

More than 1 in 200 people have an inherited cardiovascular condition

CLINICAL IMPLICATIONS OF CARDIOLOGY GENETIC TESTING

The underlying molecular etiology of heritable arrhythmias, cardiomyopathies, aortopathies, and other heart conditions can be identified through genetic testing. Genetic testing is an important aid in the diagnosis of these diseases and provides important prognostic and therapeutic information.¹⁻⁷

- DIAGNOSIS**
- Confirms a clinical diagnosis
 - Differentiates from other causes
 - Fulfills diagnostic criteria for complex heart disorders
 - Identifies at-risk asymptomatic and pre-symptomatic family members

- PROGNOSIS**
- Identifies patients and family members at highest risk for life-threatening arrhythmias, aortic dissections, or heart failure
 - Predicts response to pharmacotherapies

- MANAGEMENT**
- Informs avoidance of certain medications and guides lifestyle modifications to avoid triggering cardiac events
 - Aids in the decision to place implantable cardiac defibrillator or pacemaker and directs treatment options such as enzyme replacement therapy, early surgical intervention, and heart transplantation
 - Enables appropriate screening intervals and interventions for at-risk family members

GUIDELINES AND CONSENSUS STATEMENTS RECOMMEND GENETIC TESTING

Guidelines and consensus statements endorsed by numerous professional societies, including ACCF, AHA, HFSA, HRS, and PACES, recommend cardiology genetic testing.⁴⁻⁷

Heart Rhythm Society (2011) Consensus Recommendations⁴

	CONDITION	PATIENTS WITH ESTABLISHED OR SUSPECTED DIAGNOSIS	FAMILY
ARRHYTHMIAS	Brugada syndrome	Can be useful	Recommended
	Catecholaminergic polymorphic ventricular tachycardia	Recommended	Recommended
	Long QT syndrome	Recommended: For diagnosed/suspected patients or asymptomatic patients with idiopathic, serial QTc values >480 ms (prepuberty) or >500 ms (adults) May be considered: For asymptomatic patients with idiopathic, serial QTc values >460 ms (prepuberty) or >480 ms (adults)	Recommended
	Short QT syndrome	May be considered	Recommended
	Postmortem SIDS/SUDS	May be considered: In the setting of autopsy-negative sudden unexplained death syndrome (SUDS)	Recommended
CARDIOMYOPATHIES	Arrhythmogenic right ventricular cardiomyopathy	Can be useful: For patients satisfying 2010 task force diagnostic criteria May be considered: For patients with 1 major or 2 minor criteria Not recommended: For patients with only a single, minor criterion	Recommended
	Hypertrophic cardiomyopathy	Recommended	Recommended
	Dilated cardiomyopathy	Can be useful	Recommended
	Left ventricular noncompaction	Can be useful	Recommended

INVITAE CARDIOLOGY GENETIC TESTS

ARRHYTHMIA AND CARDIOMYOPATHY		CARDIOMYOPATHY AND SKELETAL MUSCLE DISEASE	
Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel	up to 149 genes	Invitae Cardiomyopathy and Skeletal Muscle Disease Panel	up to 158 genes
ARRHYTHMIA		AORTOPATHY AND CONNECTIVE TISSUE DISORDERS	
Invitae Arrhythmia Comprehensive Panel	up to 73 genes	Invitae Aortopathy Comprehensive Panel	up to 25 genes
Invitae Arrhythmogenic Cardiomyopathy Panel	up to 24 genes	Invitae Ehlers-Danlos Syndrome Panel	15 genes
Invitae Brugada Syndrome Panel	up to 20 genes	Invitae Loeys-Dietz Syndrome Panel	up to 6 genes
Invitae Catecholaminergic Polymorphic Ventricular Tachycardia Panel	8 genes	Invitae Marfan Syndrome Test	1 gene
Invitae Long QT Syndrome Panel	up to 17 genes	FAMILIAL HYPERCHOLESTEROLEMIA	
Invitae Short QT Syndrome Panel	up to 6 genes	Invitae Familial Hypercholesterolemia Panel	4 genes
CARDIOMYOPATHY		PULMONARY HYPERTENSION	
Invitae Cardiomyopathy Comprehensive Panel	up to 106 genes	Invitae Pulmonary Arterial Hypertension Panel	up to 9 genes
Invitae Arrhythmogenic Cardiomyopathy Panel	up to 23 genes	Invitae Hereditary Hemorrhagic Telangiectasia Panel	5 genes
Invitae Dilated Cardiomyopathy Panel	up to 70 genes	Invitae Capillary Malformation-Arteriovenous Malformation Syndrome Test	up to 4 genes
Invitae Hypertrophic Cardiomyopathy Panel	up to 60 genes	CONGENITAL HEART DISEASE	
Invitae Left Ventricular Noncompaction Panel	up to 19 genes	Invitae Congenital Heart Disease Panel	42 genes
Invitae Transthyretin Amyloidosis Test	1 gene	Invitae CHARGE Syndrome Test	1 gene
Invitae Hereditary Hemochromatosis Panel	5 genes	Invitae Costello Syndrome Test	1 gene
Invitae RASopathies Comprehensive Panel	18 genes	Invitae Holt-Oram Syndrome Test	1 gene
		Invitae RASopathies Comprehensive Panel	18 genes
		Invitae Sotos Syndrome Test	1 gene

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