Natural State Genomics

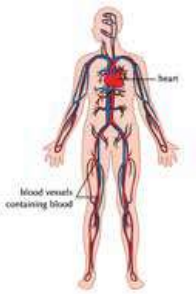
Cardiology
Information for Patients

More than one in 200 people have an inherited cardiovascular condition.

Cardiovascular disorders affect the heart and blood vessels that carry oxygen and nutrients throughout the body. Cardiovascular disorders may occur due to several factors, including heredity.

Some people might carry genetic variants that predispose them to cardiovascular disease and may be asymptomatic. Others may experience fainting, chest pain, shortness of breath, an irregular heartbeat, heart failure, stroke, and/or sudden cardiac death.

Depending on the condition, there might be alternatives to reduce the impact of cardiovascular diseases. Often patients benefit from changes in diet and lifestyle, and the use of medications, surgery, and medical devices (i.e. pacemaker).



What are genes and mutations?

Genes are the instructions in our DNA that tell our bodies how to grow and develop. We all have millions of genetic changes and most do not impact our health. However, some changes may be harmful and cause disease by preventing a gene from working properly. These harmful changes are called mutations.

For most genes, we inherit two copies: one from our mother and one from our father. A person that carries a mutation has a 50% (1 out of 2) chance of passing on the mutation that is associated to disease or the benign change in the other copy of the gene.

Your physician may consider genetic testing if you have a personal or family history of:

- Cardiomyopathy
- Arrhythmia
- Vascular disease
- Extremely high levels of cholesterol in the blood
- Premature heart failure
- Heart attack or stroke at an early age
- Sudden cardiac death, unexplained or accidental death at a young age
- Several relatives have the same type of cardiovascular disease (cardiomyopathy, arrhythmia, extremely high levels of cholesterol in blood, vascular disease)
- Relative with a known disease-causing mutation

Condition	Organ/Tissue Affected	Cause	Medical Issues
Cardiomyopathy	Heart	The heart muscle may become thick, stiff, enlarged, or replaced with scar tissue	Heart failure; shortness of breath; fatigue; swelling of ankles, feet, legs, abdomen, and veins in the neck; dizziness; chest pain; heart murmurs; sudden cardiac death
Arrhythmia	Heart	Irregular heartbeat	Heart palpitations; chest pain; shortness of breath; fainting; weakness; dizziness; sweating; anxiety
Vascular disorders	Blood Vessels	Affects blood flow by weakening, blocking, or damaging vessels	Chest pain; shortness of breath; arterial disease, aneurysm; stroke; death
Familial Hypercholesterolemia	Arteries of the Heart and Brain	Extremely high levels of cholesterol in the blood create blockages in the arteries	Premature heart disease: heart attack, stroke; cholesterol deposits on ankles, hands, and eyes

What are the benefits of genetic testing?

Genetic testing for cardiovascular disorders can:

- Establish or confirm the appropriate diagnosis
- Identify risks for other health concerns that are associated with the condition
- Provide information useful for the best management plan for those with a positive test result
- Allow at-risk family members to undergo genetic testing.
- Provides options for family planning

Possible results of genetic testing

There are three possible results from genetic testing:

⊕POSITIVE:
A pathogenic/likely pathogenic variant is detected. Knowing the specific gene involved tells you how epilepsy in you and your family was inherited. It can also give a doctor insight into your risk for other health conditions associated with the genetic mutation(s).

⊖NEGATIVE:
No mutation detected; benign variant might be detected. No mutation that explains the condition is identified. A true negative result is only when the disease-causing mutation in the family is known is not identified. Management will be determined by personal and family medical history.

? UNCERTAIN:
A variant of unknown significance is detected. There is not enough evidence to classify the change as pathogenic or benign. Management will be based on personal and family medical history.

What does a positive result mean for my family?

A positive result is important for your family, and it is important to share genetic test results whenever possible. Parents, siblings, and children of someone with a cardiovascular condition could have as high as a 50% chance to also have a mutation.

Is genetic testing covered by health insurance?

NS Genomics accepts all commercial and private healthcare insurance plans, which will determine coverage of genetic testing. Prior to testing, NS Genomics' insurance specialists will contact you if there is an anticipated out-of-pocket cost that exceeds $250.

If I have genetic testing, can my employer or health insurance company discriminate against me?

No. The Genetic Information Nondiscrimination Act (GINA) protects Americans from discrimination by an employer or a health insurance company based on genetic testing results and genetic information. Importantly, GINA does not offer protections for disability, long term care, or life insurance. It also does not apply to members of the U.S. military or employees of the Federal government, Indian Health Service, or Veterans Health Administration. For detailed information about GINA, please visit : www.eeoc.gov/laws/types/genetic.cfm

Where can I learn more?

Sudden Arrhythmia Death Syndromes (SADS) Foundation: | www.sads.org

Heart Failure Society of America (HFSA): | www.hfsa.org

The FH Foundation: | thefhfoundation.org

American Heart Association: | www.heart.org/HEARTORG

Genetic and Rare Disease Information Center: | rarediseases.info.nih.gov

References

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