



Description	# of Genes	Genes	ICD-10 Suggestions*
LIPIDEMIAS			
Familial Hypercholesterolemia			
Includes the genes most commonly associated with familial hypercholesterolemia.	4	APOB LDLR LDLRAP1 PCSK9	E78.01 Familial hypercholesterolemia Z82.41 Family history of sudden cardiac death Z84.81 Family history of carrier of genetic disease Z86.74 Personal history of sudden cardiac arrest
ARRHYTHMIAS			
Pan Arrhythmia			
Includes the genes most commonly associated with heritable arrhythmias, including long QT syndrome, short QT syndrome, arrhythmogenic cardiomyopathy, catecholaminergic polymorphic ventricular tachycardia, and Brugada syndrome.	42	ABCC9 ACTN2 ANK2 CACNA1C CACNA2D1 CACNB2 CALM1 CALM2 CALM3 CASQ2 CAV3 DES DSC2 DSG2 DSP EMD GPD1L HCN4 JUP KCNE1 KCNE2 KCNE3 KCNH2 KCNJ2 KCNQ1 LDB3 LMNA PKP2 PLN PRKAG2 RBM20 RYR2 SCN10A SCN4B SCN5A SNTA1 TGFB3 TMEM43 TNNI3 TNNT2 TRDN TTN	I45.81 Long QT syndrome I49.9 Catecholaminergic polymorphic ventricular tachycardia (CPVT) I49.9 Short QT syndrome Q23.8 Brugada syndrome R55 Syncope and collapse R94.31 Abnormal electrocardiogram (ECG or EKG) Z82.41 Family history of sudden cardiac death Z84.81 Family history of carrier of genetic disease Z86.74 Personal history of sudden cardiac arrest
Emerging Evidence Genes			
Add-on genes for which there is emerging evidence that mutations in these genes are related to heritable arrhythmias.	14	AKAP9 ANKRD1 CTNNA3 KCND3 KCNE5 KCNJ5 KCNJ8 PDLIM3 RANGRF SCN1B SCN2B SCN3B SLMAP TRPM4	
Brugada Syndrome			
Includes the genes most commonly associated with Brugada syndrome.	10	ABCC9 CACNA1C CACNB2 GPD1L HCN4 KCNE3 KCNH2 PKP2 SCN10A SCN5A	Q23.8 Brugada syndrome R55 Syncope and collapse R94.31 Abnormal electrocardiogram (ECG or EKG) Z82.41 Family history of sudden cardiac death Z84.81 Family history of carrier of genetic disease Z86.74 Personal history of sudden cardiac arrest
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to Brugada syndrome.	10	CACNA2D1 KCND3 KCNE5 KCNJ8 RANGRF SCN1B SCN2B SCN3B SLMAP TRPM4	
Catecholaminergic Polymorphic Ventricular Tachycardia			
Includes the genes most commonly associated with catecholaminergic polymorphic ventricular tachycardia syndrome.	8	ANK2 CALM1 CALM2 CALM3 CASQ2 KCNJ2 RYR2 TRDN	I46.2 Cardiac arrest I49.9 Catecholaminergic polymorphic ventricular tachycardia (CPVT) R55 Syncope and collapse R94.31 Abnormal electrocardiogram (ECG or EKG) Z82.41 Family history of sudden cardiac death Z84.81 Family history of carrier of genetic disease Z86.74 Personal history of sudden cardiac arrest
Long QT Syndrome			
Includes the genes most commonly associated with long QT syndrome.	15	ANK2 CACNA1C CALM1 CALM2 CALM3 CAV3 KCNE1 KCNE2 KCNH2 KCNJ2 KCNQ1 SCN4B SCN5A SNTA1 TRDN	I45.81 Long QT syndrome I46.2 Cardiac arrest R55 Syncope and collapse R94.31 Abnormal electrocardiogram (ECG or EKG) Z82.41 Family history of sudden cardiac death Z84.81 Family history of carrier of genetic disease Z86.74 Personal history of sudden cardiac arrest
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to long QT syndrome.	2	AKAP9 KCNJ5	
Short QT Syndrome			
Includes the genes most commonly associated with short QT syndrome.	6	CACNA1C CACNA2D1 CACNB2 KCNH2 KCNJ2 KCNQ1	I49.9 Short QT syndrome R55 Syncope and collapse R94.31 Abnormal electrocardiogram (ECG or EKG) Z82.41 Family history of sudden cardiac death Z84.81 Family history of carrier of genetic disease Z86.74 Personal history of sudden cardiac arrest

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Description	# of Genes	Genes	ICD-10 Suggestions*
CARDIOMYOPATHIES			
Pan Cardiomyopathy			
Includes genes most commonly associated with heritable cardiomyopathies, including dilated cardiomyopathy, hypertrophic cardiomyopathy, arrhythmogenic cardiomyopathy, left ventricular noncompaction, and restrictive cardiomyopathy.	52	<p>ABCC9 ACTC1 ACTN2 AGL BAG3 CACNA1C CAV3 CRYAB</p> <p>CSRP3 DES DMD DOLK DSC2 DSG2 DSP EMD EYA4</p> <p>FHL1 FKRP FKTN FLNC GAA GLA HCN4 JUP LAMP2</p> <p>LDB3 LMNA MYBPC3 MYH7 MYL2 MYL3 PKP2 PLN</p> <p>PRKAG2 RAF1 RBM20 RYR2 SCN5A SGCD SLC22A5 TAZ</p> <p>TCAP TGFB3 TMEM43 TNNC1 TNNI3 TNNT2 TPM1 TTN</p> <p>TTR VCL</p>	<p>I42.0 Dilated cardiomyopathy</p> <p>I42.1 Hypertrophic obstructive cardiomyopathy</p> <p>I42.2 Hypertrophic non-obstructive cardiomyopathy</p> <p>I42.5 Cardiomyopathy, other restrictive</p> <p>I42.8 Other cardiomyopathies</p> <p>I42.9 Cardiomyopathy, unspecified</p> <p>I43 Syndromic cardiomyopathy</p> <p>R55 Syncope and collapse</p> <p>Z82.41 Family history of sudden cardiac death</p> <p>Z84.81 Family history of carrier of genetic disease</p> <p>Z86.74 Personal history of sudden cardiac arrest</p>
Emerging Evidence Genes			
Add-on genes for which there is emerging evidence that mutations in these genes are related to heritable cardiomyopathies.	27	<p>ANKRD1 CALR3 CHRM2 CTF1 CTNNA3 DTNA FHL2 GATA4</p> <p>GATA6 GATAD1 ILK JPH2 LAMA4 LRRC10 MYH6 MYLK2</p> <p>MYOM1 MYOZ2 MYPN NEBL NEXN NKX2-5 NPPA PDLIM3</p> <p>PLEKHM2 TMPO TXNRD2</p>	
RASopathies			
Add-on RASopathy genes for a related group of disorders, including Noonan syndrome and cardiofaciocutaneous syndrome, that often include HCM as a feature.	17	<p>A2ML1 BRAF CBL HRAS KRAS MAP2K1 MAP2K2 NF1</p> <p>NRAS PTPN11 RASA1 RIT1 RRAS SHOC2 SOS1 SOS2</p> <p>SPRED1</p>	
Recessive Pediatric Syndrome			
Add-on genes related to recessive, pediatric-onset syndromes that may have cardiomyopathy as a feature.	8	<p>ACADVL ALMS1 CPT2 DNAJC19 ELAC2 MTO1 SDHA</p> <p>TMEM70</p>	
Arrhythmogenic Cardiomyopathy			
Includes genes that cause disorders that can present with both arrhythmias and cardiomyopathies, including arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVC).	20	<p>ACTN2 DES DSC2 DSG2 DSP EMD JUP LDB3</p> <p>LMNA PKP2 PLN PRKAG2 RBM20 RYR2 SCN5A</p> <p>TGFB3 TMEM43 TNNI3 TNNT2 TTN</p>	<p>I42.9 Cardiomyopathy, unspecified</p> <p>I43 Syndromic cardiomyopathy</p> <p>I47.2 Ventricular tachycardia</p> <p>I49.8 Other specified cardiac arrhythmias</p> <p>I51.7 Ventricular hypertrophy</p> <p>R94.31 Abnormal electrocardiogram (ECG or EKG)</p> <p>Z82.41 Family history of sudden cardiac death</p> <p>Z84.81 Family history of carrier of genetic disease</p> <p>Z86.74 Personal history of sudden cardiac arrest</p>
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to arrhythmogenic cardiomyopathy.	3	<p>ANKRD1 CTNNA3 PDLIM3</p>	
Dilated Cardiomyopathy			
Includes the genes most commonly involved in dilated cardiomyopathy (DCM). Includes genes that cause primary DCM as well as syndromic conditions with DCM as a symptom, including Danon disease, Duchenne and Becker muscular dystrophy, Emery-Dreifuss muscular dystrophy, and transthyretin amyloidosis.	42	<p>ABCC9 ACTC1 ACTN2 BAG3 CAV3 CRYAB CSRP3</p> <p>DES DMD DOLK DSC2 DSG2 DSP EMD EYA4</p> <p>FKRP FKTN FLNC JUP LAMP2 LDB3 LMNA MYBPC3</p> <p>MYH7 PKP2 PLN RAF1 RBM20 RYR2 SCN5A SGCD</p> <p>SLC22A5 TAZ TCAP TMEM43 TNNC1 TNNI3 TNNT2</p> <p>TPM1 TTN TTR VCL</p>	<p>I42.0 Dilated cardiomyopathy</p> <p>I42.9 Cardiomyopathy, unspecified</p> <p>I43 Syndromic cardiomyopathy</p> <p>I51.7 Ventricular hypertrophy</p> <p>Z82.41 Family history of sudden cardiac death</p> <p>Z84.81 Family history of carrier of genetic disease</p> <p>Z86.74 Personal history of sudden cardiac arrest</p>
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to DCM.	20	<p>ANKRD1 CHRM2 CTF1 FHL2 GATA4 GATA6 GATAD1</p> <p>ILK LAMA4 LRRC10 MYH6 MYPN NEBL NEXN</p> <p>NKX2-5 NPPA PLEKHM2 PDLIM3 TMPO TXNRD2</p>	
Recessive Pediatric Syndrome			
Add-on genes related to recessive, pediatric-onset syndromes that may have DCM as a symptom.	6	<p>ACADVL ALMS1 CPT2 DNAJC19 SDHA TMEM70</p>	

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Description	# of Genes	Genes	ICD-10 Suggestions*
CARDIOMYOPATHIES, CONTINUED			
Hypertrophic Cardiomyopathy			
Includes the genes most commonly involved in hypertrophic cardiomyopathy (HCM). Includes genes that cause primary HCM as well as syndromic conditions with HCM as a feature, including Danon disease, Fabry disease, and Pompe disease.	27	ACTC1 ACTN2 AGL BAG3 CACNA1C CAV3 CSRP3 DES FHL1 FLNC GAA GLA LAMP2 LDB3 MYBPC3 MYH7 MYL2 MYL3 PLN PRKAG2 TCAP TNNC1 TNNI3 TNNT2 TPM1 TTR VCL	I42.1 Hypertrophic obstructive cardiomyopathy I42.2 Hypertrophic non-obstructive cardiomyopathy I42.5 Cardiomyopathy, other restrictive I43 Syndromic cardiomyopathy Z82.41 Family history of sudden cardiac death Z84.81 Family history of carrier of genetic disease Z86.74 Personal history of sudden cardiac arrest
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to HCM.	11	ANKRD1 CALR3 GATA4 JPH2 MYH6 MYLK2 MYOM1 MYOZ2 MYPN NEXN PDLIM3	
RASopathies			
Add-on RASopathy genes, a related group of disorders, including Noonan syndrome and cardiofaciocutaneous syndrome, that often include HCM as a feature.	18	A2ML1 BRAF CBL HRAS KRAS MAP2K1 MAP2K2 NF1 NRAS PTPN11 RAF1 RASA1 RIT1 RRAS SHOC2 SOS1 SOS2 SPRED1	
Recessive Pediatric Syndromes			
Add-on genes related to recessive, pediatric-onset syndromes that may have hypertrophic cardiomyopathy as a feature.	4	ACADVL CPT2 ELAC2 MTO1	
Left Ventricular Noncompaction			
Includes genes most commonly involved in left ventricular noncompaction (LVNC).	17	ACTC1 ACTN2 DSP HCN4 LAMP2 LDB3 LMNA MYBPC3 MYH7 PLN RYR2 SCN5A TAZ TNNI3 TNNT2 TPM1 VCL	Q23.8 Brugada syndrome R55 Syncope and collapse R94.31 Abnormal electrocardiogram (ECG or EKG) Z82.41 Family history of sudden cardiac death Z84.81 Family history of carrier of genetic disease Z86.74 Personal history of sudden cardiac arrest
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to LVNC.	2	DTNA PLEKHM2	
Restrictive Cardiomyopathy			
Includes genes most commonly involved with restrictive cardiomyopathy.	8	ACTC1 BAG3 DES MYBPC3 MYH7 TNNI3 TNNT2 TTR	I46.2 Cardiac arrest I49.9 Catecholaminergic polymorphic ventricular tachycardia (CPVT) R55 Syncope and collapse R94.31 Abnormal electrocardiogram (ECG or EKG) Z82.41 Family history of sudden cardiac death Z84.81 Family history of carrier of genetic disease Z86.74 Personal history of sudden cardiac arrest
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to Brugada syndrome.	2	FLNC MYPN	

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Description	# of Genes	Genes	ICD-10 Suggestions*
ARRHYTHMIAS AND CARDIOMYOPATHIES			
Pan Arrhythmia and Cardiomyopathy			
Includes the genes most commonly associated with heritable arrhythmias and cardiomyopathies, including long QT syndrome, short QT syndrome, arrhythmogenic cardiomyopathy, catecholaminergic polymorphic ventricular tachycardia, Brugada syndrome, hypertrophic cardiomyopathy, dilated cardiomyopathy, left ventricular noncompaction, and restrictive cardiomyopathy.	70	ABC9 ACT1 ACTN2 AGL ANK2 BAG3 CACNA1C CACNA2D1 CACNB2 CALM1 CALM2 CALM3 CASQ2 CAV3 CRYAB CSRP3 DES DMD DOLK DSC2 DSG2 DSP EMD EYA4 FHL1 FKRP FKTN FLNC GAA GLA GPD1L HCN4 JUP KCNE1 KCNE2 KCNE3 KCNH2 KCNJ2 KCNQ1 LAMP2 LDB3 LMNA MYBPC3 MYH7 MYL2 MYL3 PKP2 PLN PRKAG2 RAF1 RBM20 RYR2 SCN10A SCN4B SCN5A SGCD SLC22A5 SNTA1 TAZ TCAP TGFB3 TMEM43 TNNC1 TNNI3 TNNT2 TPM1 TRDN TTN TTR VCL	I42.0 Dilated cardiomyopathy I42.1 Hypertrophic obstructive cardiomyopathy I42.2 Hypertrophic non-obstructive cardiomyopathy I42.5 Cardiomyopathy, other restrictive I42.8 Other cardiomyopathies I42.9 Cardiomyopathy, unspecified I43 Syndromic cardiomyopathy I45.81 Long QT syndrome I49.9 Catecholaminergic polymorphic ventricular tachycardia (CPVT) I49.9 Short QT syndrome Q23.8 Brugada syndrome R55 Syncope and collapse R94.31 Abnormal electrocardiogram (ECG or EKG) Z82.41 Family history of sudden cardiac death Z84.81 Family history of carrier of genetic disease Z86.74 Personal history of sudden cardiac arrest
Emerging Evidence Genes			
Add-on genes for which there is emerging evidence that mutations in these genes are related to heritable arrhythmias.	38	AKAP9 ANKRD1 CALR3 CHRM2 CTF1 CTNNA3 DTNA FHL2 GATA4 GATA6 GATAD1 ILK JPH2 KCND3 KCNE5 KCNJ8 LAMA4 LRRC10 MYH6 MYLK2 MYO1 MYOZ2 MYPN NEBL NEXN NKX2-5 NPPA PDLIM3 PLEKHM2 RANGRF SCN1B SCN2B SCN3B SLMAP TMPO TRPM4 TXNRD2	
RASopathies			
Add-on RASopathy genes for a related group of disorders, including Noonan syndrome and cardiofaciocutaneous syndrome, that often include HCM as a symptom.	17	A2ML1 BRAF CBL HRAS KRAS MAP2K1 MAP2K2 NF1 NRAS PTPN11 RASA1 RIT1 RRAS SHOC2 SOS1 SOS2 SPRED1	
Recessive Pediatric Syndromes			
Add-on genes related to recessive, pediatric-onset syndromes that may have cardiomyopathy as a symptom.	8	ACADVL ALMS1 CPT2 DNAJC19 ELAC2 MTO1 SDHA TMEM70	

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