Is Genetic Testing for Heart Disease Right for Me?

Heart disease is a leading cause of death worldwide and often runs in families.

Certain heart conditions, such as those in the Figure, are caused by a single harmful DNA variation (or mutation) while others, such as heart attack and high blood pressure, are usually caused by the combined effect of many smaller-impact DNA variations. Tests for the latter are emerging but are not yet reliable enough for common use. Tests for the former can be ordered by cardiologists and genetic counselors. These tests can help patients and their families know who is at risk for disease.

How Do I Know If Genetic Testing Is Right for Me?

If anyone in your family is diagnosed with any of the diseases in the Figure or if you recognize any of the red flags, discuss with your primary care physician whether you should be referred to a cardiologist. The cardiologist may recommend specific heart tests or an appointment with a genetic counselor to discuss your family's heart health. If they think your heart test results and family history are suspicious of an inherited heart condition, they may recommend genetic testing.

What Types of Genetic Tests Are Available for Heart Problems?

A physician or genetic counselor can order a genetic test that looks at many genes to try to find the single variation causing the condition in the family. These genetic tests are performed at a medical-grade laboratory, are often covered by insurance, and are different from genetic tests ordered online that focus on ancestry.

What Types of Results Can I Get on a Multigene Test?

A positive result means that the cause of your family's heart problems was found in the DNA. This may help physicians diagnose your condition more accurately and personalize your treatment. However, often the largest benefit of genetic testing is for the family: if the underlying cause of the heart problem is known, other family members who may be at risk of having the same disease can find out if they carry the same genetic variation. If they do, they might also develop the heart condition and should be followed by a cardiologist.

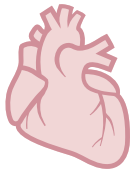
A negative result means the cause of your family's heart problems was not found in the region of DNA tested.

An uncertain result happens when a variation in the DNA is found that is suspicious for causing the disease. We all carry genetic variations, good and bad, in our DNA. Sometimes, we cannot tell if the variation found causes heart problems. Family members should not get tested for these genetic variations because we do not know what it means for their future.

Conditions or Health Histories That Are Red Flags for Inherited Cardiovascular Disease

Diagnosis of one of the following conditions in the family:

Cardiomyopathy (heart muscle disease)
- Hypertrophic, dilated (nonischemic), peripartum, or restrictive cardiomyopathy
- Arrhythmogenic (right) ventricular cardiomyopathy
- Left ventricular noncompaction cardiomyopathy



Arrhythmia (abnormal heart rhythms)
- Long QT or Brugada syndrome
- Catecholaminergic polymorphic ventricular tachycardia
- Unexplained cardiac arrest and/or sudden death
- Sudden infant death syndrome
- Atrial fibrillation (in patients aged <45 y)

Connective tissue disease
- Vascular Ehlers-Danlos, Marfan, or Loeys-Dietz syndrome
- Aortic aneurysm or dissection (in patients aged <50 y)

Other inherited conditions
- Familial hypercholesterolemia
- Familial or unexplained pulmonary hypertension
- Heart attack or coronary artery disease
- Heart defects
- Familial amyloidosis

Personal or family history of the following heart procedures:

All patients aged <50 y
- Implantable cardioverter defibrillator
- Pacemaker implant

All patients aged <60 y
- Left ventricular assist device
- Heart transplant

Female patients aged <65 y and male patients aged <60 y
- Coronary artery bypass or stent surgery

What Can't a Genetic Test Tell Me About My Family's Heart Problem?

A positive genetic test result in a patient does not predict what their exact problems will be. Similarly, finding the genetic variant that causes the family disease in other family members does not mean they will definitely develop the disease. Rather, it shows they are at risk and suggests they should have close follow-up. If a family member is found not to carry the variant that causes the family disease, they are at low risk and do not need close follow-up.

A negative or uncertain genetic test result in a patient does not rule out an inherited heart condition: it only rules out a cause in the genes tested. Because scientists are always discovering new genes, your family may have an answer in the future. Until then, talk to your cardiologist or genetic counselor to learn who in your family needs to see a cardiologist and how often.

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