

Hereditary Cardiovascular Genetic Testing

REFERENCE GUIDE



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More than 1 in 200 people have an inherited cardiovascular condition. Ambry's mission is to provide the most advanced genetic testing information available to help you identify those at-risk and determine the best treatment options. If we know a patient has a disease-causing genetic change, not only does it mean better disease management, but it also indicates that we can test others in the family and provide them with potentially life-saving information.

We test for:

ARRHYTHMIA

RhythmFirst/RhythmNext (long QT syndrome*, Brugada syndrome**)

CPVTNext (catecholaminergic polymorphic ventricular tachycardia)*

ARVCNext (arrhythmogenic right ventricular cardiomyopathy)**

CardioNext

CustomNext-*Cardio*

CARDIOMYOPATHY

HCMFirst/HCMNext (hypertrophic cardiomyopathy)*

DCMNext (dilated cardiomyopathy)*

ARVCNext (arrhythmogenic right ventricular cardiomyopathy)**

LVNCNext (left ventricular non-compaction)**

CMNext (cardiomyopathy)

CardioNext

CustomNext-*Cardio*

AORTIC ANEURYSMS/ DISSECTIONS

TAADNext (thoracic aortic aneurysms/dissections)

Marfan syndrome

Ehlers-Danlos syndrome, vascular type

CustomNext-*Cardio*

FAMILIAL HYPERCHOLESTEROLEMIA

FHNext

* Class I recommendation by Heart Rhythm Society and European Heart Rhythm Association

** Class IIa recommendation by Heart Rhythm Society and European Heart Rhythm Association

Choose the Right Test

Our hereditary cardiovascular testing menu includes large multi-gene panels, single gene testing, and customizable panel options for inherited arrhythmias, cardiomyopathies, aortic aneurysms/dissections, and familial hypercholesterolemia.

	TO CONFIRM A DIAGNOSIS OR RISK	TEST NAME	YIELD [^]	GENES	TURNAROUND TIME
Cardiomyopathy	Hypertrophic cardiomyopathy (HCM)	HCMFirst <i>(reflex testing available)</i> HCMNext <i>(reflex testing available)</i>	50%	MYBPC3, MYH7	2-3 weeks
	Dilated cardiomyopathy (DCM) with cardiac conduction disease and/or family history of premature sudden death	DCMNext	30-40%	36 genes	2-3 weeks
	Left ventricular non-compaction (LVNC)	LVNCNext	unknown	8 genes	2-3 weeks
	Arrhythmogenic right ventricular cardiomyopathy (ARVC)	ARVCNext	50-60%	9 genes	2-3 weeks
Arrhythmia	Clinical suspicion of catecholaminergic polymorphic ventricular tachycardia (CPVT)	CPVTNext	50-60%	6 genes	2-3 weeks
	Long QT syndrome (LQTS), clinical suspicion or QT interval >480ms (adolescents) or >500ms (adults) OR Clinical suspicion of Brugada syndrome (BrS), type I OR Unclear arrhythmia diagnosis that appears to be inherited OR Sudden cardiac arrest/death at 35 years of age or younger, with a normal heart structure	RhythmFirst <i>(reflex testing available)</i> RhythmNext <i>(reflex testing available)</i>	~70% for LQTS; 16-30% for BrS	12 genes	2-3 weeks
			75% for LQTS; 15-30% for BrS	36 genes	
Other	Hypercholesterolemia and a family history of same*	FHNext	70%	APOB, LDLR, PCSK9, LDLRAP1 + pharmacogenetic c.521T>C SNP in SLCO1B1	2-3 weeks
	Thoracic aortic aneurysm/dissection (TAAD) and a family history of TAAD or sudden death	TAADNext	30-40%	22 genes	2-3 weeks

Class I Recommendations** Class II Recommendations** Indications for cardiovascular genetic testing

* Adults (20 yrs of age or older): LDL cholesterol >190 mg/dl or non-HDL cholesterol > 220 mg/dl
Children, adolescents and young adults (under 20 yrs of age): LDL cholesterol > 160 mg/dl or non-HDL cholesterol > 190 mg/dl

** Adapted from Ackerman MJ, et al. Heart Rhythm Society/European Heart Rhythm Association expert consensus statement on the state of genetic testing for the channelopathies and cardiomyopathies. *Heart Rhythm*. 2011 Aug;8(8):1308-39.

[^] Theoretical yield from medical literature

Please visit ambrygen.com for information about these options

Over 1 Million Tests Completed

MOVING SCIENCE FORWARD



Purposeful Confirmatory Testing

Many labs validate their tests based on certain limited studies. That's why we participated and led the largest study of its kind (20,000 cases) guiding us to utilize confirmatory testing when we see specific well-defined thresholds. Our mission is to get it right the first time.



Understanding Disease Better Through Free Data Sharing

Identifying an individual's genetic information is nothing new—it's what we do with it that is unique. When labs share genomic information, we can together accelerate the understanding of human disease. Through AmbryShare, we leverage de-identified information to collaborate with others and help people everywhere find answers.



SuperLab

Our 65,000 square foot highly-automated CLIA/CAP certified lab produces some of the fastest turnaround times in the industry, without compromising testing accuracy or specificity.



Ambry's Translational Genomics (ATG) Lab

As an advanced diagnostic lab, it's our responsibility to ensure the results you get from us are accurate and that classification is as complete and robust as possible. Our ATG lab is a unique laboratory that provides an additional service at no additional cost for you and your patients to generate more precise data potentially bringing clarity to some variants of unknown significance (VUS). This helps to actively drive down the rate of VUS results and can give you an increased understanding of your patient's results, so you can better provide medical management recommendations and improve health outcomes.



Educational Webinar Series

Our free six-part "Mastering Cardiogenetics" webinar series features an overview of cardiovascular genetics and how it can apply to your practice: ambrygen.com/mastering-cardiogenetics

About Ambry



Just as no two fingerprints are alike, the way disease presents itself in every individual is different. Since 1999, our mission has always been about understanding disease better, so treatments and cures can be found faster. Every sample that arrives in our lab is viewed as a person with a life and a story that is unique to only them. By providing advanced confirmation genetic testing for inherited and non-inherited diseases, we can help you make more informed and responsible treatment decisions with your patients.