

Hereditary Cardiovascular
Conditions
PATIENT GUIDE

GENETIC TESTING FOR INHERITED
CARDIOVASCULAR DISEASES

Hereditary Cardiovascular Conditions

PATIENT GUIDE

Understanding The Basics

34 million people
worldwide have Familial
Hypercholesterolemia,
90% remain undiagnosed



As many as
1 in 500 adults
may have a
cardiomyopathy
condition

WHAT IS AN INHERITED CARDIOVASCULAR DISORDER?

There are many different disorders that involve the heart. Some of these are more likely to run in families than others. Inherited heart diseases are those that run in families and are caused by a change (or mutation) 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. These conditions will often be asymptomatic that may lead to sudden death as 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 200** people have an inherited cardiovascular condition



There is a **50% chance** of inheriting an autosomal dominant cardiovascular disorder



Each year in the United States, **approximately 210,000** die suddenly and unexpectedly due to Sudden Cardiac Arrest

CAUSES FOR INHERITED CARDIOVASCULAR DISORDERS

Most inherited heart disorders are passed down in an autosomal dominant pattern and often show up in multiple generations in a family. These disorders can also be associated with death at a young age due to sudden cardiac arrest or an aortic dissection. Knowing if you are at an increased risk for one of these disorders can help make sure that you and your relatives get the proper medical care to prevent any serious events.

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 you have a family history of high cholesterol, heart attacks or stroke, you may have an increased risk for heart disease.

Should You Have Genetic Testing?

IF YOU ANSWER “YES” TO ANY OF THE QUESTIONS BELOW, hereditary cardiovascular genetic testing may be something for you and/or your family members to consider.

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?

Your healthcare provider may identify other reasons why it may be beneficial for you to consider genetic testing.

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

Understanding Disease Better Through Quality Testing

YOUR GENES CARRY A STORY THAT IS UNIQUE TO YOU AND MAKE YOU WHO YOU ARE. GENETIC TESTING CAN HELP YOU BETTER UNDERSTAND AND MANAGE THE CARDIOVASCULAR DISORDER IN YOUR FAMILY.

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

Ambry participates in important studies to expand our knowledge of cardiovascular disorders. To that end, our testing menu is continually being updated to include the most important genes for you and your family members.

Your healthcare provider has determined that the best test for you is:

- | | |
|--------------------------------------|--------------------------------------|
| <input type="checkbox"/> TAADNext® | <input type="checkbox"/> HCMNext® |
| <input type="checkbox"/> FHNNext® | <input type="checkbox"/> LongQTNext™ |
| <input type="checkbox"/> RhythmNext® | <input type="checkbox"/> NoonanNext™ |
| <input type="checkbox"/> DCMNext® | <input type="checkbox"/> CPVTNext® |
| <input type="checkbox"/> CMNext® | <input type="checkbox"/> ARVCNext® |
| <input type="checkbox"/> CardioNext® | |

VISIT OUR WEBSITE

See updated information on which genes are included on the test your healthcare provider selected above: ambrygen.com/patient/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 or rule out a suspected inherited heart disease or help determine your chance of developing or passing it on
- 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 mutation following a sudden unexplained death with autopsy findings
- Offer family members genetic testing (for a familial mutation) and implement medical surveillance to only those that need it
- Clarify risks to family members, including the inheritance pattern
- Reduce healthcare costs, resources, and anxiety for families


AMBRY GENETICS OFFERS NO-COST TESTING TO CLOSE RELATIVES

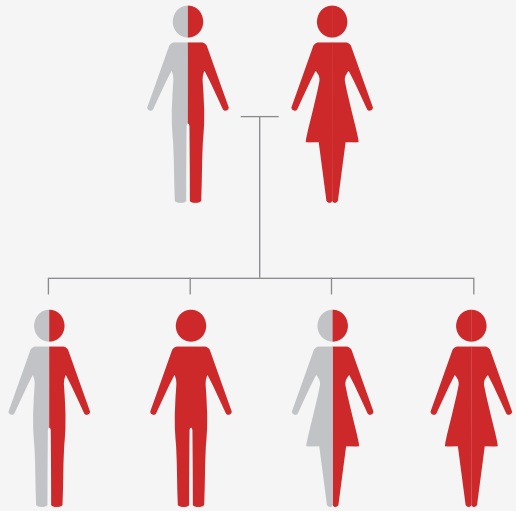
This is available for the specific genetic mutation identified in the first family member tested at Ambry within 90 days of the original report date.

FOR YOUR FAMILY MEMBERS:

If you test positive for a genetic mutation, your close family members (like your parents, brothers, sisters and children) have a 50/50 random chance of also having the same mutation.

 Has genetic mutation

 No mutation



Possible Genetic Test Results

POSITIVE

A mutation was found in at least one of your genes tested

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

Genetic testing for certain family members may be recommended.

NEGATIVE

No genetic mutations 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 change was found, but it is unclear if this change causes the cardiovascular disorder or not

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.

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

Resources For You

Ambry Genetics' Patient
Education Website

ambrygen.com/patient

FH Foundation

theafhfoundation.org

Hypertrophic Cardiomyopathy
Association

4hcm.org

Marfan Foundation

marfan.org

Children's Cardiomyopathy
Foundation

childrenscardiomyopathy.org

National Lipid Association

learnyourlipids.com

Genetic Information
Nondiscrimination Act

ginahelp.org

Sudden Arrhythmia Death
Syndrome SADS Foundation

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

2 WHAT WILL HAPPEN WHEN MY RESULTS ARE READY?

Your healthcare provider will receive your results; they will not be sent directly to you. Every healthcare provider may have a different method and time frame for reviewing your results with you, so it is important to discuss this process with them when your test is performed. Your healthcare provider will discuss recommended next steps based on your test results.

3 WILL MY 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. Depending on where you live in the world, you may have different (or fewer) laws in this area. Visit ginahelp.org to learn more.

4 HOW WILL MY TEST RESULTS BE PROTECTED?

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

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

It is important to share your results with your 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, your healthcare provider may be able to offer some advice.

6 WILL GENETIC TESTING BE COVERED BY MY INSURANCE?

Many insurance plans cover genetic testing, and Ambry Genetics is contracted with the majority of U.S. health plans. Your out-of-pocket cost may vary based on your individual plan. A team of dedicated specialists is available to help you get access to the genetic testing you need, and provide further details about our 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)?

Your insurance company sends you an EOB to explain any services paid on your behalf. You can contact us directly to speak with a Billing specialist with any questions or concerns about Ambry Genetics genetic testing that appears on your EOB. It is important to remember that insurance companies can take several weeks or even a couple of months to process claims.

STILL HAVE QUESTIONS?

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

Elevating the Standard of Care™

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