ONLINE APPENDIX

Contemporary and Future Approaches to Precision Medicine in Inherited Cardiomyopathies

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ONLINE TABLE 1. Genes with cardiomyopathy-associated rare variants.

Gene	Protein	Rare variants*			Common variants†	Notes
		HCM	G-DCM	ARVC/ ACM		
ABCC9	ATP binding cassette C9		+			Potassium channel subunit. Single report in AD DCM. Also associated with AR Cantu syndrome.
ACAD9	Acyl CoA dehydrogenase 9	+	+			Mitochondrial function protein. DCM/HCM with AR mitochondrial syndrome.
ACTA1	Actin alpha 1		+			Skeletal sarcomeric protein. Rare cardiac involvement with AD myopathy.
ACTC1	Actin alpha cardiac muscle	+++	++			Sarcomeric protein. Primary association is AD HCM. Also moderate association with AD DCM/LVNC and CHD.
ACTN2	Actin alpha 2	++	++		Y	Skeletal and cardiac sarcomeric protein. Mixed AD phenotypes within families reported including HCM, DCM and arrhythmias. Also associated with LVNC.
AKAP9	A kinase anchor protein 9		+			Cell signaling protein. Single report of AD DCM in a cohort study.
ALG6	Alpha 1,3 glucosyltransferase		+			Glycosylation protein. DCM associated with AR congenital glycosylation disorder.
ALMS1	Alstrom syndrome protein 1		+			Centrosome protein. DCM with AR Alstrom syndrome
ALPK3	Alpha kinase 3	+++	++		Y	Nuclear kinase protein. Biallelic variants cause pediatric DCM transitioning to HCM in adulthood with dysmorphic features. Heterozygous rare variants may cause mild cardiomyopathy.
ANKRD1	Ankyrin Repeat Domain 1	+	+			Transcription factor. Rare reports in AD HCM and DCM. Also associated with CHD.

ANO5	Anoctamin 5		+			Chloride channel protein. Rare cardiac involvement with AR muscular dystrophy.
BAG3	BAG cochaperone 3	+	+++		Y	Chaperone protein. Strong association with severe AD DCM. Also associated with HCM/RCM in the setting of AD myofibrillar myopathy.
BRAF	B-Raf Proto-Oncogene, Serine/Threonine Kinase	+				Oncogene protein. Associated with HCM in AD Noonan syndrome
CALR3	Calreticulin 3	+				Chaperone protein. Two VUS's reported in one AD HCM cohort study.
CASZ1	Castor Zinc Finger Protein 1		+			Transcription factor. Rare reports of AD HCM. One report in CHD.
CAV3	Caveolin-3	+	+			Scaffolding protein. Associated with DCM with AD muscular dystrophy. Isolated AD/AR DCM and AD HCM rarely reported.
CAVIN4	Caveolae associated protein 4		+			Z-line protein. Reported in several cases in one AD DCM cohort study.
CDC25B	Cell division cycle 25B		+			Cell division phosphatase protein. AR DCM with syndromic features in one report.
CDH2	Cadherin 2			+		Cell adhesion protein. Two reports (3 families) with AD ARVC.
CHKB	Choline-kinase beta		+			Phospholipid biosynthesis protein. DCM reported in association with AR muscular dystrophy and syndromic features.
CHRM2	Cholinergic receptor, muscarinic 2		+			Muscarinic receptor protein. AD DCM reported in one large Han Chinese family.
CMYA3	Cardiomyopathy-associated protein 3		+			Intercalated disc maturation protein. Three probands reported with DCM; two are heterozygote VUS's, one compound heterozygote. No segregation.
CNBP	Cellular nucleic acid binding protein		+			Nucleic acid binding protein. DCM reported in association with AD myotonic dystrophy 2

CRYAB	alpha-B crystallin		+			Chaperone protein. DCM/HCM/RCM reported in association with myofibrillar myopathy. Rare reports of isolated DCM and RCM.
CSRP3	Cysteine and glycine rich protein 3	++	+		Y	Cytoskeletal regulatory protein. Moderate association with AD HCM. Associated with DCM in two cohort studies.
CTNNA3	Catenin alpha 3			+		Cell-cell adhesion protein. Rare association with AD ARVC.
CTF1	Cardiotrophin 1		+			Cytokine protein. Rare association with AD DCM.
DES	Desmin	+	+++	++		Cytoskeletal protein. Strong association with AD DCM and ACM overlap phenotypes that can co-occur with CD and myopathy. Also associated with AD RCM + CD. One report in HCM.
DLG1	Discs large homolog 1	+				Scaffolding protein. Single report of AD HCM in an expanded cohort.
DMD	Dystrophin		+++			Cytoskeletal protein. Associated with DCM in Duchenne's and Becker muscular dystrophy; female carriers can present with Isolated DCM. Can present in males as X-linked DCM without skeletal myopathy.
DMPK	Dystrophia myotonica protein kinase		+			Nuclear matrix protein. Associated with DCM (+/- CD, AA, VA) in AD myotonic dystrophy 1.
DNAJC19	DNAJ/HSP40 homolog C19		+			Mitochondrial membrane protein. Associated with DCM in AR mitochondrial syndrome
DOLK	Dolichol kinase		+			Glycosylation protein. Associated with DCM in AR congenital glycosylation disorder.
DPM3	Dolichol P Mannose synthase subunit 3		+			Endoplasmic reticulum protein. Associated with DCM in AD muscular dystrophy.

DSC2	Desmocollin		+	+++		Desmosomal protein. Strong association with AD ARVC. Isolated AD DCM cases also described.
DSG2	Desmoglein		+	+++		Desmosomal protein. Strong association with AD ARVC. Isolated AD DCM cases also described.
DSP	Desmoplakin		+++	+++		Desmosomal protein. Strong association with ACM with left and/or right ventricular involvement (ARVC or ALVC). ALVC can present like DCM. Acute episodes can mimic myocarditis. Also associated with LVNC.
DTNA	Dystrobrevin alpha		+			Dystrophin complex protein. Two reports of AD LVNC with systolic dysfunction.
EEF1A2	Eukaryotic translation elongation factor 1A2		+			tRNA transport protein. One family report of AR DCM with syndromic features.
EMD	Emerin		+			Nuclear membrane protein. Associated with DCM in XL Emery-Dreifuss muscular dystrophy. Isolated AD DCM cases rarely reported.
EPG5	Ectopic P granules protein		+			Autophagy protein. Associated with DCM in AR Vici syndrome
EYA4	Eyes absent 4		+	+		Transcription factor. Associated with syndrome of AD sensorineural hearing loss +/- DCM.
FBXO32	F-box only protein 32		++			Autophagy protein. Moderate association with severe AR DCM.
FHL2	Four and a half LIM domain protein 2	+	+			Extracellular membrane assembly protein. Several heterozygote variants isolated in one DCM/HCM cohort.
FHOD3	Formin homology 2 domain containing 3	+++	+		Y	Sarcomeric function protein. Emerging strong association with AD HCM. One report in AD DCM.

FKRP	Fukutin related protein		+			Post translation modification protein. Associated with DCM in AR muscular dystrophy. Rare case reports of DCM preceding muscular dystrophy in childhood.
FKTN	Fukutin related protein		+			Glycosylation protein. Associated with DCM in AR Fukuyama muscular dystrophy. Isolated AR DCM also described.
FLNC	Filamin C	++	+++	++	Y	Cytoskeletal protein. Strong association with AD arrhythmic DCM. Moderate association with AD ACM and HCM. Also associated with AD myofibrillar myopathy.
FOXD4	Forkhead box D4		+			Transcription factor. Associated with AD DCM in one family report (2 individuals).
GAA	Glucosidase Alpha Acid	+				Galactosidase protein. Associated with HCM in AR congenital glycosylation disorder.
GLA	Galactosidase alpha	+++				Galactosidase protein. Associated with HCM phenocopy in X-linked Fabry's disease.
FXN	Frataxin	+	+			Mitochondrial protein. Associated with HCM and/or DCM in the setting of Friedrich's ataxia.
GATA4	Gata binding factor 4	+	+			Transcription factor. Two family reports in AD DCM. One variant in an HCM cohort. Also associated with CHD, AF and LVNC.
GATA5	Gata binding factor 5		+			Transcription factor. One family report of AD DCM. Also associated with CHD and AF.
GATA6	Gata binding factor 6	+	+			Transcription factor. Two family reports of AD DCM. Also associated with CHD and AF.
GATAD1	Gata zinc finger domain containg 1		+			Transcription factor. One family report of AR DCM.

HAND1	Hand and neural crest derivatives expressed 1		+			Transcription factor. One heterozygote missense variant identified in a DCM cohort.
HAND2	Hand and neural crest derivatives expressed 2		+		Y	Transcription factor. One family report of AD DCM.
HCN4	Hyperpolarization Activated Cyclic Nucleotide Gated Potassium Channel 4		+			Potassium channel protein. Associated with a cardiac syndrome of LVNC, CD and AF. Some individuals have reduced LVEF.
HFE	Homeostatic iron regulator		+			Iron regulation protein. Associated with DCM and CD in AR haemochromatosis.
HRAS	Hras proto-oncogene GTPase	+				Oncogene protein. Associated with HCM in AD Costello syndrome.
ILK	Integrin linked kinase		+	+		Transmembrane signal transduction protein. Rare cases of AD ARVC. One variant in a DCM cohort.
ISL1	ISL Lim homeobox 1		+			Transcription factor. One family report of AD DCM.
ITPA	Inosine triphosphatase		+			Phosphatase protein. Associated with infantile DCM in Martsolf syndrome.
JPH2	Junctophilin 2	++	+			Cytoskeletal protein. Moderate association with AD HCM. Rare cases of AR/AD DCM.
JUP	Junction plakoglobin			+++		Junctional plaque protein. Strong association with AD ARVC. Also associated with AR Naxos disease.
KCNJ12	Inwardly rectifying potassium channel J12		+			Potassium channel protein. One family report of AD DCM, possibly arrhythmia mediated.
KCNJ2	Inwardly rectifying potassium channel J2		+			Potassium channel protein. Three family reports of DCM in AD LQT (Anderson Tawil syndrome), possibly arrhythmia mediated. Also associated with AF and SQT.
KCNQ1	Voltage gated potassium channel KQT like 1		+			Potassium channel protein. Two reports of AD DCM with VPBs, possibly arrhythmia

						mediated. Also associated with LQT, AF and SQT.
KLF5	Kruppel like factor 5		+			Transcription factor. One family report of AD DCM.
KLF10	Kruppel like factor 10	+				Transcription factor. Several variants identified in an HCM cohort.
KRAS	Kras proto-oncogene GTPase	+				Oncogene protein. Associated with HCM in AD Noonan syndrome.
LAMA2	Laminin alpha 2		+			Extracellular matrix protein. Associated with DCM with AR muscular dystrophy.
LAMA4	Laminin alpha 4		+			Extracellular matrix protein. Variants identified in 3 DCM cohorts. No family data.
LAMP2	Lysosome associated membrane protein 2	+++	++			Membrane glycoprotein. Associated with HCM phenocopy with X-linked Danon disease. Female carriers can present with isolated DCM or HCM.
LDB3	Lim domain binding 3	+	++			Sarcomeric stabilization protein. Associated with AD DCM in three families. Variants isolated in several DCM cohorts and one HCM cohort. Also associated with LVNC. Also associated with late onset AD myofibrillar myopathy.
LIMS2	Lim zinc finger domain containing 2		+			Extracellular matrix protein. Associated with DCM with AR muscular dystrophy.
LMNA	Lamin A/C		+++	+		Nuclear lamina protein. Strong association with DCM with CD and AA/VA. Also associated with LVNC. Also associated with AD Emery-Dreifuss muscular dystrophy.
LMOD2	Leiomodin 2		+			Actin regulation protein. One report of homozygous nonsense variants in an infant with DCM.

LRRC10	Leucine rich repeat containing protein 10		+			Actin binding protein. Associated with AD DCM in two small families from one cohort. Homozygous variants reported in one infant with DCM.
MAP2K1	Mitogen activated protein kinase kinase 1	+				Protein kinase. Associated with HCM in AD Noonan syndrome.
MAP2K2	Mitogen activated protein kinase kinase 2	+				Protein kinase. Associated with HCM in AD Noonan syndrome.
MIB1	Mindbomb E3 Ubiquitin Protein Ligase 1		+			Ubiquitin protein ligase protein. Primary association is with AD LVNC, some with impaired ventricular contraction.
MTATP6	Mitochondrial ATP synthetase 6	+	+			Mitochondrial protein. Associated with HCM or DCM with mitochondrial syndrome.
MT-CO2	Mitochondrial cytochrome C oxidase 1		+			Mitochondrial protein. Associated with mitochondrial syndrome and isolated DCM cases also described.
MT-CO3	Mitochondrial cytochrome C oxidase 2		+			Mitochondrial protein. Associated with mitochondrial syndrome and isolated DCM cases also described.
MT-CYB	Mitochondrial cytochrome B	+	+			Mitochondrial protein. Associated with mitochondrial syndrome and isolated DCM and HCM cases also described.
MT-ND1	Mitochondrial NADH dehydrogenase 1	+	+			Mitochondrial protein. Associated with DCM in mitochondrial syndrome. Isolated HCM cases also described.
MT-ND2	Mitochondrial NADH dehydrogenase 2		+			Mitochondrial protein. Associated with mitochondrial syndrome and isolated DCM cases also reported.
MT-ND5	Mitochondrial NADH dehydrogenase 5		+			Mitochondrial protein. Associated with HCM in mitochondrial syndrome. Isolated DCM cases also described.

MT-RNR1	Mitochondrial ribosomal RNA small subunit 125		+			Mitochondrial protein. Associated with mitochondrial syndrome and isolated DCM and RCM cases also described.
MT-RNR2	Mitochondrial ribosomal RNA large subunit 16s	+	+			Mitochondrial protein. Associated with DCM or HCM in mitochondrial syndrome.
MT-TE	Mitochondrial tRNA Glutamic acid		+			Mitochondrial protein. One report of neonatal DCM with myopathy.
MT-TI	Mitochondrial tRNA isoleucine	+	+			Mitochondrial protein. Associated with DCM in mitochondrial syndrome. Isolated HCM cases also described.
MT-TK	Mitochondrial tRNA lysine	+	+			Mitochondrial protein. Associated with HCM or DCM in mitochondrial syndrome.
MT-TL1	Mitochondrial tRNA leucine	+	+			Mitochondrial protein. Associated with HCM or DCM in mitochondrial syndrome. Isolated HCM cases also described.
MT-TP	Mitochondrial tRNA proline		+			Mitochondrial protein. Associated with mitochondrial syndrome and isolated DCM cases also described.
MTTS1	Mitochondrial tRNA Serine 1		+			Mitochondrial protein. Associated with mitochondrial syndrome and isolated DCM cases also described.
MYBPC3	Myosin binding protein C	+++	+	+		Sarcomeric protein. Strong association with AD HCM. Associated with AD DCM in numerous cohort studies but no family studies. Minor association with ACM and LVNC.
MYBPHL	Myosin binding protein H-like		+			Sarcomeric protein. One family report of nonsense variant causing AD DCM with CD and AA.
MYH6	Myosin heavy chain alpha	+	+		Y	Sarcomeric protein. Primary association is with CHD (atrial septal defect). Limited association with cardiomyopathy.

MYH7	Myosin heavy chain beta	+++	++	+		Sarcomeric protein. Strong association is with AD HCM and DCM. Also associated with LVNC and AD Laing distal myopathy.
MYL2	Myosin light chain 2	+++	+			Sarcomeric protein. Strong association with AD HCM. One family report for AD DCM.
MYL3	Myosin light chain 3	+++	+	+		Sarcomeric protein. Strong association with AD HCM. Single variants identified in one DCM cohort and one ARVC cohort.
MYLK2	Myosin light chain kinase 2	+				Sarcomeric protein. Two VUS's in one HCM patient reported.
MYOM1	Myomesin 1	+	+			Sarcomeric protein. One small family report in AD HCM. Single variant identified in a DCM cohort.
MYOZ2	Myozenin 2	+				Calcineurin signaling protein. One small family report in AD HCM. Several variants identified in cohorts are not rare.
MYPN	Myopalladin	+	+			Sarcomeric protein. Associated with AD DCM in two small families and several cohorts. Associated with AD HCM in several cohorts. Also associated with AR nemaline myopathy which can present with DCM.
NEBL	Nebulette	+	+			Sarcomeric protein. Associated with AD DCM in two cohort studies. Associated with AD HCM and LVNC in one cohort study.
NEXN	Nexilin	+	++			Actin binding protein. Associated with AD DCM in several cohort studies and rare variants enriched in a large DCM case control study. No family data. Associated with AD HCM in two isolated cases.
NKX2.5	NK2 homeobox 5		++		Y	Transcription factor. Associated with AD DCM with AVB and CHD (atrial septal

						defect) in four families and two sporadic cases.
NONO	Non-POU Domain Containing Octamer Binding		+			RNA binding protein. Associated with X-linked DCM/LVNC in a syndrome with intellectual impairment +/- CHD.
NRAS	Nras proto-oncogene GTPase	+				Oncogene protein. Associated with HCM in AD Noonan syndrome
OBSCN	Obscurin	+	+			Sarcomeric assembly protein. Heterozygous rare variants isolated in in one DCM cohort and one HCM cohort.
OGT	O-linked N-acetylglucosamine transferase		+			Glycosylation protein. One report of AR DCM with sensory neuropathy.
PDLIM3	PDZ and LIM domain 3	+	+			Cytoskeletal protein. Associated with AD HCM in two cohort and AD DCM in one cohort.
PGM1	Phosphoglucomutase1		+			Glycosylation protein. Associated with DCM with AR congenital glycosylation disorder.
PKP2	Plakophilin		+	+++		Desmosomal protein. Strong association with ARVC. Associated with AD DCM in several cohorts, no family studies.
PLEKHM2	Pleckstrin homology domain containing protein M2		+			Microtubule associated lysosome positioning protein. Associated with AR DCM with VA in one consanguineous Bedouin family. Also associated with LVNC.
PLN	Phospholamban	+	+++	+++	Y	Sarcoplasmic reticulum Ca^{2+}/ATPase protein. Strongly associated with AD DCM with VAs, ACM and overlap phenotypes.
PNPLA2	Patatin like phospholipase domain containing protein 2		+			Lipid metabolism enzyme. Associated with DCM with AR myopathy.
POMT1	Protein O mannosyl transferase 1		+			Glycosylation protein. Associated with DCM in AR muscular dystrophy.

POMT2	Protein O mannosyl transferase 2		+			Glycosylation protein. Associated with DCM in AR muscular dystrophy.
PPP131RL	Protein phosphatase 1, regulatory subunit 13 like		++			Desmosomal protein. Associated with severe pediatric AR DCM in six families.
PRDM16	PR domain containing protein 16		+			Transcription factor. Associated with AD DCM and LVNC in one cohort. De-novo variants in two pediatric DCM cases. May be the cause of DCM in 1p36 deletion syndrome.
PRKAG2	Protein kinase AMP-activated non-catalytic subunit gamma 2	+++				Protein kinase. Associated with HCM phenocopy with VPB, AA and CD in AD glycogen storage disease.
PSEN1	Presenelin 1		+	+		Endoprotease complex protein. Two promoter variants in one DCM cohort. One family report in AD DCM but variant not rare. One variant in an ARVC cohort.
PSEN2	Presenelin 2	+				Endoprotease complex protein. One family report in AD DCM but variant not rare. Variants isolated on one HCM cohort.
PTPN11	Protein Tyrosine Phosphatase Non-Receptor Type 11	+				Cell signaling protein. Associated with HCM in AD Noonan syndrome
RAF-1	V-RAF-1 murine leukemia viral oncogene homolog 1	+				Cell-cell signaling protein. Associated with HCM in AD Noonan syndrome.
RBM20	RNA binding protein		+++		Y	RNA binding protein. Strong association with arrhythmogenic DCM. Also associated with LVNC.
RIT1	Ras Like Without CAAX 1	+				Cell signaling protein. Associated with HCM in AD Noonan syndrome.
RMND1	Required for meiotic nuclear division 1		+			Mitochondrial protein. Associated with DCM in mitochondrial syndrome.

RRAGC	Ras-related GTP binding protein C		+			Cell-cell signaling protein. One report of de-novo heterozygous variant in pediatric DCM and dysmorphism.
RYR2	Ryanodine receptor 2	+	+			Sarcoplasmic reticulum calcium channel protein. Primary association is with CPVT. Associated with AD DCM in two families; possibly an arrhythmic mechanism. Associated with AD HCM in two cohorts.
SCN5A	Voltage gated sodium channel 5A		+++	+		Voltage gated sodium channel protein. Strong association with DCM with CD or arrhythmias. Variants isolated in several ARVC cohorts but true ARVC diagnosis questionable in some of these patients. Also strongly association with Brugada syndrome and LQT.
SDHA	Succinate dehydrogenase complex flavoprotein subunit A		+			Mitochondrial protein. Associated with DCM in both AD and AR mitochondrial syndrome.
SGCA	Alpha sarcoglycan		+			Cytoskeletal extracellular matrix linker protein. Associated with DCM with AR muscular dystrophy.
SGCB	Beta sarcoglycan		+			Cytoskeletal extracellular matrix linker protein. Associated with DCM with AR muscular dystrophy.
SGCD	Delta sarcoglycan		+			Cytoskeletal extracellular matrix linker protein. Associated with AD DCM in several cohorts but most variants reported are not rare.
SGCG	Gamma sarcoglyan		+			Cytoskeletal extracellular matrix linker protein. Associated with DCM with AR muscular dystrophy.
SLC22A5	Solute-carrier family 22		+			Carnitine transport protein. Associated with DCM in AR systemic carnitine deficiency.

SMYD1	SET and MYND domain containing 1	+	+			Histone methylation protein. Homozygous variants in one infant with DCM and CHD. De-novo heterozygous variant in one adult with HCM.
SOD2	Superoxide dismutase 2		+			Mitochondrial protein. Homozygous variants in one case of severe neonatal DCM.
SOS1	SOS Ras/Rac Guanine Nucleotide Exchange Factor 1	+				Cell signaling protein. Associated with HCM in AD Noonan syndrome.
SYNE1	Spectrin repeat containing nuclear envelope protein 1		+			Nuclear envelope protein. Associated with DCM with AD muscular dystrophy. Five probands reported with isolated AD DCM, no family data.
SYNM	Synemin		+			Cytoskeletal extracellular matrix linker protein. One family report of AD DCM.
TAF1A	Tata box binding protein associated factor 1A		+			Transcription factor. One family report of two sisters with compound heterozygote mutations and severe pediatric DCM.
TAX1BP3	Tax1 Binding protein 3		+			Intracellular signaling protein. Associated with AR infantile DCM with septo-optic dysplasia in one report.
TAZ	Tafazzin	+	+++			Mitochondrial protein. Associated with DCM/LVNC in X-linked Barth syndrome. Also a strong association with isolated X-linked pediatric onset DCM/LVNC. One variant in an HCM cohort.
TBX20	T box 20		+			Transcription factor. Associated with AD DCM in two family reports, some relatives with CHD. Associated with CHD in two family reports.
TBX5	T box 5		+			Transcription factor. Associated with DCM and CHD in atypical Holt-Oram syndrome. Isolated DCM cases reported.

TCAP	Telethonin	+	+			Sarcomeric protein. Variants identified in several DCM and HCM cohorts. No family studies.
TGFB3	Transforming Growth Factor Beta 3			+		Growth factor protein. Associated with AD ARVC in two families. Also associated with TAAD.
TJP1	Tight junction protein 1			+		Scaffolding protein. Associated with AD ARVC in one family and several cohorts.
TMEM43	Transmembrane protein 70		++	+++		Nuclear membrane protein. Newfoundland founder mutation causing AC. 10-15% of cases meet criteria for DCM.
TMEM70	Transmembrane protein 70	+	+			Mitochondrial protein. Associated with neonatal DCM or HCM with AR mitochondrial syndrome
TNNC1	Troponin C1	++	++			Sarcomeric protein. Moderate association with both AD DCM and HCM.
TNNI3	Troponin I3	+++	++			Sarcomeric protein. Strongly associated with HCM and RCM. Moderate association with AD DCM.
TNNI3K	Troponin I3 interacting kinase		++			Protein kinase. Five published families with a cardiac syndrome of CD, AA, VA and DCM (~50% of carriers have DCM).
TNNT2	Troponin T2	+++	+++			Sarcomeric protein. Strong association with both AD DCM and HCM. Also associated with RCM and LVNC.
TOR1AIP1	Torsin A interacting protein 1		+			Nuclear membrane protein. Associated with DCM in AR muscular dystrophy.
TPM1	Alpha tropomyosin	+++	+++			Sarcomeric protein. Strong association with both AD DCM and HCM. Also associated with LVNC and RCM.
TRIM63	Tripartite Motif Containing 63	+				Proteosomal degradation regulation protein. Three probands reported with AD HCM. One

						homozygote case with HCM and skeletal myopathy.
TRPM4	Transient receptor potential cation channel		+			Ion channel protein. One variant isolated in a DCM cohort. Strongly associated with CD.
TSFM	Ts Translation elongation factor, mitochondrial	+	+			Mitochondrial protein. Associated with DCM or HCM with AR mitochondrial syndrome
TTN	Titin	+	+++	+	Y	Sarcomeric protein. Strongly associated with AD DCM. Associated with HCM in several cohorts and one family. Associated with ARVC in one family. Also associated with AR myopathy. Also associated with LVNC and RCM.
TTR	Transthyretin	+++	+			Transport protein. Associated with HCM phenocopy and RCM with AD amyloidosis. Rare DCM reports in AD amyloidosis.
TUFM	Tu Translation elongation factor, mitochondrial		+			Mitochondrial protein. Associated with DCM in AR mitochondrial syndrome.
TXNRD2	Thioredoxin reductase 2		+			Mitochondrial protein. Associated with isolated AD DCM in four probands. No family segregation data.
UBR1	Ubiquitin protein ligase E3 component N recognin 1		+			Proteolytic pathway protein. Associated with DCM in AR Johanson-Blizzard syndrome.
VCL	Vinculin	+	++		Y	Cytoskeletal protein. Associated with AD DCM in several cohorts and enriched in one large case control study. Associated with AD HCM in one cohort study.
XRCC4	X-Ray Repair Cross Complementing 4		+			DNA repair protein. Associated with DCM in an AR neurodevelopmental syndrome
ZBTB17	Zinc finger and BTB domain containing protein		+			Transcription factor. One small family report of AD DCM.

*Classification of genes according to level of human genetic evidence for roles in disease causation: +++ Gene has been associated with primary presentation of cardiomyopathy in multiple cases in the literature, with at least 5 instances of family segregation or de novo mutations; ++ Primary cardiomyopathy phenotypes reported in more than 10 individual cases, or 3-5 instances of family segregation or de novo cases (or 2-3 instances if recessive inheritance); + Gene of uncertain significance. Primary cardiomyopathy phenotypes reported in up to 10 individual cases, 1-3 instances of family segregation or do novo cases, OR cardiomyopathy is reported primarily in cases with syndromic features or skeletal myopathy.
†Target genes implicated in genome-wide association studies (GWAS) of heart failure, DCM, or echocardiographic/CMR parameters of cardiac structure and function.
AA = atrial arrhythmias; ACM = arrhythmogenic cardiomyopathy; AD = autosomal dominant; AF = atrial fibrillation; ALVC = arrhythmogenic left ventricular cardiomyopathy; AR = autosomal recessive; ARVC = arrhythmogenic right ventricular cardiomyopathy; CD = conduction defects: CHD = congenital heart disease; DCM = dilated cardiomyopathy; G-DCM = genetically-mediated DCM; HCM = hypertrophy cardiomyopathy; LQT = long QT syndrome; LVNC = left ventricular non-compaction; RCM = restrictive cardiomyopathy; SQT = short QT syndrome; TAAD = thoracic aortic aneurysm and dissection; VA = ventricular arrhythmias; VPB = ventricular premature beats; VUS = variant of uncertain significance.

ONLINE TABLE 2. Genes suspected to carry common variants that influence cardiac structural and functional traits

Gene	Rare variants	Protein	dbSNP	Variant location* (risk allele)	Cardiac trait	Ref.†
ABO	N	ABO, alpha 1-3-N-acetylgalactosaminyltransferase and alpha1-4-galactosyltransferase	rs600038	9:133276354T>C (C)	HF	(1)
ACTN2	Y	Alpha actinin 2	rs10925197	1:23667877G>C (C)	LVEF	(2)
AGAP5	N	ArfGAP with GTPase domain, ankyrin repeat and PH domain 5	rs4746140	10:73657491G>C (G)	HF	(1)
AGO2	N	Argonaute RISC catalytic component 2	rs1962104	8:140625230C>T (T)	LVESV, LVESVi	(2)
AKR1A1	N	Aldo-keto reductase family 1 member A1	rs753562515	1:45541360CAA>C (CAA)	LVEDV, LVESV, LVESVi	(2)
ALPK3	Y	Alpha kinase 3	rs3803403	15:84839914C>G (G)	DCM	(3)
ATP5SL	N	Distal membrane arm assembly complex 2 (*DMAC2*)	rs75124807	19:41439217CT>C (CT)	LVEDV	(2)
			rs16975238	19:41439080T>A (T)	LVESV	(2)
ATXN2	N	Ataxin 2	rs35350651	12:111469627AC>A (A)	LVEDVi	(2)
			rs11065979	12:111621753C>T (C)	SV	(2)
			rs4766578	12:111466567A>T (T)	HF	(1)
			rs10774625	12:111472415A>G (G)	LVEDD	(4)
B3GNT7	N	UDP-GlcNAc:BetaGal beta-1,3-N-acetlyglucosomyltransfrase 7	rs146724382	2:231424120TTTC>T (TTTC)	SVi	(2)
BAG3	Y	BAG cochaperone 3	rs17617337	10:119667372C>T (C)	HF, EF	(1,5)
			rs72840788	10:119656173G>A (G)	LVEF, LVEDV, LVEDVi, LVESV, LVESVi	(2,8)
			rs2234962	10:119670121T>C (T)	All cause HF, NICM, DCM	(3,6,7)

			rs7071853	10:119552094T>C (T)	LVEDV	(8)
BEND3	N	BEN domain containing 3	rs9480737	6:107121073A>G (A)	LVEDVi, SVi	(2)
C1orf86	N	FA core complex associated protein 20 (*FAAP20*)	rs2503715	1:2212668G>A (A)	LVEF	(2)
CACNB4	N	Calcium voltage-gated channel auxillary subunit beta 4	rs150793926	2:151924550G>GTA (G)	DCM	(9)
CCDC141	N	Coiled-coil domain containing 141	rs1873164	2:178888822A>G (G)	LVEDV, LVEDVi	(2)
			rs7573293	2:178888518T>C (C)	SV, SVi	(2)
CDKN1A	N	Cyclin dependent kinase inhibitor 1A	rs146170154	6:36678991 C>CTA (C)	LVMass/EDVratio	(8)
			rs9470361	6:36655602G>A (A)	FS	(4)
			rs3176326	6:36679512G>A (G)	LVEF, LVESVi	(2)
			rs730506	6:36678191G>C (G)	LVESV	(2)
			rs4135240	6:36679903T>C (T)	HF	(1)
CDKN2B-AS1	N	CDKN2B antisense RNA 1	rs1556516	9:22100177G>C (C)	HF	(1)
			rs7857118	9:22124141T>A (T)	All cause HF	(6)
CELSR2	N	Cadherin EGF LAG seven-pass G-type receptor 2	rs660240	1:109275216C>T (C)	HF	(1)
CLCNKA	N	Chloride voltage-gated channel Ka	rs28579893	1:16021039G>A (A)	LVEDV	(2)
			rs945425	1:16021917C>T (T)	LVESVi, LVEF	(2,8)
			rs12138073	1:16028463C>T (T)	NICM	(6)
CSRP3	Y	Cysteine and glycine rich protein 3	rs721067	11:19191179T>A (T)	LVEF	(2)
			rs11604807	11:19209620T>C (T)	LVESV	(2)
DBX1	N	Developing brain homeobox 1	rs11025521	11:20348660G>T (G)	FS	(10)
DEFB136	N	Defensin beta 136	rs36029352	8:11929416T>C (C)	LVEF	(2)
DERL3	N	Derlin 3	rs5760061	22:23836092A>G (G)	LVESV, LVESVi, LVESD, FS, LVEF	(2,10)
			rs6003909	22:23839465G>A (A)	LVMass/EDVratio	(8)
DNAJC18	N	DnaJ heat shock protein family (HSP40) member C18	rs35999985	5:139421136G>A (A)	LVEF	(2)

EPHB1	N	EPH receptor B1	rs13092177	3:134736952G>T (G)	LVESV	(2)
FAM241A	N	Family with sequence similarity 241 member A	rs17042102	4:110747470G>A (A)	HF	(1)
FHOD3	Y	Formin homology 2 domain containing 3	rs2047273	18:36604896T>C (T)	LVEF	(2)
			rs2303510	18:36744128G>A (G)	DCM	(3)
FLNC	Y	Filamin C	rs3807309	7:128832084G>A (G)	LVEF	(2)
			rs34373805	7:128846309C>T (C)	LVESV, LVESVi	(2)
			rs757995302	7:128848928GC>G (G)	DCM	(11)
			rs2291569	7:128848680G>A (G)	DCM	(3)
FNDC3B	N	Fibronectin type III domain containing 3B	rs1499813	3:172042637 T>C (T)	LVESV, LVESVi	(2)
FTO	N	FTO alpha-ketoglutarate dependent dioxygenase	rs56094641	16:53772541A>G (G)	HF	(1)
GAREM1	N	GRB2 associated regulator of MAPK subtype 1	rs11874741	18:32497896A>G (A)	LVEDD, LVESD	(10)
GDF5	N	Growth differentiation factor 5	rs143384	20:35437976A>G (A)	SV	(2)
GJA1	N	Gap junction protein alpha 1 (connexin-43)	rs1919865	6:121799859A>T (A)	SVi	(2)
HAND2	Y	Heart and neural crest derivatives expressed 2	rs12499670	4:173700342C>T (T)	LVEDVi	(2)
HCG22	N	HLA complex group 22	rs9262636	6:31058071A>G (G)	DCM	(12)
HECTD4	N	HECT domain E3 ubiquitin protein ligase 4	rs10850034	12:112379717T>A (T)	LVEDV, LVEDVi, LVESV	(2)
			rs11066188	12:112172910G>A (G)	SV	(2)
HLA-B	N	Major histocompatibility complex, class I, B	rs4521636	6:31344516 C>T (T)	LVEDV	(2)
			rs111721712	6:31347630C>CT (C)	SV	(2)
HLA-DQA2	N	Major histocompatibility complex, class II, DQ alpha 2	rs9275587	6:32712602T>C (T)	LVEDV	(2)
HLA-DQB1	N	Major histocompatibility complex, class II, DQ beta 1	rs9274626	6:32668263C>T (T)	LVEF, LVESV	(2)
			rs28391274	6:32656009A>G (A)	SV	(2)

HLF	N	HLF transcription factor PAR BZIP family member	rs12452367	17:55297249T>C (T)	LVEF, LVESV, LVESVi	(2)
HSPB7	N	Heat shock protein family B (small) member 7	rs1739837	1:16011438T>C (C)	LVEF	(2)
			rs1048302	1:16014384G>T (T)	LVESV	(2)
ILF3	N	Interleukin enhancer binding factor 3	rs71164107	19:10654802C>CG (CG)	LVESV	(2)
INPP5F	N	Inositol polyphosphate-5-phosphatase F	rs3188055	10:119827370A>G (G)	DCM	(3)
KLHL3	N	Kelch like family member 3	rs11745324	5:137676482G>A (G)	HF	(1)
LLPH	N	LLP homolog, long-term synaptic facilitation factor	rs7306710	12:65982311C>T (T)	LVEDV	(2)
			rs10400419	12:65996188C>T (T)	SV	(2)
LMF1	N	Lipase maturation factor 1	rs5029142	16:938070T>A (T)	LVEF	(2)
			rs3829491	16:954834T>C (T)	LVESV	(2)
			rs8063213	16:942961T> (T)	LVESVi	(2)
LPA	N	Lipoprotein (A)	rs55730499	6:160584578C>T (T)	HF	(1)
			rs140570886	6:160591981T>C (C)	HF	(1)
MAP3K7CL	N	MAP3K7 C-terminal like	rs12627426	21:29147136 T>A (A)	All cause HF	(6)
MAPT	N	Microtubule associated protein tau	rs242562	17:45949373G>A (G)	LVESV, LVESVi	(2)
MECOM	N	MDS1 and EVI1 complex locus	rs6777123	3:169585282C>A (A)	LVEDV	(2)
MITF	N	Melanocyte inducing transcription factor	rs56099248	3:69808622C>T (C)	LVEF	(2)
			rs79502300	3:69807602C>T (C)	LVESVi	(2)
MLF1	N	Myeloid leukemia factor 1	rs2886037	3:158588625A>G (G)	LVESVi	(2)
MLIP	N	Muscular *LMNA* interacting protein	rs4712056	6:54124728G>A (G)	DCM	(3)
MTSS1	N	MTSS I-BAR domain containing 1	rs1324581981	8:124846296GA>G (GA)	LVEDV, LVEDVi, LVESV, LVESVi	(2)
			rs200712209	8:124846297 GA>G (GA)	LVESV	(8)
			rs34866937	8:124847608G>A (G)	LVEF, FS, LVEDD, LVESD	(8,10)
			rs12541595	8:124845117G>T (T)	LVEDD	(4)
MYH6	Y	Myosin heavy chain 6	rs422068	14:23395595T>C (T)	SV	(2)

			rs376439	14:23399820A>G (A)	SVi	(2)
MYO1C	N	Myosin 1C	rs2302455	17:1470901G>A (G)	LVEDV, LVESV, LVESVi	(2)
			rs7502466	17:1469676G>A (G)	LVEDVi	(2)
NEDD4L	N	NEDD4 like E3 ubiquitin protein ligase	rs10871753	18:5828963T>G (G)	LVEF, LVESV, LVESVi	(2)
NKX2-5	Y	NK2 homeobox 5	rs888690	5:173209127C>T (T)	SV	(2)
			rs758277832	5:173233109G>C (C)	DCM	(11)
NMB	N	Neuromedin B	rs1051168	15:84657289G>T (T)	DCM	(3)
NOS3	N	Nitric oxide synthase 3	rs3918226	7:150993088C>T (C)	LVEDV	(2)
PITX2	N	Paired like homeodomain 2	rs17042102	4:110747470G>A (A)	HF	(1)
			rs2634071	4:110748064C>T (T)	NICM	(6)
			rs1906609	4:110745295G>T (T)	All cause HF	(6)
PKD1	N	Polycystin 1, transient receptor potential channel interacting	rs71385734	16:2110502T>G (T)	LVEDV, LVESV	(2)
PLN	Y	Phospholamban	rs11153730	6:118346359T>C (T)	LVEDV, LVESV	(2)
			rs72967533	6:118333857T>C (T)	LVEDVi, SV, SVi	(2)
PRKCA	N	Protein kinase C alpha	rs9897002	17:66290376A>G (A)	LVESV	(2)
			rs9892651	17:66307675T>C (C)	LVESVi	(2)
PROB1	N	Proline rich basic protein 1	rs11748963	5:139394348T>C (T)	LVESVi	(2)
PTK2	N	Protein tyrosine kinase 2	rs4073554	8:140694133C>T (T)	LVEF	(2)
PXN	N	Paxillin	rs116904997	12:120230731G>A (G)	LVESV, LVESVi	(2)
RBM20	Y	RNA binding motif protein 20	rs189569984	10:110784367C>T (C)	LVEF, LVESVi	(2)
RNF207	N	Ring finger protein 207	rs709208	1:6212077A>G (A)	LVESVi	(2)
RPH3A	N	Rabphilin 3A	rs2891403	12:112699767G>A (A)	SV	(2)
RPL22	N	Ribosomal protein 22	rs114300540	1:6188122C>T (C)	LVESV	(2)
RRAS2	N	RAS related 2	rs10832164	11:14026933T>C (C)	LVESV	(2)
			rs11023059	11:14042845A>G (A)	LVESVi	(2)
RSPH6A	N	Radial spoke head 6 homolog A	rs12460541	19:45808819G>A (G)	LVEDV	(2)
			rs9797817	19:45809087C>T (C)	LVEDVi	(2)

			rs10421891	19:45812551A>G (A)	LVESV, LVESVi	(2)
RYBP	N	RING1 and YY1 binding protein	rs73839819	3:72530683A>G (G)	All cause HF	(6)
SESTD1	N	SEC14 and spectrin domain containing 1	rs190093681	2:179229625C>T (C)	LVESVi	(2)
SH2B3	N	SH2B adaptor protein 3	rs3184504	12:111446804C>T (C)	LVEDV, LVESV	(2)
			rs7310615	12:111427245G>C (C)	LVEDV	(8)
SLC1A4	N	Solute carrier family 1 member 4	rs6546120	2:65011273G>T (G)	FS, LVEF, LVESD	(10)
SLC35F1	N	Solute carrier family 35 member F1	rs11153730	6:118346359T>C (T)	LVEDD	(4)
SLC39A8	N	Solute carrier family 39 member 8	rs13107325	4:102267552C>T (T)	DCM	(3)
SMARCB1	N	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily B, member 1	rs2070458	22:23817120T>A (A)	LVEF	(2)
			rs5760061	22:23836092G>A (G)	LVEF, LVESD	(10)
			rs5760054	22:23819530C>T (C)	FS	(10)
SP3	N	Sp3 transcription factor	rs539762056	2:174027948A>AT (A)	LVEDV	(2)
SPATS2L	N	Spermatogenesis associated serine rich 2 like	rs767987273	2:200305786 CA>C (CA)	LVEDVi	(2)
			rs35858375	2:200333900CATT>C (CATT)	LVESVi	(2)
SPCS3	N	Signal peptidase complex subunit 3	rs1454157	4:176437647C>T (C)	LVMass	(4)
SPEN	N	Spen family transcriptional repressor	rs1976402	1:15817284G>A (G)	LVEDVi	(2)
			rs848210	1:15933318G>A (A)	DCM	(3)
SSPN	N	Sarcospan	rs113819537	12:26195496C>G (C)	LVEF, LVESV	(2)
SURF1	N	SURF1 cytochrome C oxidase assembly factor	rs600038	9:133276354T>C (C)	HF	(1)
SURF6	N	Surfeit 6	rs579459	9:133278724T>C (C)	SVi	(2)
SYNPO2L	N	Synaptopodin 2 like	rs4746140	10:73657491G>C (G)	HF	(1)
TMEM40	N	Transmembrane protein 40	rs11719526	3:12761814C>T (C)	LVEF	(5)
TTN	Y	Titin	rs2562845	2:178649706T>C (T)	LVEF, LVESV, LVESVi	(2)
			rs2042995	2:178693639T>C (T)	LVEF, LVEDV, LVESV	(8)
			rs2255167	2:178693555T>A (T)	LVMass	(8)

			rs3829746	2:178562809T>C (C)	DCM	(3)
VCL	Y	Vinculin	rs3812625	10:73997944A>G (A)	LVEDD	(10)
VEGFA	N	Vascular endothelial growth factor A	rs2146324	6:43789126C>A (A)	LVEDV, SV	(2)
			rs6458349	6:43792052A>G (G)	LVEDVi, SVi	(2)
XPC	N	XPC complex subunit, DNA damage recognition and repair factor	rs73028849	3:14231266G>C (G)	LVEDV, LVEDVi, LVESVi	(2)
			rs11710541	3:14250179T>C (T)	LVEF, LVESV	(2)
ZBTB17	N	Zinc finger and BTB domain containing 17	rs10927875	1:15972817C>T (C)	DCM, NICM	(3,6,7)
ZNF592	N	Zinc finger protein 592	rs149369954	15:84805731TTTTG>T (TTTTG)	LVEDV, LVESV, LVESVi, LVMass/EDVratio	(2,8)
			rs8023658	15:84779989G>T (T)	LVEF	(2)
ZNF638	N	Zinc finger protein 638	rs7605066	2:71302201T>C (T)	LVEDV	(2)

*Variants are denoted by their genomic location (hg38), risk allele is in brackets.
† (1) Shah et al. GWAS meta-analysis of heart failure comprising 47,309 cases and 930,014 controls.
(2) Pirruccello et al. GWAS of CMR-derived left ventricular measurements in 36,041 UK Biobank participants, with replication in 2184 participants from the Multi-Ethnic Study of Atherosclerosis.
(3) Esslinger et al. EWAS in 2796 dilated cardiomyopathy patients and 6877 control subjects from 6 populations of European ancestry.
(4) Wild et al. GWAS meta-analysis of echocardiographic traits including 46,533 individuals from 30 studies.
(5) Choquet et al. GWAS of ejection fraction in 26,638 adults from the Genetic Epidemiology Research on Adult Health and Aging and the UK Biobank cohort.
(6) Aragam et al. GWAS of heart failure in 394,156 participants from the UK Biobank.
(7) Villard et al. GWAS of dilated cardiomyopathy in 1179 DCM patients and 1108 controls.
(8) Aung et al. GWAS of CMR-derived left ventricular traits in 16,923 European UK Biobank participants.
(9) Xu et al. GWAS for idiopathic dilated cardiomyopathy in African Americans, 662 unrelated cases and 1167 controls.
(10) Kanai et al. GWAS of 58 quantitative traits including blood biomarkers, blood pressure and echocardiography in 162,255 Japanese individuals.
(11) Sveinbjornsson et al. GWAS of dilated cardiomyopathy in 424 cases and 337 689 population controls in Iceland.
(12) Meder et al. GWAS of dilated cardiomyopathy in 4100 cases and 7600 controls.

DCM = dilated cardiomyopathy; FS = fractional shortening; HF = heart failure; LVEDD = left ventricular end-diastolic diameter; LVEDV = left ventricular end-diastolic volume; LVEDVi = indexed left ventricular end-diastolic volume; LVEF = left ventricular ejection fraction; LVESD = left ventricular end-systolic diameter; LVESV = left ventricular end-systolic diameter; LVESVi = indexed left ventricular end-systolic diameter; LVMass = left ventricular mass; N= no; NICM = non-ischemic cardiomyopathy; SV = stroke volume; SVi = indexed stroke volume; Y =y es.

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