (LQTS)** is characterized by prolongation of the QT interval on ECG and is associated with increased risk for syncope, ventricular arrhythmia, and sudden cardiac death in individuals with normal heart structure.<sup>9-11</sup> Genetic predisposition to a cardiac arrhythmia may occur as an isolated feature or may be part of a multisystemic disorder, such as Jervell and Lange-

Nielsen syndrome, Timothy syndrome, Andersen-Tawil syndrome, Naxos disease, Carvajal syndrome and muscular dystrophy.<sup>1,12-15</sup>

Several risk factors may also contribute towards development of a cardiomyopathy, including genetic disorders, hypertension, ischemic heart disease and infection. There are several different genetic cardiomyopathies. **Hypertrophic cardiomyopathy (HCM)** is characterized by myocardial hypertrophy and myocyte disarray in the absence of other cardiac or systemic causes.<sup>16-18</sup> **Dilated cardiomyopathy (DCM)** usually presents with one or more of the following: i) heart failure with symptoms of congestion (edema, orthopnea or paroxysmal dyspnea), ii) reduced cardiac output resulting in fatigue or dyspnea on exertion, arrhythmias and/or conduction system disease and iii) thromboembolic disease or stroke, mainly from left ventricular mural thrombus. However, some individuals with a DCM pathogenic variant may also be asymptomatic.<sup>15,19</sup> **Left ventricular non-compaction (LVNC)** is characterized by abnormal trabeculations in the left ventricle, most frequently at the apex, and can share the same clinical presentation as DCM, ranging from asymptomatic disease to progressive deterioration of cardiac function, arrhythmias, thromboembolic events, or sudden cardiac death.<sup>19,20</sup> **Noonan syndrome (NS)** is a relatively common multi-system disorder that may include HCM, facial dysmorphism, congenital heart defects, short stature, skeletal malformations, motor delay, learning disabilities, and impaired blood clotting ability.<sup>21</sup> Cardiomyopathy can also be a presenting feature of other inherited disorders, such as Danon disease, Fabry disease, mitochondrial myopathy, or muscular dystrophy<sup>15-19</sup>.

**Inheritance Pattern/Genetics:** Autosomal Dominant, Autosomal Recessive, X-linked, or Mitochondrial

## Test Methods:

Using genomic DNA from the submitted specimen, the complete coding regions and splice site junctions of the genes on this panel are enriched using a proprietary targeted capture system developed by GeneDx for next-generation sequencing with CNV calling (NGS-CNV) (Only exons 1-44 for CACNA1C, only the KCNQ1-binding domains including Ser1570 residue for AKAP9, and excluding exon 6 of the PKP2 gene and the following genomic regions of the TTN gene: chr2:179527692-179527782, 179523898-179523982, 179523731-179523815). The enriched targets are simultaneously sequenced with paired-end reads on an Illumina platform. Bi-directional sequence reads are assembled and aligned to reference sequences based on NCBI RefSeq transcripts and human genome build GRCh37/UCSC hg19. After gene specific filtering, data are analyzed to identify sequence variants and most deletions and duplications involving coding exons. Alternative sequencing or copy number detection methods are used to analyze regions with inadequate sequence or copy number data. Reportable variants include pathogenic variants, likely pathogenic variants and variants of uncertain significance. Likely benign and benign variants, if present, are not routinely reported but are available upon request.

## Test Sensitivity:

The clinical sensitivity of sequencing and deletion/duplication analysis of the genes included in the Combined Cardiac Panel depends in part on the patient's clinical phenotype and family history. In general, the sensitivity is highest for individuals with clearly defined cardiomyopathy or arrhythmia and a family history of disease. The technical sensitivity of sequencing is estimated to be > 99% at detecting single nucleotide events. It will not reliably detect deletions greater than 20 base pairs, insertions or rearrangements greater than 10 base pairs, or low-level mosaicism. The copy number assessment methods used with this test cannot reliably detect copy number variants of less than 500 base pairs or mosaicism and cannot identify balanced chromosome aberrations. Assessment of exon-level copy number events is dependent on the inherent sequence properties of the targeted regions, including shared homology and exon size. For the HRAS gene, FKR gene and the mitochondrial genes, sequencing but not deletion/duplication analysis, is performed. Gene specific exclusions for exon-level deletion/duplication testing for this panel are: CALM1, EMD, KCNE1L, SCN1B and TAZ genes only whole gene deletions or duplications may be detected.

Gene	Protein	Inheritance	Disease Association(s)
<i>ABCC9</i>	ATP-BINDING CASSETTE, SUBFAMILY C, MEMBER 9	AD	DCM, BrS, Cantu syndrome and related disorders
<i>ACTC1</i>	ACTIN, ALPHA, CARDIAC MUSCLE	AD	CHD, DCM, HCM, LVNC
<i>ACTN2</i>	ACTININ, ALPHA-2	AD	DCM, HCM
<i>AKAP9</i>	A-KINASE ANCHOR PROTEIN 9	AD	LQTS, Cardiomyopathy
<i>ALMS1</i>	CENTROSOME AND BASAL BODY ASSOCIATED PROTEIN	AR	Alstrom syndrome, infantile DCM, mitogenic cardiomyopathy
<i>ALPK3</i>	ALPHA-KINASE 3	AR	Pediatric Cardiomyopathy
<i>ANK2</i>	ANKYRIN 2	AD	Arrhythmia, LQTS
<i>ANKRD1</i>	ANKYRIN REPEAT DOMAIN-CONTAINING PROTEIN 1	AD	DCM
<i>BAG3</i>	BCL2-ASSOCIATED ATHANOGENE 3	AD	DCM, myofibrillar myopathy
<i>BRAF</i>	V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1	AD	Noonan/CFC/Costello syndromes
<i>CACNA1C</i>	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, L TYPE, ALPHA-1C SUBUNIT	AD	BrS, Timothy syndrome, LQTS
<i>CACNA2D1</i>	CALCIUM CHANNEL, VOLTSGE-DEPENDENT ALPHA-2/DELTA SUBUNIT 1	AD	BrS
<i>CACNB2</i>	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, BETA-2 SUBUNIT	AD	BrS
<i>CALM1</i>	CALMODULIN 1	AD	LQTS, CPVT
<i>CALM2</i>	CALMODULIN 2	AD	LQTS, CPVT
<i>CALM3</i>	CALMODULIN 3	AD	LQTS, CPVT
<i>CASQ2</i>	CALSEQUESTRIN 2	AR	CPVT
<i>CAV3</i>	CAVEOLIN 3	AD	HCM, LQTS, LGMD, Tateyama-type distal myopathy, SIDS, rippling muscle disease
<i>CHRM2</i>	M2-MUSCARINIC ACETYLCHOLINE RECEPTOR	AD	DCM
<i>CRYAB</i>	CRYSTALLIN, ALPHA-B	AD, AR	DCM, myofibrillar myopathy
<i>CSRP3</i>	CYSTEINE- AND GLYCINE-RICH PROTEIN 3	AD	HCM, DCM
<i>CTNNA3</i>	CATENIN ALPHA 3	AD, AR	ARVC, Autism
<i>DES</i>	DESMIN	AD, AR	DCM, ARVC, myopathy, AV block, LGMD
<i>DMD</i>	DYSTROPHIN	XL	DMD, BMD, DCM
<i>DOLK</i>	DOLICHOL KINASE	AR	DCM, congenital disorder of glycosylation type LM

Gene (cont.)	Protein	Inheritance	Disease Association(s)
<i>DSC2</i>	DESMOCOLLIN	AD, AR	ARVC, ARVC+skin and hair findings, DCM
<i>DSG2</i>	DESMOGLEIN	AD	ARVC, DCM
<i>DSP</i>	DESMOPLAKIN	AD, AR	ARVC, DCM, Carvajal syndrome and related disorders
<i>DTNA</i>	DYSTROBREVIN, ALPHA	AD	LVNC, CHD
<i>EMD</i>	EMERIN	XL	EMD
<i>FHL1</i>	FOUR-AND-A-HALF LIM DOMAINS 1	XL	HCM, EMD, Myofibrillar myopathy
<i>FKRP</i>	FUKUTIN RELATED PROTEIN	AR	Muscular dystrophy
<i>FKTN</i>	FUKUTIN	AR	DCM, LGMD, Fukuyama Congenital Muscular Dystrophy
<i>FLNC</i>	FILAMIN C	AD	RCM, HCM, ARVC, DCM, myopathy
<i>GAA</i>	GLUCOSIDASE, ALPHA, ACID	AR	Pompe Disease (Glycogen storage disease II)
<i>GATA4</i>	GATA-BINDING PROTEIN 4	AD	AF, CHD, cardiomyopathy, SUDS
<i>GATA5</i>	GATA-BINDING PROTEIN 5	AD	AF, congenital heart defects, cardiomyopathy
<i>GATA6</i>	GATA-BINDING PROTEIN 5	AD	AF, CHD, pancreatic agenesis, diaphragmatic hernia, diabetes, cardiomyopathy
<i>GATAD1</i>	GATA ZINC FINGER DOMAIN-CONTAINING PROTEIN 1	AR	DCM
<i>GJA5</i>	GAP JUNCTION PROTEIN, ALPHA-5	AD	AF, HB, SADS, SIDS, congenital heart defects
<i>GLA</i>	GALACTOSIDASE, ALPHA	XL	Fabry disease
<i>GNB5</i>	GUANINE NUCLEOTIDE-BINDING PROTEIN, BETA-5	AR	Intellectual developmental disorder with cardiac arrhythmia
<i>GDPII</i>	GLYCEROL-3-PHOSPHATE DEHYDROGENASE 1-LIKE	AD	BrS
<i>HCN4</i>	HYPERPOLARIZATION-ACTIVATED CYCLIC NUCLEOTIDE-GATED POTASSIUM CHANNEL 4	AD	BrS, SSS, tachycardia, LVNC, AF, AV block, bradycardia
<i>HFE</i>	HUMAN HEMOCHROMATOSIS PROTEIN (HFE)	AR	Hereditary Hemochromatosis, cardiomyopathy
<i>HRAS</i>	V-HA-RAS HARVEY RAT SARCOMA VIRAL ONCOGENE HOMOLOG	AD	Costello syndrome
<i>ILK</i>	INTEGRIN-LINKED KINASE	AD	DCM
<i>JPH2</i>	JUNCTOPHILIN 2	AD	HCM
<i>JUP</i>	JUNCTION PLAKOGLOBIN	AD, AR	ARVC, Naxos Disease and related disorders
<i>KCNA5</i>	POTASSIUM CHANNEL, VOLTAGE-GATED, SHAKER-RELATED SUBFAMILY, MEMBER 5	AD	AF
<i>KCND3</i>	POTASSIUM CHANNEL, VOLTAGE-GATED, SHAL-RELATED SUBFAMILY, MEMBER 3	AD	BrS, SIDS, Spinocerebellar ataxia
<i>KCNE1</i>	POTASSIUM CHANNEL, VOLTAGE-GATED, ISK-RELATED SUBFAMILY, MEMBER 1	AD, AR	LQTS, JLNS
<i>KCNE2</i>	POTASSIUM CHANNEL, VOLTAGE-GATED, ISK-RELATED SUBFAMILY, MEMBER 2	AD	LQTS
<i>KCNE3</i>	POTASSIUM CHANNEL, VOLTAGE-GATED, ISK-RELATED SUBFAMILY, MEMBER 3	AD	BrS
<i>KCNE1L (KCNE5)</i>	POTASSIUM CHANNEL, VOLTAGE-GATED, ISK-RELATED FAMILY, MEMBER 1-LIKE	XL	BrS, AF, VF
<i>KCNH2 (HERG)</i>	POTASSIUM CHANNEL, VOLTAGE-GATED, SUBFAMILY H, MEMBER 2	AD	LQTS, SQTS
<i>KCNJ2</i>	POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 2	AD	Andersen-Tawil syndrome, SQTS
<i>KCNJ5</i>	POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 5	AD	LQTS
<i>KCNJ8</i>	POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 8	AD	ERS, SIDS
<i>KCNQ1</i>	POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 1	AD, AR	JLNS, LQTS, SQTS
<i>KRAS</i>	V-KI-RAS2 KIRSTEN RAT SARCOMA VIRAL ONCOGENE HOMOLOG	AD	Noonan/CFC/Costello
<i>LAMA4</i>	LAMININ, ALPHA-4	AD	DCM

Gene (cont.)	Protein	Inheritance	Disease Association(s)
<i>LAMP2</i>	LYSOSOME-ASSOCIATED MEMBRANE PROTEIN 2	XL	Danon disease
<i>LDB3</i>	LIM DOMAIN-BINDING 3	AD	DCM, LVNC, ARVC, LDB3-related myopathy
<i>LMNA</i>	LAMIN A/C	AD, AR	DCM, LVNC, ARVC, LDB3-related myopathy
<i>LRRC10</i>	LEUCINE-RICH REPEAT-CONTAINING PROTEIN 10	AD, AR	DCM
<i>MAP2K1</i>	MITOGEN-ACTIVATED PROTEIN KINASE KINASE 1	AD	Noonan/CFC/Costello
<i>MAP2K2</i>	MITOGEN-ACTIVATED PROTEIN KINASE KINASE 2	AD	Noonan/CFC/Costello
<i>MIB1</i>	MINDBOMB E3 UBIQUITIN PROTEIN LIGASE 1	AD	LVNC
<i>MTND1</i>	mtDNA ENCODED COMPLEX I, SUBUNIT ND1	MITO	Cardiomyopathy, myopathy
<i>MTND5</i>	mtDNA ENCODED COMPLEX I, SUBUNIT ND5	MITO	Cardiomyopathy, myopathy
<i>MTND6</i>	mtDNA ENCODED COMPLEX I, SUBUNIT ND6	MITO	Cardiomyopathy, myopathy
<i>MTTD</i>	MITOCHONDRIAL tRNA FOR ASPARTIC ACID	MITO	Cardiomyopathy, myopathy
<i>MTTG</i>	MITOCHONDRIAL tRNA FOR GLYCINE	MITO	Cardiomyopathy, myopathy
<i>MTTH</i>	MITOCHONDRIAL tRNA FOR HISTIDINE	MITO	Cardiomyopathy, myopathy
<i>MTTI</i>	MITOCHONDRIAL tRNA FOR ISOLEUCINE	MITO	Cardiomyopathy, myopathy
<i>MTTK</i>	MITOCHONDRIAL tRNA FOR LYSINE	MITO	Cardiomyopathy, myopathy
<i>MTTL1</i>	MITOCHONDRIAL tRNA FOR LEUCINE 1	MITO	Cardiomyopathy, myopathy
<i>MTTL2</i>	MITOCHONDRIAL tRNA FOR LEUCINE 2	MITO	Cardiomyopathy, myopathy
<i>MTTM</i>	MITOCHONDRIAL tRNA FOR METHIONINE	MITO	Cardiomyopathy, myopathy
<i>MTTQ</i>	MITOCHONDRIAL tRNA FOR GLUTAMINE	MITO	Cardiomyopathy, myopathy
<i>MTTS1</i>	MITOCHONDRIAL tRNA FOR SERINE 1	MITO	Cardiomyopathy, myopathy
<i>MTTS2</i>	MITOCHONDRIAL tRNA FOR SERINE 2	MITO	Cardiomyopathy, myopathy
<i>MURC</i>	MUSCLE-RELATED COILED-COIL PROTEIN	AD	DCM
<i>MYBPC3</i>	MYOSIN-BINDING PROTEIN C, CARDIAC	AD	HCM, DCM
<i>MYH6</i>	MYOSIN, HEAVY CHAIN 6, CARDIAC MUSCLE, ALPHA	AD	CHD, DCM, HCM, SSS
<i>MYH7</i>	MYOSIN, HEAVY CHAIN 7, CARDIAC MUSCLE, BETA	AD	DCM, HCM, myopathy
<i>MYL2</i>	MYOSIN, LIGHT CHAIN 2, REGULATORY, CARDIAC, SLOW	AD, AR	HCM, muscle fiber disease
<i>MYL3</i>	MYOSIN, LIGHT CHAIN 3, ALKALI, VENTRICULAR, SKELETAL, SLOW	AD, AR	HCM
<i>MYL4</i>	MYOSIN, LIGHT CHAIN 4, ALKALI, ATRIAL, EMBRYONIC	AD, AR	AF
<i>MYLK2</i>	MYOSIN LIGHT CHAIN KINASE 2	AD	HCM
<i>MYOZ2</i>	MYOZENIN 2	AD	HCM
<i>MYPN</i>	MYOPALLADIN	AD	DCM, RCM, HCM
<i>NEBL</i>	NEBULETTE	AD	DCM, endocardial fibroelastosis
<i>NEXN</i>	NEXILIN	AD	DCM, HCM
<i>NKX2-5</i>	NK2 HOMEODOMAIN 5	AD	CHD, CCD, cardiomyopathy
<i>NRAS</i>	NEUROBLASTOMA RAS VIRAL ONCOGENE HOMOLOG	AD	Noonan/CFC/Costello syndromes
<i>PDLIM3</i>	PDZ AND LIM DOMAIN PROTEIN 3	AD	HCM, DCM
<i>PKP2</i>	PLAKOPHILIN 2	AD	ARVC, BrS
<i>PLN</i>	PHOSPHOLAMBAN	AD	DCM, HCM, ARVC
<i>PPA2</i>	PYROPHOSPHATASE, INORGANIC, 2	AR	Cardiomyopathy, infantile cardiac arrest
<i>PRDM16</i>	PR DOMAIN CONTAINING 16	AD	DCM, LVNC
<i>PRKAG2</i>	PROTEIN KINASE, AMP-ACTIVATED, NONCATALYTIC, GAMMA2	AD	HCM, Wolff-Parkinson-White syndrome
<i>PTPN11</i>	PROTEIN-TYROSINE PHOSPHATASE, NONRECEPTOR-TYPE 11	AD	Noonan/CFC/Costello syndromes
<i>RAF1</i>	V-RAF-1 MURINE LEUKEMIA VIRAL ONCOGENE HOMOLOG 1	AD	Noonan/CFC/Costello syndromes
<i>RBM20</i>	RNA-BINDING MOTIF PROTEIN 20	AD	DCM
<i>RIT1</i>	RAS-LIKE WITHOUT CAAX 1	AD	Noonan syndrome
<i>RANGRF</i>	RAN GUANINE NUCLEOTIDE RELEASE FACTOR	AD	BrS
<i>RYR2</i>	RYANODINE RECEPTOR 2	AD	ARVC, CPVT, DCM

Gene (cont.)	Protein	Inheritance	Disease Association(s)
<i>SCN10A</i>	SODIUM CHANNEL, VOLTAGE-GATED, TYPE X, ALPHA SUBUNIT	AD	BrS
<i>SCN1B</i>	SODIUM CHANNEL, VOLTAGE-GATED, TYPE I, BETA SUBUNIT	AD, AR	BrS, Cardiac conduction disease
<i>SCN2B</i>	SODIUM CHANNEL, VOLTAGE-GATED, TYPE II, BETA SUBUNIT	AD	BrS, AF
<i>SCN3B</i>	SODIUM CHANNEL, VOLTAGE-GATED, TYPE III, BETA SUBUNIT	AD	BrS, AF, VF, SIDS
<i>SCN4B</i>	SODIUM CHANNEL, VOLTAGE-GATED, TYPE IV, BETA SUBUNIT	AD	LQTS
<i>SCN5A</i>	SODIUM CHANNEL, VOLTAGE-GATED, TYPE V, ALPHA SUBUNIT	AD	BrS, DCM, Heart block, LQTS, SSS, SIDS, ARVC, ARVC-like disease
<i>SGCD</i>	SARCOGLYCAN, DELTA	AD, AR	DCM, LGMD
<i>SHOC2</i>	SOC-2 HOMOLOG	AD	Noonan-like syndrome with loose Anagen Hair 1
<i>SNTA1</i>	ALPHA SYNTROPHIN	AD	LQTS
<i>SOS1</i>	SON OF SEVENLESS, DROSOPHILA, HOMOLOG 1	AD	Noonan/CFC/Costello syndromes
<i>TAZ</i>	TFAZZIN	XL	DCM, LVNC, Barth syndrome
<i>TBX20</i>	T-BOX 20	AD	CHD, DCM, LVNC
<i>TCAP</i>	TITIN-CAP (TELETHONIN)	AD, AR	HCM, DCM, LGMD
<i>TECL</i>	TRANS-2,3-ENOYL-CoA REDUCTASE-LIKE PROTEIN	AR	CPVT3
<i>TGFB3</i>	TRANSFORMING GROWTH FACTOR BETA 3	AD	ARVC, Loeys-Dietz syndrome, TAAD
<i>TMEM43</i>	TRANSMEMBRANE PROTEIN 43	AD	ARVC, EMD
<i>TMPO</i>	THYMOPOIETIN	AD	DCM
<i>TNNC1</i>	TROPONIN C, SLOW	AD	DCM, HCM
<i>TNNI3</i>	TROPONIN I, CARDIAC	AD	DCM, HCM, RCM
<i>TNNT2</i>	TROPONIN T2, CARDIAC	AD	DCM, HCM, RCM, LVNC
<i>TOR1AIP1</i>	TORSIN-1A-INTERACTING PROTEIN 1	AR	LGMD, Contractures, DCM
<i>TPM1</i>	TROPOMYOSIN 1	AD	DCM, HCM
<i>TRDN</i>	TRIADIN	AR	CPVT, LQTS
<i>TRPM4</i>	TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY M, MEMBER 4	AD	HB, BrS
<i>TTN</i>	TITIN	AD, AR	DCM, ARVC, TTN-related myopathies and muscular dystrophies
<i>TTR</i>	TRANSTHYRETIN	AD	TTR-related amyloidosis
<i>TXNRD2</i>	THIOREDOXIN REDUCTASE 2	AD, AR	DCM
<i>VCL</i>	VINCULIN	AD	HCM, DCM, LVNC

Abbreviations: AD- Autosomal dominant; AF- Atrial Fibrillation; AR- Autosomal recessive; ARVC – Arrhythmogenic Right Ventricular Cardiomyopathy; BMD – Becker Muscular Dystrophy; BrS – Brugada Syndrome; CHD – Congenital Heart Defects; CPVT – Catecholaminergic Polymorphic Ventricular Tachycardia; DCM – Dilated Cardiomyopathy; DMD- Duchenne Muscular Dystrophy; EMD – Emery Dreifuss Muscular Dystrophy; ERS-Early repolarization syndrome; HB- Heart Block; HCM – Hypertrophic Cardiomyopathy; JLNS – Jervell and Lange-Nielsen Syndrome; LGMD – Limb Girdle Muscular Dystrophy; LQTS – Long QT Syndrome; LVNC – Left Ventricular Non-Compaction; RCM – Restrictive Cardiomyopathy; SIDS – Sudden Infant Death Syndrome; SSS – Sick Sinus Syndrome; VF-Ventricular fibrillation; XL- X-linked

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