INVITAE CARDIOLOGY GENETIC TESTS

A comprehensive menu for heritable heart and vascular conditions curated by medical genetics experts for specific indications and clinical scenarios.

CLINICAL AREA: CARDIOLOGY

Test name	# gene(s)	Gene list
Arrhythmia and Cardiomyopathy		
Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel	67	ABCC9, ACTC1, ACTN2, AGL, ANK2, BAG3, CACNA1C, 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, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, LAMP2, LMNA, MYBPC3, MYH7, MYL2, MYL3, MYL4, NKX2-5, PKP2, PLN, PRKAG2, RAF1, RBM20, RYR2, SCN5A, SGCD, SLC22A5, TAZ, TCAP, TGFB3, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TTN, TTR, VCL
Add-on preliminary-evidence genes	47	AKAP9, ANKRD1, CACNA2D1, CALR3, CHRM2, CTF1, CTNNA3, DTNA, FHL2, GATA4, GATA6, GATAD1, GJA5, ILK, JPH2, KCND3, KCNE3, KCNE5, KCNJ5, KCNJ8, KCNK3, LAMA4, LDB3, LRRC10, MED12, MYH6, MYLK2, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NPPA, PDLIM3, PLEKHM2, PRDM16, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SLMAP, SNTA1, TMPO, TRPM4, TXNRD2
Add-on RASopathy genes not included in panel	17	A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1
Add-on genes associated with autosomal recessive syndromic pediatric cardiomyopathy	8	ACADVL, ALMS1, CPT2, DNAJC19, ELAC2, MTO1, SDHA, TMEM70
Add-on sudden unexpected death in epilepsy (SUDEP) genes for arrhythmia and cardiomyopathy	11	DEPDC5, KCNA1, KCNQ2, KCNQ3, KCNT1, PCDH19, PRRT2, SCN1A, SCN8A, SCN9A, SLC2A1
Arrhythmia		
Invitae Arrhythmia Comprehensive Panel	39	ABCC9, ACTN2, ANK2, CACNA1C, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, DES, DSC2, DSG2, DSP, EMD, FLNC, GPD1L, HCN4, JUP, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, LMNA, MYL4, NKX2-5, PKP2, PLN, PRKAG2, RBM20, RYR2, SCN5A, TMEM43, TNNI3, TNNT2, TRDN, TTN
Add-on preliminary-evidence genes	25	AKAP9, ANKRD1, CACNA2D1, CTNNA3, GATA6, GJA5, KCND3, KCNE3, KCNE5, KCNJ5, KCNJ8, KCNK3, LDB3, NPPA, PDLIM3, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SLMAP, SNTA1, TGFB3, TRPM4
Add-on sudden unexpected death in epilepsy (SUDEP) genes for arrhythmia	11	DEPDC5, KCNA1, KCNQ2, KCNQ3, KCNT1, PCDH19, PRRT2, SCN1A, SCN8A, SCN9A, SLC2A1
Invitae Arrhythmogenic Cardiomyopathy Panel	19	ACTN2, DES, DSC2, DSG2, DSP, EMD, FLNC, JUP, LMNA, PKP2, PLN, PRKAG2, RBM20, RYR2, SCN5A,TMEM43, TNNI3, TNNT2, TTN
Add-on preliminary-evidence genes	5	ANKRD1, CTNNA3, LDB3, PDLIM3, TGFB3
Invitae Brugada Syndrome Test	1	SCN5A
Add-on preliminary-evidence genes	19	ABCC9, CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNH2, KCNE3, KCNE5, KCNJ8, PKP2, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SLMAP, TRPM4
Invitae Catecholaminergic Polymorphic Ventricular Tachycardia Panel	8	ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TRDN
Invitae Long QT Syndrome Panel	13	ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN5A, TRDN
Add-on preliminary-evidence genes	4	AKAP9, KCNJ5, SCN4B, SNTA1
Invitae Short QT Syndrome Panel	5	CACNA1C, CACNB2, KCNH2, KCNJ2, KCNQ1
Add-on preliminary-evidence gene	1	CACNA2D1

INVITAE CARDIOLOGY GENETIC TESTS

CLINICAL AREA: CARDIOLOGY

Test name	# gene(s)	Gene list
Cardiomyopathy		
Invitae Cardiomyopathy Comprehensive Panel	50	ABCC9, ACTC1, ACTN2, AGL, BAG3, CACNA1C, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GAA, GLA, HCN4, JUP, LAMP2, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, PRKAG2, RAF1, RBM20, RYR2, SCN5A, SGCD, SLC22A5, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL
Add-on preliminary-evidence genes	31	ANKRD1, CALR3, CHRM2, CTF1, CTNNA3, DTNA, FHL2, GATA4, GATA6, GATAD1, ILK, JPH2, LAMA4, LDB3, LRRC10, MED12, MYH6, MYLK2, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PLEKHM2, PRDM16, TGFB3, TMPO, TXNRD2
Add-on RASopathy genes not included in panel	17	A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1
Add-on autosomal recessive syndromic pediatric cardiomyopathy genes	8	ACADVL, ALMS1, CPT2, DNAJC19, ELAC2, MTO1, SDHA, TMEM70
Invitae Arrhythmogenic Cardiomyopathy Panel	19	ACTN2, DES, DSC2, DSG2, DSP, EMD, JUP, LMNA, PKP2, PLN, PRKAG2, RBM20, RYR2, SCN5A, TGFB3, TMEM43, TNNI3, TNNT2, TTN
Add-on preliminary-evidence genes	4	ANKRD1, CTNNA3, LDB3, PDLIM3
Invitae Dilated Cardiomyopathy Panel	41	ABCC9, ACTC1, ACTN2, BAG3, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FKRP, FKTN, FLNC, JUP, LAMP2, LMNA, MYBPC3, MYH7, PKP2, PLN, RAF1, RBM20, RYR2, SCN5A, SGCD, SLC22A5, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL
Add-on preliminary-evidence genes	23	ANKRD1, CHRM2, CTF1, FHL2, GATA4, GATA6, GATAD1, ILK, LAMA4, LDB3, LRRC10, MED12, MYH6, MYPN, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PLEKHM2, PRDM16, TMPO, TXNRD2
Add-on autosomal recessive syndromic pediatric cardiomyopathy genes	6	ACADVL, ALMS1, CPT2, DNAJC19, SDHA, TMEM70
Invitae Hypertrophic Cardiomyopathy Panel	26	ACTC1, ACTN2, AGL, BAG3, CACNA1C, CAV3, CSRP3, DES, FHL1, FLNC, GAA, GLA, LAMP2, MYBPC3, MYH7, MYL2, MYL3, PLN, PRKAG2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL
Add-on preliminary-evidence genes	12	ANKRD1, CALR3, GATA4, JPH2, LDB3, MYH6, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, PDLIM3
Add-on RASopathy genes	18	A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1
Add-on genes associated with autosomal recessive syndromic pediatric cardiomyopathy	4	ACADVL, CPT2, ELAC2, MTO1
Invitae Left Ventricular Noncompaction Panel	15	ACTC1, DSP, HCN4 , LAMP2, LMNA, MYBPC3, MYH7, PLN, RYR2, SCN5A, TAZ, TNNI3, TNNT2, TPM1, VCL
Add-on preliminary-evidence genes	4	DTNA, LDB3, PLEKHM2, PRDM16
Invitae Transthyretin Amyloidosis Test	1	TTR
Invitae Hereditary Hemochromatosis Panel	5	HAMP, HFE, HJV, SLC40A1, TFR2
Invitae RASopathies Comprehensive Panel	18	A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1

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Cardiomyopathy and Skeletal Muscle Disease		
Invitae Cardiomyopathy and Skeletal Muscle Disease Panel	116	ABCC9, ACTA1, ACTC1, ACTN2, AGL, ANO5, ATP2A1, B3GALNT2, B4GAT1, BAG3, BIN1, CACNA1C, CAPN3, CAV3, CCDC78, CFL2, CHKB, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, CSRP3, DAG1, DES, DMD, DNAJB6, DNM2, DOLK, DPM1, DPM2, DPM3, DSC2, DSG2, DSP, DYSF, EMD, EYA4, FHL1, FKBP14, FKRP, FKTN, FLNC, GAA, GLA, GMPPB, GNE, GYS1, HCN4, ISPD, ITGA7, JUP, KBTBD13, KLHL40, KLHL41, LAMA2, LAMP2, LARGE1, LMNA, LMOD3, MATR3, MEGF10, MTM1, MYBPC3, MYH7, MYL2, MYL3, MYOT, MYPN, NEB, PKP2, PLEC, PLN, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRKAG2, RAF1, RBM20, RXYLT1, RYR1, RYR2, SCN5A, SELENON, SGCA, SGCB, SGCD, SGCG, SLC22A5, SQSTM1, STAC3, STIM1, TAZ, TCAP, TIA1, TMEM43, TNNC1, TNNI3, TNNT1, TNNT2, TNPO3, TOR1AIP1, TPM1, TPM2, TPM3, TRAPPC11, TRIM32, TTN, TTR, VCL, VCP
Add-on preliminary-evidence genes	36	ANKRD1, CALR3, CHRM2, CTF1, CTNNA3, DTNA, FHL2, GATA4, GATA6, GATAD1, HNRNPDL, ILK, JPH2, LAMA4, LDB3, LIMS2, LRRC10, MYF6, MYH6, MYLK2, MYOM1, MYOZ2, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PLEKHM2, PRDM16, SUN1, SUN2, SYNE1, SYNE2, TGFB3, TMPO, TXNRD2
Add-on autosomal recessive syndromic pediatric cardiomyopathy genes	7	ACADVL, ALMS1, DNAJC19, ELAC2, MTO1, SDHA, TMEM70
Aortopathy and Connective Tissue Disorders		
Invitae Aortopathy Comprehensive Panel	24	ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, FBN1, FBN2, FLNA, FOXE3, MED12, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2
Add-on preliminary-evidence genes	3	HCN4, MAT2A, SMAD6
Invitae Ehlers-Danlos Syndrome Panel	15	ADAMTS2, ATP7A, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, CRTAP, FLNA, FKBP14, P3H1, PLOD1, SLC39A13
Invitae Loeys-Dietz Syndrome Panel	6	FBN1, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2
Invitae Marfan Syndrome Test	1	FBN1
Familial Hypercholesterolemia		
Invitae Familial Hypercholesterolemia Panel	4	APOB, LDLR, LDLRAP1, PCSK9
Add-on genes for clinical overlapping conditions	2	ABCG5, ABCG8
Pulmonary Hypertension		
Invitae Pulmonary Arterial Hypertension Panel	5	ACVRL1, BMPR2, CAV1, ENG, SMAD9
Add-on preliminary-evidence genes	4	BMPR1B, GDF2, KCNA5, KCNK3
Invitae Hereditary Hemorrhagic Telangiectasia Panel	5	ACVRL1, ENG, GDF2, RASA1, SMAD4
Invitae Capillary Malformation-Arteriovenous Malformation Syndrome Test	1	RASA1
Add-on hereditary hemorrhagic telangiectasia genes	3	ACVRL1, ENG, SMAD4
Congenital Heart Disease		
Invitae Congenital Heart Disease Panel	42	ACTC1, ACVR2B, ALMS1, BCOR, BRAF, CBL, CHD7, CRELD1, ELN, FOXH1, GATA4, GATA6, GDF1, GJA1, GPC3, HAND1, HRAS, JAG1, KRAS, LEFTY2, MAP2K1, MAP2K2, MED13L, MEIS2, MYH6, NKX2-5, NKX2-6, NODAL, NOTCH1, NR2F2, NRAS, NSD1, PTPN11, RAF1, RIT1, SHOC2, SMAD6, SOS1, TBX1, TBX5, ZFPM2, ZIC3
Invitae CHARGE Syndrome Test	1	CHD7
Invitae Holt-Oram Syndrome Test	1	TBX5
Invitae RASopathies Comprehensive Panel	18	A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1
Invitae Sotos Syndrome Test	1	NSD1

