



Listado de genes del Cardio Panel-Code

Disease	MedGen Category	Genes	ClinVar SNPs
Aortic valve disorder	Aortic valve disorder#C1260873	COL5A1, NOTCH1	4
Loeys-Dietz syndrome	Loeys-Dietz syndrome#C2697932	COL3A1, COL5A2, MYH11, MYLK, SMAD3, TGFB2, TGFBRI, TGFBRI2	60
Short QT Syndrome	Short QT syndrome# C2348199	CACNA1C, KCNH2, KCNJ2, KCNQ1, TRPM4	11
Catecholaminergic polymorphic ventricular tachycardia	Catecholaminergic polymorphic ventricular tachycardia#C1631597	ANK2, CALM1, DSG2, LAMA4, LMNA, MYBPC3, RYR2, SCN1B, SCN5A	32
	Catecholaminergic polymorphic ventricular tachycardia ? #CN231060	RYR2	2
Familial hypercholesterolemias	Familial hypercholesterolemias#CN118841	APOB, LDLR, LDLRAP1, PCSK9	79
Familial restrictive cardiomyopathy	Familial restrictive cardiomyopathy#C0340429	TNNI3, TNNT2	9
	Cardiomyopathy, restrictive#C0007196	CACNA1C, DSP, MYH7, TNNI3	7
Noncompaction cardiomyopathy	Left ventricular noncompaction cardiomyopathy#CN167392	DES, DSP, JPH2, JUP, LDB3, MYH7, RBM20, RYR2, SCN5A, TTN	16
	Left ventricular noncompaction 10#C3715165	MYBPC3	7
	Left ventricular noncompaction 5#C3150690	MYH7	4
	Left ventricular noncompaction 9#CN179850	TPM1	2
	Left ventricular noncompaction 9#CN179851	PRDM16	2
	Left ventricular noncompaction 9#CN179852	MIB1	2
	Left ventricular noncompaction 9#CN179853	LDB3	2
Rasopathy	Left ventricular noncompaction 9#CN179854	BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, SHOC2, SOS1, TNNT2	736
	Left ventricular noncompaction 9#CN179855	MAP2K1, MAP2K2	5
	Left ventricular noncompaction 9#CN179856	CBL	9
Arrhythmogenic right ventricular cardiomyopathy	Left ventricular noncompaction 9#CN179857	DSC2, DSP, PKP2	8
Brugada syndrome	Left ventricular noncompaction 9#CN179858	ANK2, CACNA1C, CACNB2, GPD1L, HCN4, KCNE3, MYBPC3, SCN1B, SCN3B, SCN5A, SNTA1	271
	Left ventricular noncompaction 9#CN179859	SCN5A	2
	Left ventricular noncompaction 9#CN179860	KCND3	2
	Left ventricular noncompaction 9#CN179861	CACNA1C	2
Long QT Syndrome	Left ventricular noncompaction 9#CN179862	ANK2, CACNA1C, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, KCNQ1, SCN5A	755
	Left ventricular noncompaction 9#CN179863	KCNQ1	201
	Left ventricular noncompaction 9#CN179864	AKAP9, ANK2, CACNA1C, CACNA2D1, CAV3, DSC2, DSP, GPD1L, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, MYBPC3, RYR2, SCN1B, SCN5A, SNTA1, TMEM43, TRPM4	110
	Left ventricular noncompaction 9#CN179865	KCNE2, KCNH2, SCN5A	5
	Left ventricular noncompaction 9#CN179866	KCNH2, KCNQ1	4
	Left ventricular noncompaction 9#CN179867	CALM1	3
	Left ventricular noncompaction 9#CN179868	ANK2	2
	Left ventricular noncompaction 9#CN179869	KCNH2, SSUH2	2
	Left ventricular noncompaction 9#CN179870	KCNE1, KCNH2	2
Atrial fibrillation	Left ventricular noncompaction 9#CN179871	KCNH2, SCN5A	2
	Left ventricular noncompaction 9#CN179872	ABCC9, GJA5, KCNA5, KCNE2, KCNJ2, KCNQ1, SCN5A	42
	Left ventricular noncompaction 9#CN179873	SCN5A	7
	Left ventricular noncompaction 9#CN179874	SCN3B	3
	Left ventricular noncompaction 9#CN179875	SCN1B	3
	Left ventricular noncompaction 9#CN179876	SCN2B	2
Hypertrophic cardiomyopathy	Left ventricular noncompaction 9#CN179877	NPPA	1
	Left ventricular noncompaction 9#CN179878	JPH2, MYBPC3, MYH7, MYL3, MYOZ2, TNNI3, TPM1	24
	Left ventricular noncompaction 9#CN179879	ACTN2	5
	Left ventricular noncompaction 9#CN179880	LDB3	2
Dilated cardiomyopathy	Left ventricular noncompaction 9#CN179881	MYPN	2
	Left ventricular noncompaction 9#CN179882	ACTN2, ANKRD1, CEP85L, CSRP3, DES, DSG2, AS1, LDB3, MYH6, MYH7, MYPN, SCN5A, TMPO, TTN, VCL	23
	Left ventricular noncompaction 9#CN179883	MYPN	4
Cardiomyopathy	Left ventricular noncompaction 9#CN179884	PRDM16	3
	Left ventricular noncompaction 9#CN179885	ABCC9, ACTC1, ACTN2, ALMS1, ANKRD1, BAG3, CALR3, CAV3, PLN, CRYAB, CSRP3, CTF1, DES, DSC2, DSG2, DSP, DTNA, EYA4, FHL2, FKTN, GATAD1, GPD1L, ILK, JPH2, JUP, KCNE2, KCNH2, KRAS, LAMA4, LDB3, LMNA, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, PDLIM3, PKP2, PRKAG2, RBM20, RYR2, SCN5A, SDHA, SGCD, SNTA1, SOS1, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, TXNRD2, VCL	3017
Arrhythmia	Left ventricular noncompaction 9#CN179886	ANK2	5
	Left ventricular noncompaction 9#CN179887	AKAP9, ANK2, CACNA1C, CACNB2, CASQ2, DSG2, HCN4, JUP, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, RANGRF, RYR2, SCN1B, SCN3B, SCN4B, SCN5A, SNTA1	967
Familial aortopathy (Congenital aneurysm of ascending aorta, CROGLoeys-Dietz syndrome, type 4, CROGMarfan's syndrome) Thoracic aortic aneurysms and aortic dissections	Left ventricular noncompaction 9#CN179888	COL3A1, COL5A2, FBN1, LTBP2, MYH11, TGFBRI, TGFBRI2	551
	Left ventricular noncompaction 9#CN179889	ACTA2, COL3A1, COL5A1, COL5A2, FBN1, FBN2, MYH11, MYLK, NOTCH1, TGFB2, TGFBRI, TGFBRI2	579
	Left ventricular noncompaction 9#CN179890	MYH11	1
Osler hemorrhagic telangiectasia syndrome	Left ventricular noncompaction 9#CN179891	SMAD4	20
Sudden cardiac arrest	Left ventricular noncompaction 9#CN179892	GPD1L, KCNH2, KCNQ1, SCN4B, SCN5A	19
	Left ventricular noncompaction 9#CN179893	AKAP9, ANK2, PLN, DSG2, GPD1L, KCNH2, RBM20, RYR2, SCN5A, TPM1	12
	Left ventricular noncompaction 9#CN179894	AKAP9, ANK2, CACNA2D1, CACNB2, PLN, DSC2, DSP, MYBPC3, MYH6, RYR2, SCN5A	13
Holt-Oram syndrome	Left ventricular noncompaction 9#CN179895	TBX5	7
Arteriohepatic dysplasia (Alagille syndrome)	Left ventricular noncompaction 9#CN179896	JAG1	7