Ambry Genetics®

Genetic Testing for Inherited Cardiovascular Disease

Reference Guide

Genetic Testing for Patients with Inherited Cardiovascular Disease is Recommended By:

Heart Rhythm Society/European Heart Rhythm Association[1]

Heart Failure Society of America Guideline #4[2]

American Heart Association[3]

DID YOU KNOW?

More than **1 in 60** people have an inherited cardiovascular condition.[4]

Identifying Patients with a Genetic Cause for Their Cardiovasuclar Disease Can Clarify a Diagnosis and Inform Recommendations for Personalized Medical Management.

Benefits of genetic testing for cardiovascular disease may include:

- **Clarify or Confirm a Diagnosis**
- **Guide Personalized Medical Management Recommendations**
- **Avoid Genotype-Specific Cardiac Event**
- **Identify At-Risk Family Members**

Cardiovascular Genetic Testing

Ambry Genetics Offers a Range of Options to Help Identify an Underlying Cause and Inform Management

Comprehensive Cardiomyopathy + Arrhythmia Panel

› Cardiomyopathies › Arrhythmias › Autopsy-negative sudden cardiac death[1]	CardioNext®	92 genes

Cardiomyopathy

› Cardiomyopathies (Comprehensive)	CMNext®	56 genes
Hypertrophic Cardiomyopathy[1,2]	HCCMNext®	30 genes
Dilated cardiomyopathy with cardiac conduction disease and/or family history of sudden death[1,2]	DCMNext®	37 genes
Arrhythmogenic right ventricular cardiomyopathy[2]	ARVCNext®	11 genes
Peripartum Cardiomyopathy[2]	CardioNext®	92 genes

Arrhythmia

› Arrhythmias (Comprehensive)	RhythmNext®	42 genes
Left ventricular non-compaction[1]	DCMNext®	37 genes
Long QT syndrome[1] Short QT syndrome Brugada syndrome[1]	LongQTNext®	17 genes
Catecholaminergic polymorphic ventricular tachycardia[1]	CPVTNext®	4 genes

Familial Hypercholesterolemia and Lipid Disorders

› Personal or family history of hypercholesterolemia*,[5] ▪ **Children:** LDL-C levels >160 mg/dL and >1 first-degree relative similarly affected or LDL-C levels >190 mg/dL even in the absence of family history ▪ **Adults:** LDL-C levels >190 mg/dL and >1 first-degree relative similarly affected or LDL-C levels >250 mg/dL even in the absence of a positive family history	**FHNext®**	**4 genes**
› Familial chylomicronemia syndrome	**FCSNext®**	**5 genes**
› Sitosterolemia	**Sitosterolemia**	**2 genes**
› Familial HDL deficiency › Lysosomal acid lipase deficiency › LCAT deficiency/fish-eye disease › Hyperlipoproteinemia type III › Cerebrotendinous xanthomatosis › Apolipoprotein C-III deficiency	**CustomNext-*Cardio*®**	**Select from 167 genes**

Other

› Noonan syndrome and other RASopathies	**NoonanNext®**	**18 genes**
› Thoracic aortic aneurysm/dissection, Marfan, and related disorders[6] ▪ Age at diagnosis <50 years, or ▪ Age at diagnosis 50-60 years, no hypertension, or ▪ Positive family history ▪ Syndromic features	**TAADNext®**	**35 genes**
› Familial transthyretin amyloidosis	**TTR**	**1 genes**

Guidelines recommend genetic testing for patients with these conditions

* 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

RESULTS IN 2-3 WEEKS

All panels have a turnaround time of 2-3 weeks.

Moving Science Forward

No-Cost Family Variant Testing

Testing for all blood relatives is available within 90 days of the original report if the proband was tested at Ambry. Family testing is done via specific site analysis for pathogenic or likely pathogenic variants. (Excludes SNP array and applies to single gene, panel or exome testing.)

Flexible Sample Options

Ambry accepts blood, saliva and other sample types to perform genetic testing.

Sample for Life

We periodically review variants and let you know when there is updated information, such as a reclassification. This is part of our commitment to finding answers.

Complimentary Post-Test Genetic Counseling

To help advise on the impact of test results, Ambry Genetics works with third-party genetic counseling service providers to offer no-cost, post-test genetic counseling for patients with a positive and/or VUS result.

References

1. Heart Rhythm Society (HRS)/ European Heart Rhythm Association (EHRA) Guidelines. *Heart Rhythm.* 2011 Aug;8(8):1308-39.
2. Heart Failure Society of America (HFSA) Guideline, *Journal of Cardiac Failure* (2018), https://doi.org/10.1016/j.cardfail.2018.03.004.
3. Musunuru K, et al. Genetic Testing for Inherited Cardiovascular Diseases: A Scientific Statement From the American Heart Association. *Circ Genom Precis Med.* 2020 Aug;13(4):e000067.
4. Centers for Disease Control and Prevention. "Heart Disease Prevalence." National Center for Health Statistics. https://www.cdc.gov/nchs/hus/topics/heart-disease-prevalence.htm. August 2022.
5. Sturm, Amy C., et al., Clinical Genetic Testing for Familial Hypercholesterolemia. *Journal of the American College of Cardiology.* 72 (2018) 662-680.
6. Verhagen, Judith M.A., et al., Expert consensus recommendations on the cardiogenetic care for patients with thoracic aortic disease and their first-degree relatives. *International Journal of Cardiology.* 258 (2018) 243-248.

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