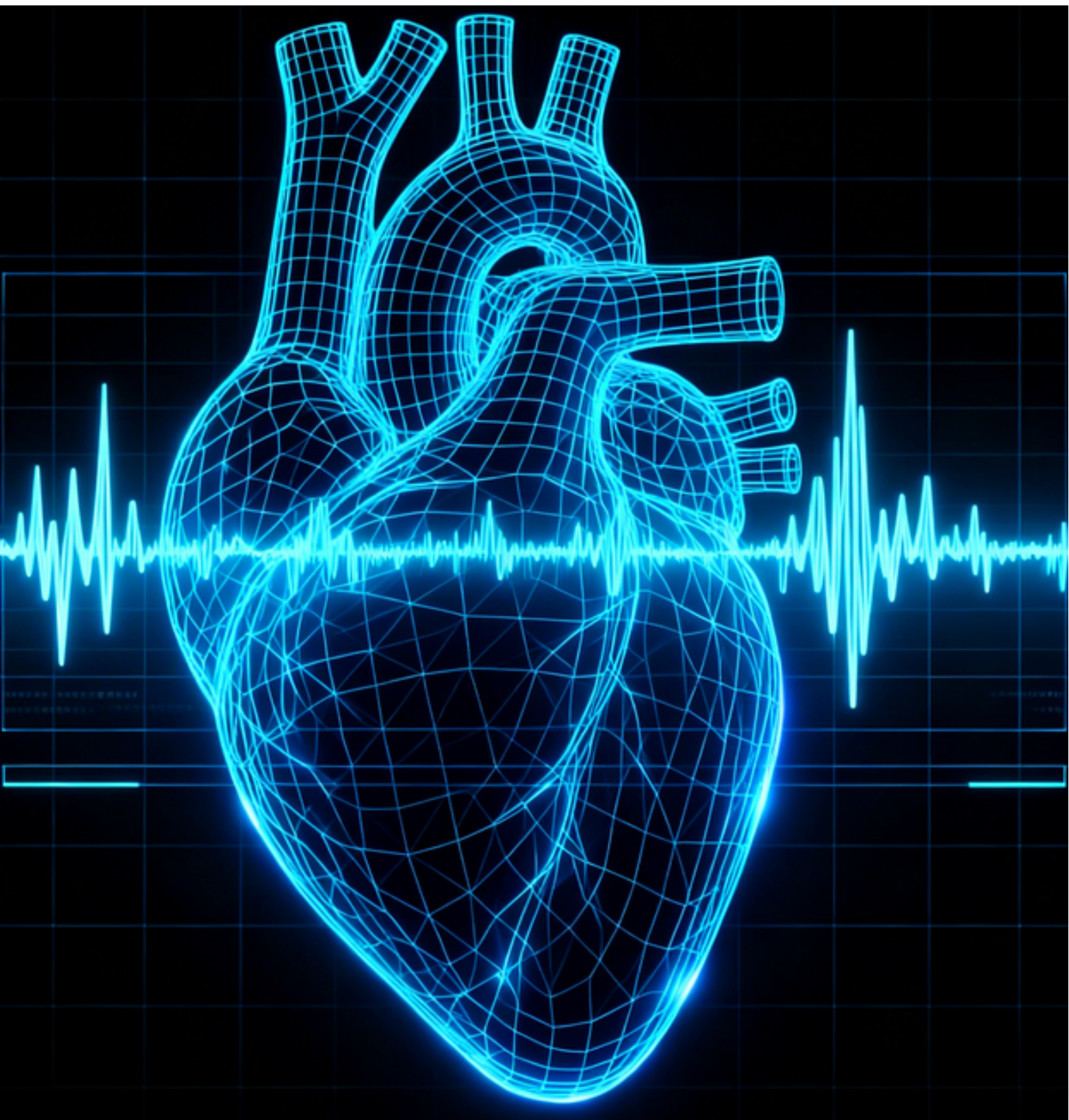


CardioRisk

A Genomic Tool for Hereditary Heart Disease — by iGenetx



What is CardioRisk?

CardioRisk is a comprehensive service for diagnosing hereditary cardiovascular disease. It combines whole exome sequencing of 100 expert-selected genes with specialist genetic counseling recommended by the ESC, AHA, and CCS for patients with cardiomyopathies and channelopathies. Scientific evidence confirms genetic testing in hereditary heart disease is cost-effective and has markedly reduced mortality.

Who is it intended for?

- Patients diagnosed with cardiomyopathy or channelopathy
- Individuals with first-degree relatives affected by hereditary heart disease
- Patients who have survived a sudden cardiac death episode
- Families in whom a pathogenic variant has been identified

Why Choose CardioRisk?

Comprehensive	Advanced Technology	Expert Interpretation	Simple
Analyzes 100 genes covering the full spectrum of hereditary heart disease in one test.	Whole Exome Sequencing with 100x average coverage — superior to standard gene panels.	Variants classified per ACMG guidelines by specialist curators using the latest evidence.	A single saliva or blood sample with pre- and post-test genetic counseling included.

Key Performance Statistics

100

Genes analyzed across all
hereditary heart disease
categories

26.5%

Diagnostic yield with WES vs.
18% with commercial panels

95%

Detection rate in HCM with WES
vs. 50% with a 5-gene panel

Technical Specifications

- Whole Exome Sequencing (WES) with 100x average coverage; >99% of target genes at $\geq 20x$
- Double-check methodology applied to all 100 genes by an expert cardiac genetics team
- Variant classification per ACMG guidelines; specialized software for detailed analysis
- Accepted samples: saliva (iGenetx kit), whole blood in EDTA, or extracted DNA

CardioRisk — Disease Coverage & Testing Process

Disease Categories, Conditions & Associated Genes (100 Genes Total)		
Primary Cardiomyopathies	Hypertrophic (HCM)	ACTC1, ACTN2, COX15, CSRP3, FHL1, FLNC, FXN, JPH2, LAMP2, LDB3, MYBPC3, MYH7, MYL2, MYL3, NEXN, NF1, PLN, PRKAG2, SLC25A4, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL
	Dilated (DCM)	ABCC9, ACTC1, ACTN2, BAG3, CRYAB, CSRP3, DES, DSG2, FKTN, LDB3, LMNA, MYBPC3, MYH7, NEXN, NF1, PLN, RBM20, SCN5A, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL
	ARVC	DES, DSC2, DSG2, DSP, JUP, LMNA, PKP2, PLN, RYR2, TGFB3, TMEM43, TTN
	Non-compaction / Restrictive	ACTC1, BAG3, CSRP3, DES, FLNC, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, SCN5A, TAZ, TNNI3, TNNT2, TPM1, TTN
Metabolic Cardiomyopathies	Fabry / Pompe / Danon / Barth / Amyloid	GLA, GAA, LAMP2, TAZ, TTR
Channelopathies & Arrhythmias	Long QT / Short QT / Brugada / CPVT	CACNA1C, CALM1, CALM2, CALM3, CAV3, CASQ2, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, SCN5A, TRDN
	Familial AF / WPW / Conduction Defect	ABCC9, LMNA, PRKAG2, RYR2, SCN5A, TNNI3, TNNT2

Vascular Syndromes	Marfan / Loeys-Dietz / Aortic Aneurysm	ACTA2, ELN, FBN1, FBN2, FLNA, LOX, MYH11, MYLK, NOTCH1, PRKG1, SKI, SMAD3, SMAD4, TGFB2, TGFB3, TGFB1, TGFB2
	Ehlers-Danlos (Vascular)	COL3A1
RASopathies & Other Syndromes	Noonan / LEOPARD / Costello / CFC	BRAF, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SOS1, SOS2
	Muscular Dystrophies with cardiac involvement	BAG3, CAV3, EMD, FHL1, FKTN, LMNA, TCAP, TMEM43, TTN
	Familial Hypercholesterolemia	ABCG5, ABCG8, APOB, APOE, LDLR, LDLRAP1, LIPA, PCSK9

How to Get Started

1 Physician prescribes the test	2 iGenetx ships a saliva collection kit	3 Sequencing & interpretation performed	4 Report sent to physician to review with patient
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"As doctors we are used to seeing the most severe cases, but with genetics you are anticipating."

View on mobile



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