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

ARRHYTHMIA AND CARDIOMYOPATHY

Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel (up to 148 genes)

Primary panel (67 genes)

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 (46 genes)

AKAP9	ANKRD1	CACNA2D1	CALR3	CHRM2	CTF1	CTNNA3	DTNA	FHL2	GATA4	GATA6	GATAD1
GJA5	ILK	JPH2	KCND3	KCNE3	KCNE5	KCNJ5	KCNJ8	KCNK3	LAMA4	LDB3	LRRC10
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 genes)

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 genes)

ACADVL	ALMS1	CPT2	DNAJC19	ELAC2	MTO1	SDHA	TMEM70

Add-on sudden unexpected death in epilepsy (SUDEP) genes for arrhythmia and cardiomyopathy (10 genes)

DEPDC5	KCNQ2	KCNQ3	KCNT1	PCDH19	PRRT2	SCN1A	SCN8A	SCN9A	SLC2A1

ARRHYTHMIA

Invitae Arrhythmia Comprehensive Panel (up to 73 genes)

Primary panel (39 genes)

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 (24 genes)

AKAP9	ANKRD1	CACNA2D1	CTNNA3	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 (10 genes)

DEPDC5	KCNQ2	KCNQ3	KCNT1	PCDH19	PRRT2	SCN1A	SCN8A	SCN9A	SLC2A1

Invitae Arrhythmogenic Cardiomyopathy Panel (up to 24 genes)

Primary panel (19 genes)

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 genes)

ANKRD1	CTNNA3	LDB3	PDLIM3	TGFB3

Invitae Brugada Syndrome Panel (up to 20 genes)

Primary panel (8 genes)

ABCC9	CACNA1C	CACNB2	GPD1L	HCN4	KCNH2	PKP2	SCN5A

Add-on preliminary-evidence genes (12 genes)

CACNA2D1	KCND3	KCNE3	KCNE5	KCNJ8	RANGRF	SCN10A	SCN1B	SCN2B	SCN3B	SLMAP	TRPM4

Invitae Catecholaminergic Polymorphic Ventricular Tachycardia Panel (8 genes)

Primary panel (8 genes)

ANK2	CALM1	CALM2	CALM3	CASQ2	KCNJ2	RYR2	TRDN

Invitae Long QT Syndrome Panel (up to 17 genes)

Primary panel (13 genes)

ANK2	CACNA1C	CALM1	CALM2	CALM3	CAV3	KCNE1	KCNE2	KCNH2	KCNJ2	KCNQ1	SCN5A
TRDN											

Add-on preliminary-evidence genes (4 genes)

AKAP9	KCNJ5	SCN4B	SNTA1

Invitae Short QT Syndrome Panel (up to 6 genes)

Primary panel (5 genes)

CACNA1C	CACNB2	KCNH2	KCNJ2	KCNQ1

Add-on preliminary-evidence gene (1 gene)

CACNA2D1

INVITAE CARDIOLOGY GENETIC TESTS (continued)

CARDIOMYOPATHY

Invitae Cardiomyopathy Comprehensive Panel (up to 105 genes)

Primary panel (50 genes)

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 (30 genes)

ANKRD1 CALR3 CHRM2 CTF1 CTNNA3 DTNA FHL2 GATA4 GATA6 GATAD1 ILK JPH2
LAMA4 LDB3 LRRC10 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 genes)

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 genes)

ACADVL ALMS1 CPT2 DNAJC19 ELAC2 MTO1 SDHA TMEM70

Invitae Arrhythmogenic Cardiomyopathy Panel (up to 23 genes)

Primary panel (19 genes)

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 genes)

ANKRD1 CTNNA3 LDB3 PDLIM3

Invitae Dilated Cardiomyopathy Panel (up to 69 genes)

Primary panel (41 genes)

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 (22 genes)

ANKRD1 CHRM2 CTF1 FHL2 GATA4 GATA6 GATAD1 ILK LAMA4 LDB3 LRRC10 MYH6
MYPN NEBL NEXN NKX2-5 NPPA PDLIM3 PLEKHM2 PRDM16 TMPO TXNRD2

Add-on genes associated with autosomal recessive syndromic pediatric cardiomyopathy (6 genes)

ACADVL ALMS1 CPT2 DNAJC19 SDHA TMEM70

Invitae Hypertrophic Cardiomyopathy Panel (up to 58 genes)

Primary panel (26 genes)

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 genes)

ANKRD1 CALR3 GATA4 JPH2 LDB3 MYH6 MYLK2 MYOM1 MYOZ2 MYPN NEXN PDLIM3

Add-on RASopathy genes (18 genes)

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 genes)

ACADVL CPT2 ELAC2 MTO1

Invitae Left Ventricular Noncompaction Panel (up to 19 genes)

Primary panel (15 genes)

ACTC1 DSP HCN4 LAMP2 LMNA MYBPC3 MYH7 PLN RYR2 SCN5A TAZ TNNI3
TNNT2 TPM1 VCL

Add-on preliminary-evidence genes (4 genes)

DTNA LDB3 PLEKHM2 PRDM16

Invitae Transthyretin Amyloidosis Test (1 gene)

Primary panel (1 gene)

TTR

Invitae Hereditary Hemochromatosis Panel (5 genes)

Primary panel (5 genes)

HAMP HFE HFE2 SLC40A1 TFR2

Invitae RASopathies Comprehensive Panel (18 genes)

Primary panel (18 genes)

A2ML1 BRAF CBL HRAS KRAS MAP2K1 MAP2K2 NF1 NRAS PTPN11 RAF1 RASA1
RIT1 RRAS SHOC2 SOS1 SOS2 SPRED1

INVITAE CARDIOLOGY GENETIC TESTS (continued)

CARDIOMYOPATHY AND SKELETAL MUSCLE DISEASE

Invitae Cardiomyopathy and Skeletal Muscle Disease Panel (up to 158 genes)

Primary panel (113 genes)

ABCC9	ACTA1	ACTC1	ACTN2	AGL	ANO5	ATP2A1	B3GALNT2	B4GAT1	BAG3	BIN1	CACNA1C
CAPN3	CAV3	CCDC78	CFL2	CHKB	CNTN1	COL6A1	COL6A2	COL6A3	CPT2	CRYAB	CSRP3
DAG1	DAG1	DES	DMD	DNAJB6	DNM2	DOLK	DPM1	DPM2	DPM3	DSC2	DSG2
DSP	DYSF	EMD	EYA4	FHL1	FKBP14	FKRP	FKTN	FLNC	GAA	GLA	GMPPB
GNE	HCN4	ISPD	ITGA7	JUP	KBTBD13	KLHL40	KLHL41	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	RYR1	RYR2
SCN5A	SELENON	SGCA	SGCB	SGCD	SGCG	SLC22A5	SQSTM1	STAC3	STIM1	TAZ	TCAP
TIA1	TMEM43	TMEM5	TNNC1	TNNI3	TNNT1	TNNT2	TNPO3	TPM1	TPM2	TPM3	TRAPPC11
TRIM32	TTN	TTR	VCL	VCP							

Add-on preliminary-evidence genes (38 genes)

ANKRD1	CALR3	CHRM2	COL12A1	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
TOR1AIP1	TXNRD2										

Add-on autosomal recessive syndromic pediatric cardiomyopathy genes (7 genes)

ACADVL	ALMS1	DNAJC19	ELAC2	MTO1	SDHA	TMEM70

AORTOPATHY AND CONNECTIVE TISSUE DISORDERS

Invitae Aortopathy Comprehensive Panel (up to 25 genes)

Primary panel (23 genes)

ACTA2	CBS	COL3A1	COL5A1	COL5A2	EFEMP2	FBN1	FBN2	FLNA	MED12	MYH11	MYLK
NOTCH1	PLOD1	PRKG1	SKI	SLC2A10	SMAD3	SMAD4	TGFB2	TGFB3	TGFBR1	TGFBR2	

Add-on preliminary-evidence genes (2 genes)

MAT2A	SMAD6

Invitae Ehlers-Danlos Syndrome Panel (14 genes)

Primary panel (14 genes)

ADAMTS2	ATP7A	CHST14	COL1A1	COL1A2	COL3A1	COL5A1	COL5A2	CRTAP	FLNA	FKBP14	P3H1
PLOD1	SLC39A13										

Invitae Loeys-Dietz Syndrome Panel (up to 6 genes)

Primary panel (4 genes)

SMAD3	TGFB2	TGFBR1	TGFBR2

Add-on clinically overlapping genes (2 genes)

FBN1	TGFB3

Invitae Marfan Syndrome Test (1 gene)

Primary panel (1 gene)

FBN1

FAMILIAL HYPERCHOLESTEROLEMIA

Invitae Familial Hypercholesterolemia Panel (4 genes)

Primary panel (4 genes)

APOB	LDLR	LDLRAP1	PCSK9

PULMONARY HYPERTENSION

Invitae Pulmonary Arterial Hypertension Panel (up to 9 genes)

Primary panel (4 genes)

ACVRL1	BMPR2	CAV1	ENG

Add-on preliminary-evidence genes (5 genes)

BMPR1B	GDF2	KCNA5	KCNK3	SMAD9

Invitae Hereditary Hemorrhagic Telangiectasia Panel (up to 5 genes)

Primary panel (4 genes)

ACVRL1	ENG	RASA1	SMAD4

Add-on preliminary-evidence gene (1 gene)

GDF2

Invitae Capillary Malformation-Arteriovenous Malformation Syndrome Test (up to 4 genes)

Primary panel (1 gene)

RASA1

Add-on hereditary hemorrhagic telangiectasia genes (3 genes)

ACVRL1	ENG	SMAD4

CONGENITAL HEART DISEASE

Invitae Congenital Heart Disease Panel (42 genes)

Primary panel (42 genes)

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 gene)

Primary panel (1 gene)

CHD7

Invitae Holt-Oram Syndrome Test (1 gene)

Primary panel (1 gene)

TBX5

Invitae RASopathies Comprehensive Panel (18 genes)

Primary panel (18 genes)

A2ML1	BRAF	CBL	HRAS	KRAS	MAP2K1	MAP2K2	NF1	NRAS	PTPN11	RAF1	RASA1
RIT1	RRAS	SHOC2	SOS1	SOS2	SPRED1						

Invitae Sotos Syndrome Test (1 gene)

Primary panel (1 gene)

NSD1

ABOUT INVITAE

Invitae's mission is to bring comprehensive genetic information into mainstream medical practice to improve the quality of healthcare for everyone. Our goal is to aggregate most of the world's genetic tests into a single service with higher quality, faster turnaround time, and lower price than many single-gene tests today.

For more information about Invitae's genetic tests, please visit www.invitae.com.

INVITAE

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