SUPPLEMENTAL MATERIAL

Table S1: Gene Panels
Khan et al. Genotype and Cardiac Outcomes in Pediatric Dilated Cardiomyopathy, 2021

Gene	Harvard Partners LMM 2007	Harvard Partners LMM 2009	GeneDx 2009	Transgenomic/ Familion 2009	Harvard Partners LMM 2011	GeneDx DCM 2011	Transgenomic/ Familion 2011	Harvard Partners LMM 2013	GeneDx 2013	Transgenomic/ Familion 2013	GeneDx 2017*	Invitae 2017*
ABCC9		✓			✓			✓			✓	✓
ACTC	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
ACTN2		✓			✓			✓	✓		✓	✓
AGL												✓
ALMS1											✓	
APLK3											✓	
ANKRD1					✓		✓	✓	✓	✓	✓	✓
BAG3								✓			✓	✓
BRAF											✓	
CACNA1C												✓
CALR3												✓
CASQ2					✓			✓				
CAV3					✓			✓			✓	✓
CHRM2											✓	✓
CRYAB					✓			✓			✓	✓
CSRP3		✓			✓			✓	✓		✓	✓
CTF1		✓			✓			✓				✓
CTNNA3												✓
DES		✓	✓		✓	✓		✓	✓		✓	✓
DMD											✓	✓
DOLK											✓	✓

Gene	Harvard Partners LMM 2007	Harvard Partners LMM 2009	GeneDx 2009	Transgenomic/ Familion 2009	Harvard Partners LMM 2011	GeneDx DCM 2011	Transgenomic/ Familion 2011	Harvard Partners LMM 2013	GeneDx 2013	Transgenomic/ Familion 2013	GeneDx 2017*	Invitae 2017*
DSC2					✓			✓			✓	✓
DSG2					✓			✓			✓	✓
DSP					✓			✓			✓	✓
DTNA					✓			✓			✓	✓
EMD		✓			✓			✓	✓		✓	✓
EYA4												✓
FHL1											✓	✓
FHL2					✓			✓				✓
FKRP											✓	✓
FKTN											✓	✓
FLNC												✓
GAA												✓
GATA4												✓
GATA6												✓
GATAD1								✓			✓	✓
GLA				✓	✓			✓			✓	✓
HCN4											✓	✓
HRAS											✓	
ILK											✓	✓
JPH2											✓	✓
JUP					✓			✓			✓	✓
KRAS											✓	
LAMA4					✓			✓			✓	✓
LAMP2			✓	✓	✓	✓		✓	✓		✓	✓

Gene	Harvard Partners LMM 2007	Harvard Partners LMM 2009	GeneDx 2009	Transgenomic/ Familion 2009	Harvard Partners LMM 2011	GeneDx DCM 2011	Transgenomic/ Familion 2011	Harvard Partners LMM 2013	GeneDx 2013	Transgenomic/ Familion 2013	GeneDx 2017*	Invitae 2017*
LDB3/ZASP	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓
LMNA	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓
LRRC10												✓
MAP2K1											✓	
MAP2K2											✓	
MIB1											✓	
MTND1 (NADH dehydro1)			✓			✓			✓		✓	
MTND5 (NADH dehydro5)			✓			✓			✓		✓	
MTND6 (NADH dehydro6)			✓			✓			✓		✓	
MTTD (tRNA Asp)						✓			✓		✓	
MTTG (tRNA Gly)											✓	
MTTH (tRNA His)			✓			✓			✓		✓	
MTTI (tRNA Ile)						✓			✓		✓	
MTTK (tRNA Lys)			✓			✓			✓		✓	
MTTL1 (tRNA Leu)			✓			✓			✓		✓	
MTTL2 (tRNA Leu2)						✓			✓		✓	
MTTM (methionine)						✓			✓		✓	
MTTQ (tRNA Gln)			✓			✓			✓		✓	
MTTS1 (tRNA Ser1)			✓			✓			✓		✓	
MTTS2 (tRNA Ser2)			✓			✓			✓		✓	
MURC											✓	
MYBPC3	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓

Gene	Harvard Partners LMM 2007	Harvard Partners LMM 2009	GeneDx 2009	Transgenomic/ Familion 2009	Harvard Partners LMM 2011	GeneDx DCM 2011	Transgenomic/ Familion 2011	Harvard Partners LMM 2013	GeneDx 2013	Transgenomic/ Familion 2013	GeneDx 2017*	Invitae 2017*
MYH6					✓			✓			✓	✓
MYH7	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
MYL2				✓	✓						✓	✓
MYL3				✓	✓			✓			✓	✓
MYLK2					✓			✓			✓	✓
MYOM1												✓
MYOZ2					✓			✓			✓	✓
MYPN											✓	✓
NEBL								✓			✓	✓
NEXN					✓			✓	✓		✓	✓
NKX2.5											✓	✓
NPPA												✓
NRAS											✓	
PDLIM3											✓	✓
PKP2					✓			✓			✓	✓
PLEKHM2												✓
PLN	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓
PRDM16											✓	✓
PRKAG2				✓	✓			✓			✓	✓
PTPN11											✓	
RAF1											✓	✓
RBM20					✓			✓	✓		✓	✓
RIT1											✓	
RYR2					✓			✓			✓	✓

Gene	Harvard Partners LMM 2007	Harvard Partners LMM 2009	GeneDx 2009	Transgenomic/ Familion 2009	Harvard Partners LMM 2011	GeneDx DCM 2011	Transgenomic/ Familion 2011	Harvard Partners LMM 2013	GeneDx 2013	Transgenomic/ Familion 2013	GeneDx 2017*	Invitae 2017*
SCN5A							✓	✓	✓	✓	✓	✓
SGCD		✓	✓		✓			✓	✓		✓	✓
SLC22A5												✓
SOS1											✓	
TAZ	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓
TCAP		✓			✓			✓			✓	✓
TGFB3					✓				✓		✓	✓
TMEM43								✓				✓
TMPO								✓			✓	✓
TNNC1				✓	✓		✓	✓	✓	✓	✓	✓
TNNI3	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
TNNT2	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
TPM1	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
TTN					✓			✓	✓		✓	✓
TTR			✓		✓	✓		✓	✓		✓	✓
TXNRD2											✓	✓
VCL		✓			✓			✓	✓		✓	✓

LMM = Laboratory for Molecular Medicine; * = deletion/duplication added in 2016

Table S2: Genotype and Variant Classification
Khan et al. Genotype and Cardiac Outcomes in Pediatric Dilated Cardiomyopathy, 2021

Gene	DNA change	Protein effect	Clinical Adjudication	Original Report Classification	External Lab Classification	Case Number [a]
ACTN2	c.26A>G	p.Gln9Arg	VUS	P	VUS	104
ACTN2	c.26A>G	p.Gln9Arg	VUS	LP	VUS	106
ACTN2	c.2630dupG	p.Ala878Cysfs*62	P/LP	VUS	VUS	1
BAG3	c.367C>T	p.Arg123X	P/LP	P	P/LP	2
LMNA	c.350A>G	p.Lys117Arg	P/LP	VUS	VUS	4
LMNA	c.868G>A	p.Glu290Lys	P/LP	VUS	P/LP	3
MT-TS2	m.12241delC		P/LP	LP	Not provided	5
MYBPC3	c.1721G>A	p.Arg574Gln	P/LP	VUS	VUS	7
MYBPC3	c.1294G>A	p.Ala432Thr	P/LP	VUS	VUS	8
MYBPC3	c.3628-41_3628-17del25	IVS32-41_IVS32-17del25	P/LP	VUS	P/LP	6
MYH7	c.3113T>C	p.Leu1038Pro	P/LP	LP	VUS	12
MYH7	c.2678C>T	p.Ala893Val	P/LP	VUS	P/LP	9
MYH7	c.4280A>T	p.Asp1427Val	P/LP	LP	VUS	17
MYH7	c.444C>G	p.Ser148Arg	P/LP	VUS	VUS	18
MYH7	c.1180G>A	p.Asp394Asn	P/LP	VUS	VUS	10
MYH7	c.541G>A	p.Gly181Arg	P/LP	VUS	VUS	15
MYH7	c.2585C>T	p.Ala862Val	P/LP	LP	VUS	16
MYH7	c.2711G>A	p.Arg904His	P/LP	P	P/LP	11
MYH7	c.844G>T	p.Asp282Tyr	P/LP	VUS	VUS	14
MYH7	c.1573G>A	p.Glu525Lys	P/LP	VUS	P/LP	13
MYH7	c.2710C>T	p.Arg904Cys	P/LP	P	P/LP	19
MYL2	c.401A>C	p.Glu134Ala	P/LP	P	VUS	12
PRDM16	c.1573dupC	p.Arg525Profs*79	P/LP	P	P/LP	20
TNNC1	c.184G>A	p.Asp62Asn	P/LP	VUS	VUS	21
TNNT2	c.421C>T	p.Arg141Trp	P/LP	P	P/LP	22
TNNT2	c.391C>T	p.Arg131Trp	P/LP	LP	P/LP	24
TNNT2	c.392G>A	p.Arg131Gln	P/LP	LP	P/LP	23
TNNT2	c.352G>A	p.Glu118Lys	P/LP	LP	VUS	25
TPM1	c.725C>T	p.Ala242Val	P/LP	P	VUS	27
TPM1	c.479G>A	p.Arg160His	P/LP	LP	P/LP	28
TPM1	c.118G>C	p.Glu40Gln	P/LP	VUS	VUS	29
TPM1	c.725C>T	p.Ala242Val	P/LP	P	VUS	26
TPM1	c.688G>A	p.Asp230Asn	P/LP	P	P/LP	30
TTN	c.28363+1G>A	Splice variant	P/LP	VUS	VUS	32
TTN	c.75793C>T	p.Arg25265*	P/LP	P	P/LP	34
TTN	c.92206_92207dupTA	p.Leu30738Hisfs*4	P/LP	P	P/LP	35
TTN	c.75703delG	p.Val25235Serfs*68	P/LP	LP	P/LP	38
TTN	c.94261_94262 dupGG	p.Leu31422Valfs*20	P/LP	P	P/LP	33
TTN	c.1800+1G>A	Splice variant	P/LP	LP	VUS	37
TTN	c.62572C>T	p.Arg20858*	P/LP	P	P/LP	36
TTN	c.102712C>T	p.Gln34238*	P/LP	VUS	P/LP	12
TTN	c.16387_16390delATCT	p.Ile5463Valfs*15	P/LP	VUS	VUS	37
TTN	c.73828G>T	p.Glu24610*	P/LP	LP	P/LP	31
TTR	c.424G>A	p.Val142Ile	P/LP	P	P/LP	16
TTR	c.424G>A	p.Val142Ile	P/LP	P	P/LP	39
VCL	c.562C>T	p.Arg188*	P/LP	P	VUS	40
			VUS only below			
ACTC1	c.623G>A	p.Arg208His	VUS	VUS	VUS	86
ACTC1	c.579G>T	p.Lys193Asn	VUS	VUS	VUS	61
ACTC1	c.119_129+14del25	p.Pro40Argfs*15	VUS	VUS	VUS	86
ACTN2	c.2306A>C	p.Lys769Thr	VUS	VUS	VUS	96
ACTN2	c.578_583del	p.His193_Arg194del	VUS	VUS	VUS	96
ACTN2	c.548G>A	p.Gly183Asp	VUS	VUS	VUS	47
ACTN2	c.1984C>T	p.Arg662Trp	VUS	VUS	VUS	11
ALMS1	c.11711G>A	p.Arg3904Gln	VUS	VUS	VUS	29
BAG3	c.474_476dup	p.Ala160dup	VUS	VUS	B/LB	96
CTF1	c.275C>A	p.Ala92Glu	VUS	VUS	VUS	24
DES	c.1009G>A	p.Ala337Thr	VUS	VUS	VUS	82
DES	c.935A>C	p.Asp312Ala	VUS	VUS	VUS	78
DES	c.170C>T	p.Ser57Leu	VUS	VUS	VUS	47
DES	c.934G>A	p.Asp312Asn	VUS	VUS	VUS	79
DMD	c.404A>G	p.Asn135Ser	VUS	VUS	VUS	77
DMD	c.9299A>G	p.Asn3100Ser	VUS	VUS	VUS	106
DSC2	c.1914G>C	p.Gln638His	VUS	VUS	B/LB	39
DSC2	c.1081G>A	p.Val361Met	VUS	VUS	VUS	61
DSC2	c.2446G>A	p.Val816Met	VUS	VUS	VUS	34
DSC2	c.26C>G	p.Ser9Cys	VUS	VUS	VUS	74

Table S2: Genotype and Variant Classification
Khan et al. Genotype and Cardiac Outcomes in Pediatric Dilated Cardiomyopathy, 2021

Gene	DNA change	Protein effect	Clinical Adjudication	Original Report Classification	External Lab Classification	Case Number [a]
DSP	c.2287T>C	p.Tyr763His	VUS	VUS	VUS	20
DSP	c.607G>A	p.Asp203Asn	VUS	VUS	VUS	100
DSP	c.4288A>T	p.Ile1430Phe	VUS	VUS	VUS	71
DSP	c.88G>A	p.Val30Met	B/LB	VUS	VUS	68
DSP	c.269A>G	p.Gln90Arg	VUS	VUS	VUS	11
DSP	c.4172A>G	p.Tyr1391Cys	VUS	VUS	VUS	50
DSP	c.2723G>A	p.Arg908His	VUS	VUS	VUS	35
DSP	c.1778A>G	p.Asn593Ser	VUS	VUS	B/LB	31
DSP	c.107G>C	p.Gly36Ala	VUS	VUS	VUS	34
DSP	c.8529_8540del12	p.Ser2843_Arg2846del	VUS	VUS	VUS	78
DSP	c.4372C>G	p.Arg1458Gly	VUS	VUS	VUS	29
DTNA	c.1228G>A	p.Asp410Asn	VUS	VUS	VUS	98
FKRP	c.404C>A	p.Ala135Asp	VUS	VUS	VUS	52
FLNC	c.6539G>A	p.Arg2180His	VUS	VUS	VUS	108
GLA	c.1153A>G	p.Thr385Ala	VUS	VUS	B/LB	61
GLA	c.427G>A	p.Ala143Thr	VUS	VUS	VUS	39
ILK	c.184G>A	p.Val62Ile	VUS	VUS	VUS	90
LAMA4	c.2599C>G	p.Leu867Val	VUS	VUS	VUS	29
LAMA4	c.2069G>A	p.Arg690His	VUS	VUS	VUS	51
LAMA4	c.1612C>T	p.Arg538Cys	VUS	VUS	B/LB	105
LDB3	c.752A>G	p.Lys251Arg	B/LB	LB	B/LB	15
LDB3	c.1606G>T	p.Val536Phe	VUS	VUS	VUS	39
LDB3	c.566C>T	p.Ser189Leu	VUS	VUS	VUS	52
LMNA	c.861T>C	Ala287Ala	B/LB	B	B/LB	13
MYBPC3	c.2980C>T	p.Leu994Phe	VUS	VUS	VUS	106
MYBPC3	c.472G>A	p.Val158Met	B/LB	LB	B/LB	72
MYBPC3	c.2870C>G	p.Thr957Ser	VUS	VUS	B/LB	12
MYBPC3	c.3682C>T	p.Arg1228Cys	VUS	VUS	VUS	100
MYBPC3	c.1519G>A	p.Gly507Arg	B/LB	VUS	B/LB	80
MYBPC3	c.472G>A	p.Val158Met	B/LB	LB	B/LB	21
MYH6	c.2401A>G	p.Ile801Val	VUS	VUS	VUS	98
MYH6	c.5089A>G	p.Thr1697Ala	VUS	VUS	VUS	73
MYH6	c.4293G>A	p.Met1431Ile	VUS	VUS	VUS	9
MYH6	c.3979-7_3979-6delTC		VUS	VUS	VUS	73
MYH7	c.5495G>A	p.Arg1832His	VUS	VUS	VUS	36
MYL2	c.141C>A	p.Asn47Lys	VUS	VUS	VUS	74
MYLK2	c.4G>A	p.Ala2Thr	B/LB	VUS	VUS	92
MYPN	c.1130G>A	p.Arg377Gln	B/LB	VUS	VUS	109
MYPN	c.3481C>A	p.Leu1161Ile	VUS	VUS	B/LB	47
MYPN	c.-166-1G>C	Splice variant	VUS	VUS	VUS	89
MYPN	c.1594G>A	p.Val532Met	VUS	VUS	VUS	35
NEBL	c.604G>A	p.Gly202Arg	VUS	VUS	VUS	2
NEBL	c.180G>C	p.Lys60Asn	B/LB	VUS	VUS	87
NKX2-5	c.632C>T	p.Pro211Leu	VUS	VUS	VUS	81
PDLIM3	c.926G>A	p.Arg309Gln	VUS	VUS	VUS	35
PDLIM3	c.697G>C	p.Val233Leu	B/LB	VUS	VUS	88
PRDM16	c.2815C>G	p.Leu939Val	VUS	VUS	VUS	11
PRKAG2	c.64G>T	p.Gly22Cys	VUS	VUS	VUS	64
RAF1	c.1721A>G	p.Tyr574Cys	VUS	VUS	VUS	95
RAF1	c.293T>C	p.Val98Ala	VUS	VUS	VUS	36
RBM20	c.1459G>A	p.Val487Met	VUS	LB	VUS	42
RBM20	c.3301G>A	p.Glu1101Lys	VUS	VUS	VUS	64
RBM20	c.2662G>A	p.Asp888Asn	VUS	VUS	B/LB	100
RBM20	c.2565_2570delACAGGA	p.Gln856_Glu857del	VUS	VUS	VUS	71
RBM20	c.2662G>A	p.Asp888Asn	VUS	VUS	B/LB	76
RYR2	c.6337G>A	p.Val2113Met	VUS	VUS	VUS	53
RYR2	c.2984T>C	p.Met995Thr	VUS	VUS	VUS	47
SCN5A	c.1425A>C	p.Arg475Ser	VUS	VUS	VUS	34
SCN5A	c.1820G>A	p.Gly607Asp	VUS	VUS	VUS	54
SCN5A	c.5972G>A	p.Arg1991Gln	VUS	VUS	VUS	50
SCN5A	c.2314G>A	p.Asp772Asn	VUS	VUS	VUS	76
SCN5A	c.1673A>G	p.His558Arg	B/LB	LB	B/LB	93
SCN5A	c.4342A>C	p.Ile1448Leu	VUS	VUS	VUS	53
SCN5A	c.3308C>A	p.Ser1103Tyr	B/LB	VUS	VUS	91
SGCD	c.717C>G	p.Asp239Glu	VUS	VUS	B/LB	71
SGCD	c.15G>C	p.Glu5Asp	B/LB	VUS	B/LB	41
SGCD	c.458A>G	p.Asp153Gly	VUS	VUS	VUS	47

Table S2: Genotype and Variant Classification
Khan et al. Genotype and Cardiac Outcomes in Pediatric Dilated Cardiomyopathy, 2021

Gene	DNA change	Protein effect	Clinical Adjudication	Original Report Classification	External Lab Classification	Case Number [a]
SOS2	c.3443_3454del	p.Pro1148_Pro1151del	VUS	VUS	VUS	108
TAZ	c.383T>C	p.Phe128Ser	B/LB	VUS	B/LB	99
TAZ	c.383T>C	p.Phe128Ser	B/LB	LB	B/LB	15
TMEM43	c.286C>G	p.Arg96Gly	VUS	VUS	VUS	78
TMPO	c.694C>T	p.Pro232Ser	VUS	VUS	VUS	36
TNNT2	c.224T>G	p.Val75Gly	VUS	VUS	VUS	93
TNNT2	c.178A>G	p.Met60Val	VUS	VUS	B/LB	22
TNNT2	c.178A>C	p.Met60Leu	VUS	VUS	VUS	53
TNNT2	c.587G>A	p.Arg196Gln	VUS	VUS	VUS	36
TTN	c.40166_40168delAAG	p.Glu13389del	VUS	VUS	VUS	106
VCL	c.2285G>A	p.Arg762Gln	VUS	VUS	VUS	12

[a] Case number is the same as in Table S4.

Table S3: Additional *TTN* Information
Khan et al. Genotype and Cardiac Outcomes in Pediatric Dilated Cardiomyopathy Patients, 2021

Gene	DNA change	Protein effect	Transcript Sequenced	Exon	PSI[a]	Protein Location	Case Number[b]
TTN	c.73828G>T	p.Glu24610*	NM_133378.4	275	100	A-band	31
TTN	c.28363+1G>A	Splice variant	NM_133378.4	122	54	I-band	32
TTN	c.102712C>T	p.Gln34238*	NM_001256850.1	312	100	A-band	12
TTN	c.94261_94262 dupGG	p.Leu31422Valfs*20	NM_001256850.1	304	100	A-band	33
TTN	c.75793C>T	p.Arg25265*	NM_001256850.1	276	100	A-band	34
TTN	c.92206_92207dupTA	p.Leu30738Hisfs*4	NM_001256850.1	298	100	A-band	35
TTN	c.62572C>T	p.Arg20858*	NM_001256850.1	269	100	A-band	36
TTN	c.1800+1G>A	Splice variant	NM_133378.4	I11[c]	N/A	Z-disk	37
TTN	c.16387_16390delATCT	p.Ile5463Valfs*15	NM_133379.3	45A	N/A	N/A	37
TTN	c.75703delG	p.Val25235Serfs*68	NM_133378.4	275	100	A-band	38
TTN	c.40166_40168delAAG	p.Glu13389del	NM_001256850.1	195	5	N/A	106

[a] PSI: Percentage spliced-in. The data shown represent exon usage in the human left ventricle, derived from patients with DCM and were obtained from Royal Brompton & Harefield (NHS Foundation Trust) at https://www.cardiodb.org/titin/titin_transcripts.php, accessed 15 Jun 2021.

[b] Case number is the same as in Table S4.

[c] Intron 11. Located in the invariant region (± 1,2) of the splice consensus sequence and predicted to cause altered splicing

Table S4: Phenotype and Genotype
Khan et al. Genotype and Cardiac Outcomes in Pediatric Dilated Cardiomyopathy, 2021

Case Number	Female Sex	Age at Presentation (Years)	Family History	Life-Threatening Cardiac Outcome	Ventricular Arrhythmia	Cardiac Arrest	Mechanical Support	Transplant	Death	Gene	cDNA	Protein	Year of Genetic Test
1	1	0	No family history	0	0	0	0	0	0	ACTN2	c.2630dupG	p.Ala878Cysfs*62	2016
2	1	0	No family history	1	0	0	1	1	0	BAG3	c.367C>T	p.Arg123*	2015
3	1	6	No family history	1	0	0	1	1	0	LMNA	c.868G>A	p.Glu290Lys	2017
4	1	1	No family history	0	0	0	0	0	0	LMNA	c.350A>G	p.Lys117Arg	2010
5	0	2	No family history	0	0	0	0	0	0	MT-TS2	m.12241delC		2015
6	0	0	1st degree FH	1	0	0	0	1	0	MYBPC3	c.3628-41_3628-17del25	IVS32-41_IVS32-17del25	2016
7	0	14	No family history	1	0	0	0	1	0	MYBPC3	c.1721G>A	p.Arg574Gln	2009
8	1	0	No family history	0	0	0	0	0	0	MYBPC3	c.1294G>A	p.Ala432Thr	2013
9	0	0	No family history	1	0	0	0	1	0	MYH7	c.2678C>T	p.Ala893Val	2014
10	1	0	No family history	1	0	0	1	1	0	MYH7	c.1180G>A	p.Asp394Asn	2012
11	0	0	1st degree FH	1	1	0	1	1	0	MYH7	c.2711G>A	p.Arg904His	2016
12	0	0	1st degree FH	1	0	1	0	1	0	MYH7	c.3113T>C	p.Leu1038Pro	2015
12	0	0	1st degree FH	1	0	1	0	1	0	MYL2	c.401A>C	p.Glu134Ala	2015
12	0	0	1st degree FH	1	0	1	0	1	0	TTN	c.102712C>T	p.Gln34238*	2015
13	1	0	No family history	1	0	0	1	1	0	MYH7	c.1573G>A	p.Glu525Lys	2008
14	1	0	1st degree FH	0	0	0	0	0	0	MYH7	c.844G>T	p.Asp282Tyr	2012
15	0	5	1st degree FH	0	0	0	0	0	0	MYH7	c.541G>A	p.Gly181Arg	2012
16	0	18	No family history	0	1	0	0	0	0	MYH7	c.2585C>T	p.Ala862Val	2015
16	0	18	No family history	0	1	0	0	0	0	TTR	c.424G>A	p.Val142Ile	2015
17	1	3	1st degree FH	0	0	0	0	0	0	MYH7	c.4280A>T	p.Asp1427Val	2014
18	1	0	No family history	0	0	0	0	0	0	MYH7	c.444C>G	p.Ser148Arg	2017
19	1	10	No family history	1	1	0	0	1	0	MYH7	c.2710C>T	p.Arg904Cys	2009
20	0	0	No family history	0	0	0	0	0	0	PRDM16	c.1573dupC	p.Arg525Profs*79	2016
21	1	15	1st degree FH	0	0	0	0	0	0	TNNC1	c.184G>A	p.Asp62Asn	2013
22	1	0	No family history	1	0	0	1	1	1	TNNT2	c.421C>T	p.Arg141Trp	2011
23	0	0	No family history	0	1	0	0	0	0	TNNT2	c.392G>A	p.Arg131Gln	2009
24	1	5	1st degree FH	0	0	0	0	0	0	TNNT2	c.391C>T	p.Arg131Trp	2013
25	0	0	No family history	1	1	0	0	1	0	TNNT2	c.352G>A	p.Glu118Lys	2013
26	1	0	1st degree FH	0	0	0	0	0	0	TPM1	c.725C>T	p.Ala242Val	2009
27	0	0	1st degree FH	0	0	0	0	0	0	TPM1	c.725C>T	p.Ala242Val	2009
28	1	0	No family history	1	0	0	0	1	0	TPM1	c.479G>A	p.Arg160His	2010
29	0	0	No family history	1	1	0	0	1	0	TPM1	c.118G>C	p.Glu40Gln	2017
30	1	0	1st degree FH	0	0	0	0	0	0	TPM1	c.688G>A	p.Asp230Asn	2007
31	1	6	1st degree FH	1	0	0	1	1	0	TTN	c.73828G>T	p.Glu24610*	2012
32	0	0	No family history	0	0	0	0	0	0	TTN	c.28363+1G>A		2013
33	0	18	1st degree FH	0	0	0	0	0	0	TTN	c.94261_94262 dupGG	p.Leu31422Valfs*20	2014
34	0	15	1st degree FH	0	1	0	0	0	0	TTN	c.75793C>T	p.Arg25265*	2016
35	0	16	No family history	1	1	0	1	1	0	TTN	c.92206_92207dupTA	p.Leu30738Hisfs*4	2014
36	1	13	No family history	1	1	0	0	1	0	TTN	c.62572C>T	p.Arg20858*	2016
37	0	1	Higher degree FH	0	0	0	0	0	0	TTN	c.1800+1G>A	Splice variant	2012
37	0	1	Higher degree FH	0	0	0	0	0	0	TTN	c.16387_16390delATCT	p.Ile5463Valfs*15	2012
38	0	14	No family history	1	1	0	0	1	0	TTN	c.75703delG	p.Val25235Serfs*68	2013
39	1	2	No family history	1	1	0	1	0	0	TTR	c.424G>A	p.Val142Ile	2015
40	1	0	No family history	1	0	0	0	1	0	VCL	c.562C>T	p.Arg188*	2010

Table S4: Phenotype and Genotype
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Case Number	Female Sex	Age at Presentation (Years)	Family History	Life-Threatening Cardiac Outcome	Ventricular Arrhythmia	Cardiac Arrest	Mechanical Support	Transplant	Death	Gene	cDNA	Protein	Year of Genetic Test
41	0	1	No family history	0	0	0	0	0	0	No P/LP Variants			
42	1	0	Higher degree FH	0	0	0	0	0	0	No P/LP Variants			
43	0	0	Higher degree FH	0	0	0	0	0	0	No P/LP Variants			
44	1	12	No family history	0	1	0	0	0	0	No P/LP Variants			
45	0	13	No family history	1	0	0	0	1	0	No P/LP Variants			
46	0	3	No family history	1	0	0	1	0	1	No P/LP Variants			
47	0	0	No family history	0	0	0	0	0	0	No P/LP Variants			
48	1	0	No family history	0	0	0	0	0	0	No P/LP Variants			
49	0	15	No family history	0	0	0	0	0	0	No P/LP Variants			
50	0	0	No family history	1	0	0	0	1	0	No P/LP Variants			
51	0	0	No family history	0	0	0	0	0	0	No P/LP Variants			
52	1	0	Higher degree FH	0	0	0	0	0	0	No P/LP Variants			
53	1	8	No family history	1	1	1	1	1	0	No P/LP Variants			
54	0	0	No family history	0	0	0	0	0	0	No P/LP Variants			
55	0	10	1st degree FH	1	1	1	0	1	0	No P/LP Variants			
56	0	0	1st degree FH	1	0	0	0	1	1	No P/LP Variants			
57	0	2	No family history	0	0	0	0	0	0	No P/LP Variants			
58	0	12	No family history	0	0	0	0	0	0	No P/LP Variants			
59	1	0	No family history	1	1	0	0	1	1	No P/LP Variants			
60	0	1	Higher degree FH	1	0	0	1	1	0	No P/LP Variants			
61	0	0	1st degree FH	0	0	0	0	0	0	No P/LP Variants			
62	1	0	1st degree FH	1	0	0	0	1	0	No P/LP Variants			
63	1	13	1st degree FH	1	0	0	0	1	0	No P/LP Variants			
64	1	1	No family history	0	0	0	0	0	0	No P/LP Variants			
65	1	1	No family history	0	0	0	0	0	0	No P/LP Variants			
66	0	14	No family history	0	1	0	0	0	0	No P/LP Variants			
67	0	0	No family history	1	0	0	0	1	0	No P/LP Variants			
68	1	13	No family history	0	1	0	0	0	0	No P/LP Variants			
69	0	10	No family history	1	0	0	0	1	1	No P/LP Variants			
70	0	16	No family history	0	1	0	0	0	0	No P/LP Variants			
71	1	0	No family history	0	0	0	0	0	0	No P/LP Variants			
72	0	1	1st degree FH	0	0	0	0	0	0	No P/LP Variants			
73	0	1	No family history	0	0	0	0	0	0	No P/LP Variants			
74	1	0	No family history	0	0	0	0	0	0	No P/LP Variants			
75	0	0	No family history	1	0	0	0	1	0	No P/LP Variants			
76	0	10	1st degree FH	1	0	0	0	1	0	No P/LP Variants			
77	1	0	No family history	1	0	1	0	1	0	No P/LP Variants			
78	1	1	No family history	1	1	1	0	0	0	No P/LP Variants			
79	0	17	Higher degree FH	0	1	0	0	0	0	No P/LP Variants			
80	1	0	No family history	1	0	0	0	1	0	No P/LP Variants			
81	0	0	No family history	0	0	0	0	0	0	No P/LP Variants			
82	1	0	No family history	0	0	0	0	0	0	No P/LP Variants			
83	0	1	Higher degree FH	1	0	0	0	1	0	No P/LP Variants			
84	0	4	Higher degree FH	1	0	0	0	1	0	No P/LP Variants			

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Case Number	Female Sex	Age at Presentation (Years)	Family History	Life-Threatening Cardiac Outcome	Ventricular Arrhythmia	Cardiac Arrest	Mechanical Support	Transplant	Death	Gene	cDNA	Protein	Year of Genetic Test
85	0	1	No family history	0	0	0	0	0	0	No P/LP Variants			
86	1	0	1st degree FH	0	0	0	0	0	0	No P/LP Variants			
87	1	0	No family history	0	0	0	0	0	0	No P/LP Variants			
88	0	0	No family history	0	0	0	0	0	0	No P/LP Variants			
89	1	4	No family history	1	1	1	1	0	0	No P/LP Variants			
90	0	1	No family history	0	0	0	0	0	0	No P/LP Variants			
91	0	1	No family history	0	0	0	0	0	0	No P/LP Variants			
92	0	16	1st degree FH	1	0	0	0	0	1	No P/LP Variants			
93	1	0	No family history	0	0	0	0	0	0	No P/LP Variants			
94	1	7	No family history	1	1	0	0	1	0	No P/LP Variants			
95	0	0	No family history	1	0	0	0	1	0	No P/LP Variants			
96	0	1	No family history	1	0	0	0	1	1	No P/LP Variants			
97	1	0	No family history	1	1	1	1	1	0	No P/LP Variants			
98	0	15	1st degree FH	0	0	0	0	0	0	No P/LP Variants			
99	1	2	No family history	0	1	0	0	0	0	No P/LP Variants			
100	0	0	No family history	1	0	0	0	1	0	No P/LP Variants			
101	1	1	No family history	1	1	0	1	1	0	No P/LP Variants			
102	0	0	1st degree FH	0	0	0	0	0	0	No P/LP Variants			
103	1	2	No family history	0	0	0	0	0	0	No P/LP Variants			
104	0	16	No family history	1	0	0	0	1	1	No P/LP Variants			
105	1	0	No family history	1	0	0	0	1	0	No P/LP Variants			
106	1	1	No family history	1	1	0	0	1	0	No P/LP Variants			
107	0	15	No family history	1	0	0	1	1	0	No P/LP Variants			
108	0	15	No family history	0	1	0	0	0	0	No P/LP Variants			
109	1	15	No family history	0	0	0	0	0	0	No P/LP Variants			

Abbreviations: * = Truncation; del = Deletion; fs = Frame shift; dup = Duplication. All variants are heterozygous, except 1 hemizygous and 1 mitochondrial variant.