

Appendix Table 1. List of main genes associated with inherited cardiomyopathy.

DISEASE	Gene OMIM id	GENE	CYTOGENETIC BAND	PROTEIN	PREVALENC	INHERITANCE
Brugada Syndrome (BrS)	601144	<i>SCN5A</i>	3p22.2	α subunit of the Nav1.5 sodium channel	20-25%	AD
	604427	<i>SCN10A</i>	3p22.2	α subunit of the Nav1.8 sodium channel	2,5-5%	AD
	606936	<i>TRPM4</i>	19q13.33	Calcium-activated non-selective ion channel	8%	AD
	611875	<i>CACNA1C</i>	12p13.33	α subunit $\alpha 1C$ of the Cav1.2 calcium channel	6-7%	AD
	601439	<i>ABCC9</i>	12p12.1	ATP-Binding Cassette, Subfamily C, Member 9	4-5%	AD
	611876	<i>CACNB2</i>	10p12.33-p12.31	β subunit Cav $\beta 2b$ of calcium channel	4-5%	AD
	602861	<i>PKP2</i>	12p11.21	Plakophilin 2	2,5%	AD
	152427	<i>KCNH2</i>	7q36.1	α subunit of the HERG potassium channel	1-2%	AD
	612838	<i>SCN1B</i>	19q13.12	β subunit Nav $\beta 1$ of sodium channel	1-2%	AD
	600465	<i>ANK3</i>	10q21.2	Ankyrin 3 (G)	Rare	AD
	114204	<i>CACNA2D1</i>	7q21.11	δ subunit Cava $2\delta 1$ of calcium channel	Rare	AD
	601513	<i>FGF12</i>	3q28-q29	Fibroblast growth factor 12	Rare	AD
	611777	<i>GPD1L</i>	3p22.3	Glycerol-3-phosphate dehydrogenase 1-like	Rare	AD
	613123	<i>HCN4</i>	15q24.1	Hyperpolarization-activated cyclic nucleotide-gated channel 4	Rare	AD
	604674	<i>HEY2</i>	6q22	Hairy/Enhancer of Split-related with YRPW motif 2	Rare	AD
	605410	<i>KCND2</i>	7q31.31	α subunit of the KV4.2 potassium channel	Rare	AD
	616399	<i>KCND3</i>	1p13.2	α subunit of the KV4.3 potassium channel	Rare	AD
	613119	<i>KCNE3</i>	11q13.4	β subunit MiRP2 of potassium channel	Rare	AD
	600935	<i>KCNJ8</i>	12p12.1	α subunit of the KIR6.1 potassium channel	Rare	AD
	607954	<i>RANGRF</i>	17p13.1	RAN guanine nucleotide release factor	Rare	AD
	601327	<i>SCN2B</i>	11q23.3	β subunit Nav $\beta 2$ of sodium channel	Rare	AD
	613120	<i>SCN3B</i>	11q24.1	β subunit Nav $\beta 3$ of sodium channel	Rare	AD
	603961	<i>SEMA3A</i>	7q21.11	Semaphorin family protein	Rare	AD
602701	<i>SLMAP</i>	3p14.3	Sarcolemma-associated protein	Rare	AD	
	152427	<i>KCNH2</i>	7q36.1	α subunit of the HERG potassium channel	75%	AD
	607542	<i>KCNQ1</i>	11p15.5-p15.4	Kv7.1 hERG/Kv11.1		AD/AR

Long QT Syndrome (LQT)	600163	<i>SCN5A</i>	3p22.2	α subunit of the Nav1.5 sodium channel	5-10%	AD		
	604001	<i>AKAP9</i>	7q21.2	A-kinase anchor protein 9		AD		
	106410	<i>ANK2</i>	4q25-q26	Ankyrin 2 (B)		AD		
	114205	<i>CACNA1C</i>	12p13.33	α subunit α 1C of the Cav1.2 calcium channel		AD		
	114180	<i>CALM1</i>	14q32.11	Calmodulin 1		AD		
	114182	<i>CALM2</i>	2p21	Calmodulin 2		AD		
	114183	<i>CALM3</i>	19q13.32	Calmodulin 3		AD		
	601253	<i>CAV3</i>	3p25.3	Caveolin 3		AD		
	176261	<i>KCNE1</i>	21q22.11-q22.12	Potassium voltage-gated channel subfamily E regulatory subunit 1		AD/AR		
	603796	<i>KCNE2</i>	21q22.11	Potassium voltage-gated channel subfamily E regulatory subunit 2		AD		
	600681	<i>KCNJ2</i>	17q24.3	Potassium inwardly-rectifying channel, subfamily J, member 2		AD/AR		
	600734	<i>KCNJ5</i>	11q24.3	Potassium inwardly-rectifying channel, subfamily J, member 5		AD		
	602235	<i>KCNQ2</i>	20q13.33	Potassium voltage-gated channel subfamily Q member 2		AD		
	180902	<i>RYR2</i>	1q43	Ryanodine Receptor 2		AD		
	600235	<i>SCN1B</i>	19q13.12	Sodium voltage-gated channel beta subunit 1		AD		
	608256	<i>SCN4B</i>	11q23.3	Sodium voltage-gated channel beta subunit 4		AD		
	601017	<i>SNTA1</i>	20q11.21	α 1-Syntrophin		AD		
	603283	<i>TRDN</i>	6q22.31	Triadin		AR		
	Short QT Syndrome (SQT)	152427	<i>KCNH2</i>	7q36.1		α subunit of the HERG potassium channel	15%	AD
		600681	<i>KCNJ2</i>	17q24.3		Potassium inwardly-rectifying channel, subfamily J, member 2	<5%	AD/AR
607542		<i>KCNQ1</i>	11p15.5-p15.4	Potassium voltage gated channel, KQT-like subfamily, member 1	<5%	AD		
611875		<i>CACNA1C</i>	12p13.33	α subunit α 1C of the Cav1.2 calcium channel	Rare	AD		
114204		<i>CACNA2D1</i>	7q21.11	δ subunit Cava2 δ 1 of calcium channel	Rare	AD		
611876		<i>CACNB2</i>	10p12.33-p12.31	β subunit Cav β 2b of calcium channel	Rare	AD		
601144		<i>SCN5A</i>	3p22.2	α subunit of the Nav1.5 sodium channel	Rare	AD		
106195		<i>SLC4A3</i>	2q35	Solute carrier family 4 (anion exchanger), member 3	Rare	AD		
	180902	<i>RYR2</i>	1q43	Ryanodine Receptor 2	60%	AD		

Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)	114251	<i>CASQ2</i>	1p13.1	Calsequestrin 2	5%	AR
	600681	<i>KCNJ2</i>	17q24.3	Potassium inwardly-rectifying channel, subfamily J, member 2	1%	AR
	106410	<i>ANK2</i>	4q25-q26	Ankyrin 2 (B)	Rare	AD
	114180	<i>CALM1</i>	14q32.11	Calmodulin 1	Rare	AD
	114182	<i>CALM2</i>	2p21	Calmodulin 2	Rare	AD
	114183	<i>CALM3</i>	19q13.32	Calmodulin 3	Rare	AD
	603283	<i>TRDN</i>	6q22.31	Triadin	Rare	AR
Arrhythmogenic Cardiomyopathy (ACM)	602861	<i>PKP2</i>	12p11.21	Plakophilin 2	30-40%	AD
	125671	<i>DSG2</i>	18q12.1	Desmoglein 2	3-20%	AD
	125647	<i>DSP</i>	6p24	Desmoplakin	3-15%	AD
	188840	<i>TTN</i>	2q31.2	Titin	0-10%	AD
	125645	<i>DSC2</i>	18q12.1	Desmocollin 2	1-8%	AD/AR
	150330	<i>LMNA</i>	1q22	Lamin A/C	0-4%	AD
	102565	<i>FLNC</i>	7q32.1	Filamin C	3%	AD
	601144	<i>SCN5A</i>	3p22.2	α subunit of the Nav1.5 sodium channel	2%	AD
	114020	<i>CDH2</i>	18q12.1	Cadherin 2 - N-Cadherin	0-2%	AD
	607667	<i>CTNNA3</i>	10q22.2	Catenin, alpha 3	0-2%	AD
	125660	<i>DES</i>	2q35	Desmin	0-2%	AD
	612048	<i>TMEM43</i>	3p25.1	Transmembrane protein 43	0-2%	AD
	173325	<i>JUP</i>	17q21	Junction plakoglobin	Rare	AD
	172405	<i>PLN</i>	6q22.31	Phospholamban	Rare	AR
	180902	<i>RYR2</i>	1q43	Ryanodine receptor 2	Rare	AD
	190230	<i>TGFB3</i>	14q24.3	Transforming Growth Factor β 3	Rare	AD
	Dilated Cardiomyopathy (DCM)	188840	<i>TTN</i>	2q31.2	Titin	12-25%
150330		<i>LMNA</i>	1q22	Lamin A/C	10%	AD
102540		<i>ACTC1</i>	15q14	Actin, alpha, cardiac muscle	5-10%	AD
600958		<i>MYBPC3</i>	11p11.2	Myosin-binding protein C, cardiac		AD
160760		<i>MYH7</i>	14q11.2	Myosin, heavy chain 7, cardiac muscle, beta		AD
160781		<i>MYL2</i>	12q24.11	Myosin light chain 2		AD
160790		<i>MYL3</i>	3p21.31	Myosin light chain 3		AD/AR
191044		<i>TNNI3</i>	19q13.42	Cardiac troponin I3		AD
191045		<i>TNNT2</i>	1q32.1	Cardiac troponin T2	AD	
191010		<i>TPM1</i>	15q22.2	Tropomyosin 1	AD	
125645		<i>DSC2</i>	18q12.1	Desmocollin 2	5%	AD
125671		<i>DSG2</i>	18q12.1	Desmoglein 2		AD
125647		<i>DSP</i>	6p24	Desmoplakin		AD

173325	<i>JUP</i>	17q21	Junction plakoglobin		AD
602861	<i>PKP2</i>	12p11.21	Plakophilin 2		AD
613171	<i>RBM20</i>	10q25.2	RNA-binding motif protein 20	1-5%	AD
601144	<i>SCN5A</i>	3p22.2	α subunit of the Nav1.5 sodium channel	2-3%	AD
608517	<i>MYPN</i>	10q21.3	Myopalladin	2%	AD
102573	<i>ACTN2</i>	1q43	Alpha-actinin 2	1%	AD
102565	<i>FLNC</i>	7q32.1	Filamin C	1%	AD
172405	<i>PLN</i>	6q22.31	Phospholamban	1%	AR
193065	<i>VCL</i>	10q22.2	Vinculin	1%	AD
601439	<i>ABCC9</i>	12p12.1	ATP-Binding Cassette, Subfamily C, Member 9	Rare	AD
602330	<i>ABLIM1</i>	10q25.3	Limatin (actin-binding LIM domain protein)	Rare	AD
606844	<i>ALMS1</i>	2p13.1	ALMS1 centrosome and basal body associated protein	Rare	AR
609599	<i>ANKRD1</i>	10q23.31	Ankyrin repeat domain-containing protein 1	Rare	AD
608662	<i>ANO5</i>	11p14.3	Anoctamin 5	Rare	AR
603883	<i>BAG3</i>	10q26.1	BCL2-associated athanogene	Rare	AD
611414	<i>CALR3</i>	19p13.11	Calreticulin 3	Rare	AD
114251	<i>CASQ2</i>	1p13.1	Calsequestrin 2	Rare	AD
601253	<i>CAV3</i>	3p25.3	Caveolin 3	Rare	AD
123590	<i>CRYAB</i>	11q23.1	Alpha B crystallin	Rare	AD
600824	<i>CSRP3</i>	11p15.1	Cysteine- and glycine-rich protein 3	Rare	AD
600435	<i>CTF1</i>	16p11.2	Cardiotrophin 1	Rare	AD
128239	<i>DAG1</i>	3p21.31	Dystroglycan, alpha	Rare	AR
125660	<i>DES</i>	2q35	Desmin	Rare	AD
300377	<i>DMD</i>	Xp21.2-p21.1	Dystrophin	Rare	XLR
605377	<i>DMPK</i>	19q13.32	Dystrophia myotonica protein kinase gene	Rare	AD
610746	<i>DOLK</i>	9q34.11	Dolichol Kinase	Rare	AR
601239	<i>DTNA</i>	18q12.1	Dystrobrevin, alpha	Rare	AD/AR
300384	<i>EMD</i>	Xq28	Emerin	Rare	XLR
603550	<i>EYA4</i>	6q23.2	Eyes absent 4	Rare	AD
300163	<i>FHL1</i>	Xq26.3	Four-and -a-half LIM domains 1	Rare	XLD/XLR
602633	<i>FHL2</i>	2q12.2	Four-and -a-half LIM domains 2	Rare	AD
614518	<i>GATAD1</i>	7q21.2	GATA Zinc Finger Domain Containing Protein 1	Rare	AD
300644	<i>GLA</i>	Xq22.1	Galactosidase, alpha	Rare	XLR
602366	<i>ILK</i>	11p15.4	Integrin-linked kinase	Rare	AD

156225	<i>LAMA2</i>	6q22.33	Laminin Alpha, 2	Rare	AR
600133	<i>LAMA4</i>	6q21	Laminin Alpha, 4	Rare	AD
309060	<i>LAMP2</i>	Xq24	Lysosome-associated membrane protein 2	Rare	XLR
605906	<i>LDB3</i>	10q23.2	LIM domain-binding 3	Rare	AD
160710	<i>MYH6</i>	14q11.2	Alpha-myosin heavy chain 6	Rare	AD
605603	<i>MYOZ1</i>	10q22.2	Myozenin 1	Rare	AD/AR
605602	<i>MYOZ2</i>	4q26	Myozenin 2	Rare	AD
605491	<i>NEBL</i>	10p12.31	Nebulette	Rare	AD
613121	<i>NEXN</i>	1p31.1	Nexilin	Rare	AD
600584	<i>NKX2-5</i>	5q35.1	NK2 homeobox 5; cardiac specific homeobox 1	Rare	AD
605900	<i>PDLIM1</i>	10q23.33	C-terminal LIM domain protein 1	Rare	AD
605889	<i>PDLIM3</i>	4q35.1	PDZ and LIM domain protein 3	Rare	AD
603422	<i>PDLIM4</i>	5q31.1	PDZ and LIM domain protein 4	Rare	AD
605557	<i>PRDM16</i>	1p36.32	PR domain containing 16	Rare	AD
602743	<i>PRKAG2</i>	7q36.1	Protein Kinase, AMP-Activated, Non-Catalytic, Gamma 2	Rare	AD
104311	<i>PSEN1</i>	14q24.2	Presenilin 1	Rare	AD
600759	<i>PSEN2</i>	1q42.13	Presenilin 2	Rare	AD
176876	<i>PTPN11</i>	12q24.13	Protein-Tyrosine Phosphatase, Non-Receptor Type, 11	Rare	AD
609591	<i>RIT1</i>	1q22	RIC-like protein without CAAX motif 1	Rare	AD
180902	<i>RYR2</i>	1q43	Ryanodine Receptor 2	Rare	AD
601411	<i>SGCD</i>	5q33.2-q33.3	Delta-sarcoglycan	Rare	AD
603377	<i>SLC22A5</i>	5q31.1	Solute Carrier Family 22 (Organic Cation Transporter), Member 5	Rare	AR
601017	<i>SNTA1</i>	20q11.21	α 1-Syntrophin	Rare	AD
182530	<i>SOS1</i>	2p22.1	SOS Ras/Rac guanine nucleotide exchange factor 1	Rare	AD
607723	<i>SUN1</i>	7p22.3	SAD1 and UNC84 domain-containing protein 1	Rare	AD
613569	<i>SUN2</i>	22q13.1	SAD1 and UNC84 domain-containing protein 2	Rare	AD
608441	<i>SYNE1</i>	6q25.2	Nesprin 1, Synaptic nuclear envelop protein 1	Rare	AD
608442	<i>SYNE2</i>	14q23.2	Nesprin 2, Synaptic nuclear envelop protein 2	Rare	AD
300394	<i>TAZ</i>	Xq28	Tafazzin	Rare	XLR

	601620	<i>TBX5</i>	12q24.21	T-box 5	Rare	AD
	604488	<i>TCAP</i>	17q12	Titin-cap; telethonin	Rare	AD/AR
	603306	<i>TCF21</i>	6q23.2	Transcription factor 21, epicardin	Rare	AD
	190230	<i>TGFB3</i>	14q24.3	Transforming Growth Factor β 3	Rare	AD
	612048	<i>TMEM43</i>	3p25.1	Transmembrane protein 43	Rare	AD
	188380	<i>TMPO</i>	12q23.1	Thymopoietin	Rare	AD
	191040	<i>TNNC1</i>	3p21.1	Cardiac troponin C	Rare	AD
	176300	<i>TTR</i>	18q12.1	Transthyretin	Rare	AD
Hypertrophic Cardiomyopathy (HCM)	600958	<i>MYBPC3</i>	11p11.2	Myosin-binding protein C, cardiac	30-40%	AD
	160760	<i>MYH7</i>	14q11.2	Myosin, heavy chain 7, cardiac muscle, beta	20-30%	AD
	191045	<i>TNNT2</i>	1q32.1	Cardiac troponin T2	10-20%	AD
	191044	<i>TNNI3</i>	19q13.42	Cardiac troponin I3	2-5%	AD
	191010	<i>TPM1</i>	15q22.2	Tropomyosin 1	2-5%	AD
	102540	<i>ACTC1</i>	15q14	Actin, alpha, cardiac muscle	1-3%	AD
	160781	<i>MYL2</i>	12q24.11	Myosin light chain 2	1-3%	AD
	160790	<i>MYL3</i>	3p21.31	Myosin light chain 3	1-3%	AD/AR
	102573	<i>ACTN2</i>	1q43	Alpha-actinin 2	Rare	AD
	603883	<i>BAG3</i>	10q26.1	BCL2-associated athanogene	Rare	AD
	611414	<i>CALR3</i>	19p13.11	Calreticulin 3	Rare	AD
	601253	<i>CAV3</i>	3p25.3	Caveolin 3	Rare	AD
	123590	<i>CRYAB</i>	11q23.1	Alpha B crystallin	Rare	AD
	600824	<i>CSRP3</i>	11p15.1	Cysteine- and glycine-rich protein 3	Rare	AD
	125660	<i>DES</i>	2q35	Desmin	Rare	AD
	300163	<i>FHL1</i>	Xq26.3	Four-and -a-half LIM domains 1	Rare	XLRD/XLRR
	602633	<i>FHL2</i>	2q12.2	Four-and -a-half LIM domains 2	Rare	AD
	102565	<i>FLNC</i>	7q32.1	Filamin C	Rare	AD
	300644	<i>GLA</i>	Xq22.1	Galactosidase, alpha	Rare	XLR
	602366	<i>ILK</i>	11p15.4	Integrin-linked kinase	Rare	AD
	605267	<i>JPH2</i>	20q13.12	Junctophilin 2	Rare	AD
	309060	<i>LAMP2</i>	Xq24	Lysosome-associated membrane protein 2	Rare	XLR
	605906	<i>LDB3</i>	10q23.2	LIM domain-binding 3	Rare	AD
	160710	<i>MYH6</i>	14q11.2	Alpha-myosin heavy chain 6	Rare	AD
	609928	<i>MYH7B</i>	20q11.22	Myosin Heavy Chain 7B	Rare	AD
	606566	<i>MYLK2</i>	20q11.21	Myosin light chain kinase 2	Rare	AD
	605603	<i>MYOZ1</i>	10q22.2	Myozenin 1	Rare	AD/AR
	605602	<i>MYOZ2</i>	4q26	Myozenin 2	Rare	AD
	608517	<i>MYPN</i>	10q21.3	Myopalladin	Rare	AD

613121	<i>NEXN</i>	1p31.1	Nexilin	Rare	AD
605900	<i>PDLIM1</i>	10q23.33	C-terminal LIM domain protein 1	Rare	AD
605889	<i>PDLIM3</i>	4q35.1	PDZ and LIM domain protein 3	Rare	AD
603422	<i>PDLIM4</i>	5q31.1	PDZ and LIM domain protein 4	Rare	AD
172405	<i>PLN</i>	6q22.31	Phospholamban	Rare	AD
602743	<i>PRKAG2</i>	7q36.1	Protein Kinase, AMP-Activated, Non-Catalytic, Gamma 2	Rare	AD
176876	<i>PTPN11</i>	12q24.13	Protein-Tyrosine Phosphatase, Non-Receptor Type, 11	Rare	AD
604488	<i>TCAP</i>	17q12	Titin-cap; telethonin	Rare	AD /AR
612418	<i>TMEM70</i>	8q21.11	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	Rare	AR
191040	<i>TNNC1</i>	3p21.1	Cardiac troponin C	Rare	AD
188840	<i>TTN</i>	2q31.2	Titin	Rare	AD
176300	<i>TTR</i>	18q12.1	Transthyretin	Rare	AD
193065	<i>VCL</i>	10q22.2	Vinculin	Rare	AD

AD, Autosomic Dominant; AR, Autosomic Recessive; XLR, X-Linked Recessive; XLD, X-Linked Dominant; Rare = <1%