BREAKTHROUGH
GENOMICS

Cardiovascular Risk Assessment Test

Genetic Screening

for Cardiovascular conditions & disease

HIPAA COMPLIANT

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

CLIA Certified

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BREAKTHROUGH GENOMICS

Cardiovascular Risk Assessment Test

This test is also available for healthy individuals who want to understand their long-term risk and treatment options for potentially debilitating heart conditions that they may develop in the near future.

Benefits to genetic testing for cardiovascular conditions include:

- ✔ Improved diagnosis and treatment options
- ✔ Informed recommendations for prevention and monitoring
- ✔ Improved clinical management decisions for existing conditions
- ✔ Enhanced understanding of potential risk to family members
- ✔ Options for family planning, including preimplantation genetic diagnosis to try to avoid passing a disease-causing mutation to offspring
- ✔ Cost-savings by eliminating the need for some unnecessary screening procedures and other more invasive tests.

The following categories of cardiovascular disease and conditions that are all included in the BREAKTHROUGH GENOMICS' Comprehensive Cardiovascular Test

Arrhythmias (covers 62 genes)

This category covers genes associated with heart rhythm problems (heart arrhythmias) that occur when the electrical signals that coordinate the heart's beats don't work properly.

Atrial fibrillation is the most common cardiac rhythm disturbance that affects many millions of people around the world. The prevalence of arrhythmias increases rapidly with age, rising to 2.3% between the ages of 40 and 60 years, and to 5.9% over the age of 65.

The role of a genetic testing is becoming an essential tool to detect and manage arrhythmia as it can inform the diagnosis, prognosis, and treatment of hereditary arrhythmia diseases for both the patient and related family members.

Aorta Abnormalities (covers 53 genes)

This category of genes test for aortic disease and associated conditions that can present with different symptoms, including ascending aortic dilatation or aneurysms. Aortic dilatation refers to the relative size of a person's aorta that is greater than the 95th percentile for the normal person's age, sex and body size. An aneurysm is defined here as a localized dilation of the aorta that is more than 150 percent of predicted. Most aortic aneurysms are associated with non-syndromic dilatation. However, at least 20% of aortic aneurysms are in the context of syndromic diseases such as Marfan syndrome (MfS), Loeys-Dietz syndrome (LDS), Shprintzen-Goldberg syndrome (SGS) and Ehlers-Danlos syndromes (EDS).

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How Are Cardiovascular Conditions Inherited?

Many inherited cardiovascular conditions are examples of an **autosomal dominant condition** where the alteration of one disease-causing gene from one parent can be passed on to the children. (See figure 1). Other cardiovascular conditions like hypertension and high LDL cholesterol levels that may have a genetic underpinning can be the result of complex patterns of inheritance and involve multiple genes.

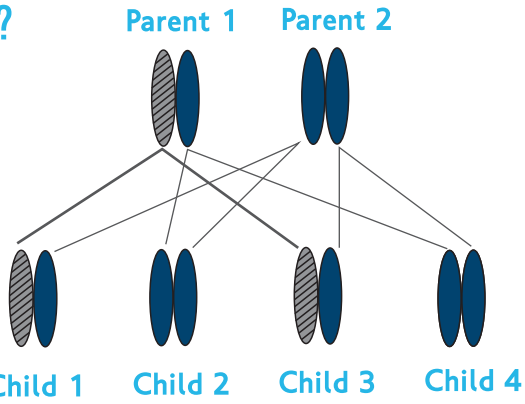


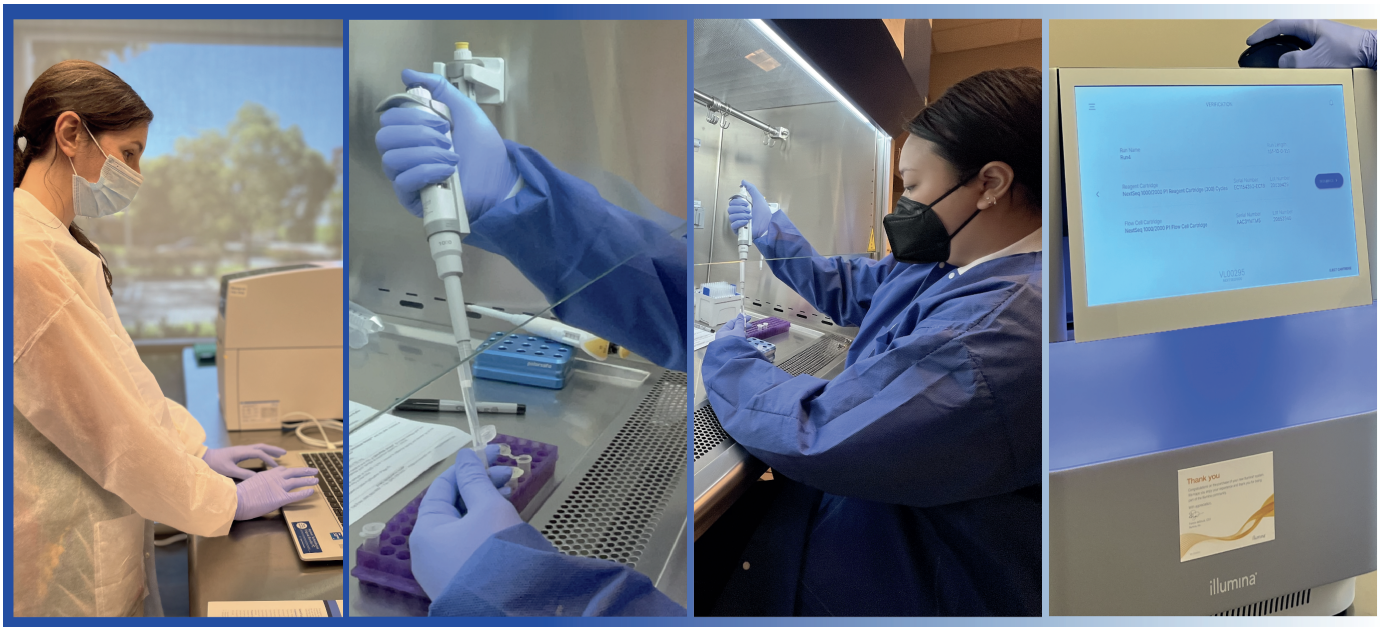
Figure 1 - this diagram shows how cardiovascular conditions can be passed down from parents to their children through an autosomal dominant inheritance pattern.

Comprehensive Cardiovascular Test from Breakthrough Genomics

Breakthrough Genomics' Comprehensive Cardiovascular Genetic Test covers nearly 260 different genes that can all impact cardiovascular health. **This test is strongly recommended for individuals with a family-history** of heart conditions or hypertension or who are concerned about their long-term risk to develop heart disease. This test is also important to help identify family members who also may also be at risk and would benefit from being tested.

Individuals with the following conditions should get tested:

- ✔ High cholesterol levels
- ✔ High blood pressure
- ✔ High levels of stress in their daily lives
- ✔ Family history of heart problems or cardiac disease
- ✔ At-risk lifestyle choices including smoking tobacco and alcohol consumption
- ✔ Heart abnormalities or defects since birth
- ✔ Family history of sudden unexplained deaths or sudden infant death syndrome (SIDS)



Identify individuals with increased risk to develop specific types of heart disease and other cardiovasular conditions

BREAKTHROUGH GENOMICS' Cardiovascular Genetic Risk Test combines advanced Whole Exome Sequencing (WES) data with next-generation bio-informatic analysis to provide comprehensive results with fast turnaround times. comprehensive assessment of an individual's predisposition to an extensive list of cardiovascular conditions and disorders. All tests receive a patient report that is individually reviewed and signed by a U.S. Board-certified medical geneticist.

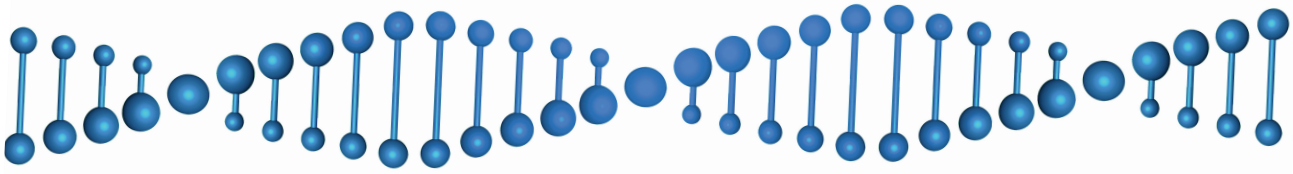
Introduction

Cardiovascular disease is one of the leading causes of death around the world. Sadly, this group of debilitating conditions accounts for over 17 million deaths annually or roughly 30% of global deaths. In the U.S., nearly 80 million people suffer from different types of cardiovascular conditions. Cardiovascular diseases (also called CVDs) include a range of inherited conditions and disorders that can affect people at any age or physical condition.

Inherited genetic variants can play a key role in determining who is most at risk for many common cardiovascular disorders including congenital heart disease, cardiomyopathy, hypertension, myocardial infarction, and others.

Fortunately today, advancements in the rapid analysis of an individual's DNA has given medical professionals the ability to order non-invasive genetic tests for anyone interested in finding out if they are at risk for cardiovascular disease. The tests require only a small amount of a person's saliva or the preparation of a dried blood spot (DBS).

After the genetic test results have been individually reviewed by a clinical geneticist and a signed report is returned to the ordering physician, a medical professional can determine if intervention is necessary and the specific follow-up, preventative treatments and other lifestyle adjustments that might be recommended for the individual to reduce their risk of developing or being impacted by an inherited heart condition.



Atrial Fibrillation (covers 19 genes)

This group of genes cover hereditary forms of atrial fibrillation. Atrial fibrillation is an irregular heart rhythm that begins in the heart's upper chambers (atria). Common symptoms of atrial fibrillation include: fatigue, heart palpitations, trouble breathing and dizziness. Atrial fibrillation is the most common type of sustained arrhythmia, affecting more than 3 million people in the United States and tens of millions more around the world.

As is the case with many cardiovascular conditions, the risk to develop atrial fibrillation increases with age.

Brugada Syndrome (covers 7 genes)

These genes are associated with Brugada Syndrome which is characterized by cardiac conduction abnormalities leading to detectable ST changes in the right precordial leads in a rest ECG.

Brugada Syndrome is responsible for 4-12% of unexpected sudden deaths and up to 20% of all sudden deaths that occur in individuals with an apparently normal heart. Clinical presentations may also include sudden infant death syndrome and sudden unexpected nocturnal death syndrome (SUNDS), a condition that is more common in individuals from Southeast Asia.

Cardiomyopathy (covers 217 genes)

This large set of genes test for inherited cardiomyopathies which are a group of diseases that affect the heart muscle. Cardiomyopathy is linked to a number of genetic diseases including Noonan's Syndrome, Danon Disease, Fabry Disease, mitochondrial myopathy, or muscular dystrophy. Cardiomyopathy can lead to heart failure, arrhythmias, or sudden death.

Congenital Structural Heart Disease (covers 125 genes)

Congenital Heart Disease is the most common type of birth defect, representing a leading cause of infant morbidity and mortality. It is estimated that Congenital Structural Heart Disease affects at least 8 out of every 1,000 newborns. Survival of infants with CHDs depends on how severe the defect is, when it is diagnosed, and how it is treated. Genetic testing for this dangerous disorder can predict recurrence risk, determine the pattern of inheritance within a family, and help to inform decisions for further family screening.

Short QT Syndrome (covers 5 genes)

Short QT Syndrome is a rare genetic disorder that is characterized by an abnormally short QT interval and an increased risk of atrial and ventricular arrhythmias. Short QT Syndrome can lead to sudden cardiac arrest which can occur in up to 40% of patients.

The correct diagnosis can help prevent and determine the right treatment for hundreds of specific heart conditions.

BREAKTHROUGH GENOMICS

A leader in the clinical interpretation of genomic data. The company provides cutting-edge genetic tests for rare disease diagnosis and other inherited conditions at its CAP and CLIA-certified, advanced clinical lab in Southern California

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