



CARDIOVASCULAR PANELS

WHAT IS THE ROLE OF CARDIOVASCULAR GENETIC TESTING?



IDENTIFY

- ◆ Genetic mutations involved in cardiovascular conditions
- ◆ Complex cardiovascular diseases
- ◆ The correct disease by differentiating between diseases with similar phenotype



EVALUATE

- ◆ Patient's level of risk
- ◆ Treatment options
- ◆ At-risk family members



MANAGE

- ◆ Clinical care
- ◆ Decision-making before an invasive treatment
- ◆ Specific therapies and predict their response

7 INDIVIDUAL PANELS AND 1 COMPREHENSIVE PANEL FOR INHERITED CARDIOVASCULAR DISEASES

AORTOPATHY (48 genes)

<i>ABCC6</i>	<i>ATP6V1A</i>	<i>C1S</i>	<i>COL4A1</i>	<i>FBLN5</i>	<i>LOX</i>	<i>PRDM5</i>	<i>SMAD4</i>
<i>ACTA2</i>	<i>ATP6V1E1</i>	<i>CBS</i>	<i>COL5A1</i>	<i>FBN1</i>	<i>LTBP4</i>	<i>PYCR1</i>	<i>TGFB2</i>
<i>ACVR1</i>	<i>B3GALT6</i>	<i>CHST14</i>	<i>COL5A2</i>	<i>FBN2</i>	<i>MFAP5</i>	<i>SKI</i>	<i>TGFB3</i>
<i>ADAMTS2</i>	<i>B4GALT7</i>	<i>COL1A1</i>	<i>DSE</i>	<i>FKBP14</i>	<i>MYH11</i>	<i>SLC2A10</i>	<i>TGFBRI</i>
<i>ALDH18A1</i>	<i>BGN</i>	<i>COL1A2</i>	<i>EFEMP2</i>	<i>FOXE3</i>	<i>MYLK</i>	<i>SLC39A13</i>	<i>TGFBR2</i>
<i>ATP6VOA2</i>	<i>CTR</i>	<i>COL3A1</i>	<i>ELN</i>	<i>GORAB</i>	<i>PLOD1</i>	<i>SMAD3</i>	<i>ZNF469</i>

ARRHYTHMIA (42 genes)

<i>ABCC9</i>	<i>CALM1</i>	<i>GNAI2</i>	<i>KCNE1</i>	<i>KCNJ8</i>	<i>SCN10A</i>	<i>SNTA1</i>
<i>AKAP9</i>	<i>CALM2</i>	<i>GPD1L</i>	<i>KCNE2</i>	<i>KCNQ1</i>	<i>SCN1B</i>	<i>TBX5</i>
<i>ANK2</i>	<i>CALM3</i>	<i>HCN4</i>	<i>KCNE3</i>	<i>LMNA</i>	<i>SCN2B</i>	<i>TECRL</i>
<i>CACNA1C</i>	<i>CASQ2</i>	<i>KCNA5</i>	<i>KCNH2</i>	<i>MYL4</i>	<i>SCN3B</i>	<i>TNNI3</i>
<i>CACNA2D1</i>	<i>CAV3</i>	<i>KCNAB2</i>	<i>KCNJ2</i>	<i>NPPA</i>	<i>SCN4B</i>	<i>TRDN</i>
<i>CACNB2</i>	<i>GJA5</i>	<i>KCND3</i>	<i>KCNJ5</i>	<i>RYR2</i>	<i>SCN5A</i>	<i>TRPM4</i>

CARDIOMYOPATHY (98 genes)

AARS2	CAV3	DSP	HCN4	MYBPC3	PLN	TAZ
ABCC9	COA5	DTNA	HRAS	MYH6	PPA2	TCAP
ACAD9	COA6	ELAC2	ILK	MYH7	PRDM16	TGFB3
ACADVL	COX15	EMD	JPH2	MYL2	PRKAG2	TK2
ACTA1	CPT2	FHL1	JUP	MYL3	RAF1	TMEM43
ACTC1	CRYAB	FKTN	KARS	MYLK2	RBM20	TMEM70
ACTN2	CSRP3	FLNC	LAMA4	MYOZ2	RYR2	TNNC1
AGK	CTNNA3	GAA	LAMP2	MYPN	SCN5A	TNNI3
AKAP9	DES	GATAD1	LDB3	NDUFB11	SCO2	TNNT2
ALMS1	DMD	GBE1	LMNA	NDUFV2	SGCD	TPM1
ALPK3	DNAJC19	GLA	MIB1	NEBL	SLC22A5	TSFM
ANKRD1	DOLK	GTPBP3	MRPL3	NEXN	SLC25A20	TTN
BAG3	DSC2	HADHA	MRPL44	PDLIM3	SLC25A3	TTR
CASQ2	DSG2	HADHB	MTO1	PKP2	SLC25A4	VCL

CONGENITAL HEART DEFECTS (80 genes)

ACTA2	CELSR2	DNAH6	FOXC1	HAND2	MYH7	PITX2	SALL4	TGFBR1
ACTC1	CELSR3	DNAI1	FOXH1	HRAS	NIPBL	PKD1L1	SEMA3D	TGFBR2
ACVR2B	CHD4	DTNA	GANAB	JAG1	NKX2-5	PLD1	SEMA3E	TLL1
AFF4	CHD7	EHMT1	GATA4	KDM5B	NKX2-6	POGZ	SMAD6	TMEM260
BMPR2	CITED2	ELN	GATA5	KMT2D	NME7	PRDM6	TAB2	TPM1
CAD	CREBBP	EVC	GATA6	MED13L	NODAL	PRKD1	TBX1	ZEB2
CCDC11	CRELD1	EVC2	GDF1	MMP21	NOTCH1	RABGAP1L	TBX20	ZFPM2
CDK13	DNAH11	FBN1	GPC3	MYH11	NOTCH2	RBFOX2	TBX5	ZIC3
CELSR1	DNAH5	FLNA	HAND1	MYH6	NR2F2	RBM10	TFAP2B	

FAMILIAL HYPERCHOLESTEROLEMIA (11 genes)

ABCA1	APOA5	LDLR	LPL
ABCG5	APOB	LDLRAP1	PCSK9
ABCG8	APOE	LIPA	

PULMONARY HYPERTENSION (11 genes)

ACVRL1	CAV1	KCNA5	SMAD9
BMPR1B	EIF2AK4	KCNK3	TBX4
BMPR2	ENG	SMAD4	

RASOPATHIES (30 genes)

AKT3	CCND2	KRAS	MAP2K2	NF2	PIK3R2	RAF1	RIT1	SHOC2	SOS2
BRAF	EPHB4	LZTR1	MRAS	NRAS	PPP1CB	RASA1	RRAS	SMARCB1	SPRED1
CBL	HRAS	MAP2K1	NF1	PIK3CA	PTPN11	RASA2	SASH1	SOS1	STAMBP

COMPREHENSIVE PANEL (292 genes)

Includes all 292 genes