

Hereditary Cardiovascular Conditions

Patient Guide

Genetic Testing for Inherited
Cardiovascular Diseases



Ambry Genetics®

A TEMPUS COMPANY

Understanding The Basics

34 million people

worldwide have Familial
Hypercholesterolemia,

90% remain undiagnosed¹



As many as
1 in 400 adults
may have a
cardiomyopathy
condition²

WHAT IS AN INHERITED CARDIOVASCULAR DISORDER?

There are many different disorders that involve the heart. Inherited heart diseases are those that run in families and are caused by a change in one gene or in a number of genes. There are many conditions of inherited heart diseases including cardiomyopathies, arrhythmias, thoracic aortic aneurysms and dissections, and familial hypercholesterolemia. Individuals with these conditions can be asymptomatic, and sometimes sudden death could be the first and only symptom.

WHAT ARE SYMPTOMS OF AN INHERITED CARDIOVASCULAR DISORDER?

Symptoms vary from person to person, but inherited cardiovascular disorders can cause shortness of breath, fatigue, chest pain, palpitations, fainting, coughing, nausea, or even sudden death in rare cases.



More than **1 in 60** people have an inherited cardiovascular condition³

There is a **50% chance** of inheriting an autosomal dominant cardiovascular disorder when you have an affected parent, child, or sibling.⁴



Each year in the United States, **approximately 320,400 people** die suddenly and unexpectedly due to Sudden Cardiac Arrest⁵

CAUSES FOR INHERITED CARDIOVASCULAR DISORDERS

Most inherited heart disorders are passed down in an autosomal dominant pattern which means that the genetic variation is inherited from just one parent. These disorders can often show up in multiple generations in a family and can be associated with death at a young age due to sudden cardiac arrest or an aortic dissection. Knowing if you or any family members are at an increased risk for one of these disorders can help prevent any serious events by getting timely medical care.

Other heart diseases can run in families in a more complicated pattern. Often, these are caused by the combination of genes that are passed down and environmental factors, including diet, exercise, smoking, ethnicity, gender and more. If there is a family history of high cholesterol, heart attacks or stroke, there is an increased risk for heart disease.

Should You Consider Genetic Testing For You or Your Child?

TALK TO A HEALTHCARE PROVIDER IF YOU ANSWER “YES” TO ANY OF THE FOLLOWING:

1

Have you/your family members* been diagnosed with any inherited heart disease/disorder?

2

Have you/your family members* been diagnosed with high cholesterol?

3

Is there any history of a sudden unexpected death or cardiac arrest in your family?

4

Does anybody in your family have a pacemaker or implantable cardioverter defibrillator (ICD) device?

5

Is there a personal or family history of unexplained fainting or passing out?

The healthcare provider may identify other reasons why genetic testing should be considered.

**“Family members” refers to blood relatives, such as brothers/sisters/parents/grandparents/children/aunts/uncles/cousins*

Understanding Disease Better Through Quality Testing

GENES CARRY A STORY THAT IS UNIQUE TO EACH PERSON. GENETIC TESTING CAN HELP BETTER UNDERSTAND AND MANAGE THE CARDIOVASCULAR DISORDER IN A FAMILY.

Genetic testing for cardiovascular disorders can include a varying number of genes that are linked to the symptoms that you or your family member has. Based on the results, the healthcare provider may discuss more specific prognosis and treatment options for you and your family.

The genetic testing recommended for you today includes one or more of the tests below.

- | | | |
|--------------------------------------|---|---|
| ☐ CardioNext® | ☐ LongQTNext™ | ☐ TAADNext® |
| ☐ CMNext® | ☐ CPVTNext® | ☐ TTR |
| ☐ RhythmNext® | ☐ FHNext® | ☐ CustomNext-Cardio® |
| ☐ HCMNext® | ☐ FCSNext® | ☐ Other _____ |
| ☐ DCMNext® | ☐ Sitosterolemia Panel | |
| ☐ ARVCNext® | ☐ NoonanNext™ | |

VISIT OUR WEBSITE

See updated information on which genes are included on the test your healthcare provider selected above: ambrygen.com/patients/cardiology

How Genetic Testing Can Impact You and Your Family



Finding a genetic cause for you or your family members' cardiovascular disorder can help:

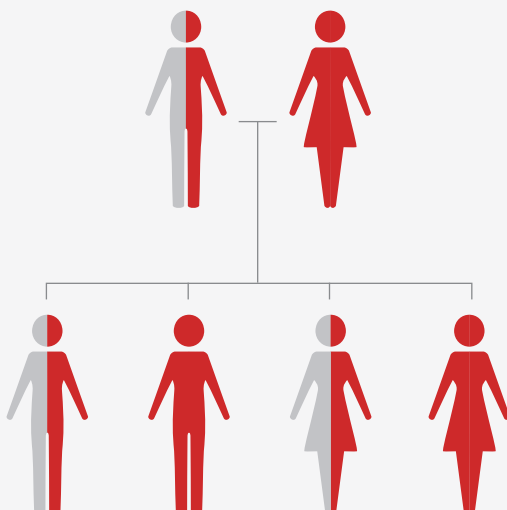
- Confirm a diagnosis, particularly when clinical criteria are unclear or borderline in an individual
- Choose a better medicine or other treatment method
- Identify an inherited variant following a sudden unexplained death
- Offer family members genetic testing and implement medical surveillance to only those that need it
- Clarify the chance for family members to inherit a genetic variant and their risk to develop symptoms
- Reduce healthcare costs, resources, and anxiety for families

FOR YOUR FAMILY MEMBERS:

If a test is positive for a genetic variant, close family members have a 50/50 random chance of also having the same mutation.

 Has genetic variant

 No genetic variant



Possible Genetic Test Results

POSITIVE

A harmful variant (genetic difference also known as mutation or pathogenic variant) was found in at least one of your genes tested.

There may be management recommendations specific to the gene that has a variant.

Genetic testing for certain family members may be recommended.

NEGATIVE

No genetic variants or differences were found in any of your genes tested.

Management recommendations are based on personal and family history.

Talk to your healthcare provider to find out if genetic testing should be considered for your family members.

VARIANT OF UNKNOWN SIGNIFICANCE (VUS)

At least one genetic variant or difference was found, but it is unclear if this difference causes an increased risk for cardiovascular disease. Although no genetic variants or differences were found in any of your genes tested, this does not rule out the possibility of a genetic component because we do not know all of the genetic causes at this time.

Management recommendations are based on personal and family history.

Talk to the healthcare provider to find out if genetic testing should be considered for family members.

It is possible to have a combination of positive and VUS results, since multiple genes are tested.

Resources

Ambry Genetics' Patient
Education Website

ambrygen.com/patients

The Family Heart Foundation

familyheart.org

Hypertrophic
Cardiomyopathy Association

4hcm.org

The Marfan Foundation

marfan.org

Children's Cardiomyopathy
Foundation

childrenscardiomyopathy.org

Foundation of the National
Lipid Association

learnyourlipids.com

Genetic Information
Nondiscrimination Act

ginahelp.org

Foundation for Inherited
Arrhythmias

fiacardiac.org

Aortic Hope

aortichope.org

Dilated Cardiomyopathy
Foundation

dcmfoundation.org



FIND A GENETIC COUNSELOR

National Society of Genetic
Counselors

nsgc.org

Canadian Association of
Genetic Counsellors

cagc-accg.ca

Frequently Asked Questions

1 HOW IS GENETIC TESTING PERFORMED AND HOW LONG DOES IT TAKE?

Genetic testing requires a blood, buccal, or saliva sample, which is collected using a special kit that is shipped overnight to Ambry Genetics by the healthcare provider. The testing, which looks for gene variants that cause an increased risk for cardiovascular disease, takes less than three weeks to complete, and results are sent to the healthcare provider.

2 WHAT WILL HAPPEN WHEN RESULTS ARE READY?

The healthcare provider will receive the results. Every healthcare provider may have a different method and time frame for reviewing results, so it is important to discuss this process with them. The healthcare provider will discuss recommended next steps based on the test results.

3 WILL THE GENETIC TEST RESULTS AFFECT MY INSURANCE COVERAGE?

In the U.S., the Genetic Information Nondiscrimination Act (2008) prohibits discrimination by health insurance companies and employers, based on genetic information. Visit ginahelp.org to learn more.

4 HOW WILL THE TEST RESULTS BE PROTECTED?

Ambry Genetics is required by law to maintain the confidentiality of protected health information in accordance with the Health Insurance Portability and Accountability Act (HIPAA). Visit HHS.gov to learn more.

5 SHOULD I TELL FAMILY MEMBERS ABOUT THE GENETIC TEST RESULTS?

It is important to share results with family members, because they may provide additional information about their own risk for cardiovascular disease and management options. If you feel unsure about how to approach the subject, the healthcare provider may be able to offer some advice.

6 WILL GENETIC TESTING BE COVERED BY INSURANCE?

Many insurance plans cover genetic testing, and Ambry Genetics is contracted with the majority of U.S. health plans. The out-of-pocket cost may vary based on each individual plan. A team of dedicated specialists is available to help patients get access to the genetic testing needed, and provide further details about payment options. Please call or email our Billing department at +1.949.900.5795 or billing@ambrygen.com with any questions. Visit ambrygen.com/patientbilling for more information.

7 WHAT IS AN EXPLANATION OF BENEFITS (EOB)?

An EOB is a statement of benefits provided by the insurance company. This is not a bill, but it will outline the expenses the insurance company will cover for the medical care received. If you have any questions regarding the EOB, contact the insurance company directly or connect with a Billing specialist from Ambry. Please note that the processing of claims by insurance companies may take a few weeks or even a couple of months.

STILL HAVE QUESTIONS?

Talk to the healthcare provider or visit our website: ambrygen.com



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References

1. Nordestgaard, Børge G., et al. "Familial hypercholesterolaemia is underdiagnosed and undertreated in the general population: guidance for clinicians to prevent coronary heart disease: consensus statement of the European Atherosclerosis Society." *European Heart Journal* 34.45 (2013): 3478-3490.
2. Ramos López A, Sabater Molina M, Gil Ortuño C, et al. Geographic variations in the prevalence of inherited cardiac diseases and in the incidence of related sudden death. *Int J Cardiol.* 2026;446:134083. doi:10.1016/j.ijcard.2025.134083
3. <https://www.cdc.gov/nchs/hus/topics/heart-disease-prevalence.htm>
4. Cirino, Allison L., and Carolyn Y. Ho. "Genetic testing for inherited heart disease." *Circulation* 128.1 (2013): e4-e8.
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