Finding answers quickly to help guide patient care

Hereditary Cardiovascular Conditions

Genetic Testing for Inherited Cardiovascular Diseases
Know the Basics

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.

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

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

CAUSES FOR INHERITED CARDIOVASCULAR DISORDERS

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

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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:

- TAADNext
- FHNext
- RhythmNext
- DCMNext
- CMNext
- HCMNext
- LongQTNExt
- NoonanNext
- CPVTNext
- ARVCNext

VISIT OUR WEBSITE

See updated information on which genes are included on the test your healthcare provider selected above: ambrygen.com/patient/cardiotest
What are the Benefits of Genetic Testing?

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
WHAT HAPPENS IF I TEST POSITIVE FOR A GENE MUTATION?

There is a 50/50 random chance that each of your children, brothers, sisters, and parents, also has this same mutation. The picture below shows how people carry and pass on these mutations. Your family members can now be tested for this mutation, if they would like. Any family members who also carry the mutation are at increased risk to develop the cardiovascular disorder and should be followed by a cardiologist. Any family members who test negative for this mutation are likely not at increased risk for the cardiovascular disorder and should not need to have a cardiologist follow them closely.
Possible Genetic Test Results

+ POSITIVE

A mutation was found in at least one of the 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 changes were found in any of the 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’s Patient Education Website
ambrygen.com/patient

FH Foundation
thefhfoundation.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
1 HOW IS GENETIC TESTING PERFORMED AND HOW LONG DOES IT TAKE?

Genetic testing is done using a blood or saliva sample, which is collected using a special kit that is shipped overnight to Ambry (all coordinated by your healthcare provider). Testing looks for mutations that cause the cardiovascular disorder in your family. It takes between 1-3 weeks for the testing to be complete depending on which test your provider orders. 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 to contact you to discuss your results, so it is important to discuss this process with them. Based on your test results, your healthcare provider will discuss any next steps.

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 your area. Visit ginahelp.org to learn more.
4 SHOULD I TELL MY FAMILY MEMBERS ABOUT MY GENETIC TEST RESULTS?

It is important to share your results with your family members as they may provide additional information about their chance of having the same disorder. Your healthcare provider may be able to guide you on finding the best way to inform family members.

5 WILL GENETIC TESTING BE COVERED BY MY INSURANCE?

Many insurance plans cover genetic testing and Ambry is contracted with the majority of U.S. health plans. Your out-of-pocket cost may vary based on your individual plan; therefore, we offer personalized verification of insurance coverage and financial options for your genetic testing. A team of dedicated specialists is available to help you get access to the genetic testing you need and answer any questions you have about our payment options. Call or email our Billing department at +1.949.900.5795 or billing@ambrygen.com with any questions.

6 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 your EOB. Some genetic tests take weeks to process in order to receive the best results. In addition, insurance companies can take several weeks or even a couple of months to process claims.

STILL HAVE QUESTIONS?
Talk to your doctor or visit our website: ambrygen.com
Finding Answers.