Understanding Genetics

Genetics

DNA is like our body's instruction manual, and it's found in every cell. Inside our DNA, there are many genes, each with a special job to help keep us healthy. For our heart, there are hundreds of genes that make sure it can pump blood to the rest of our body.

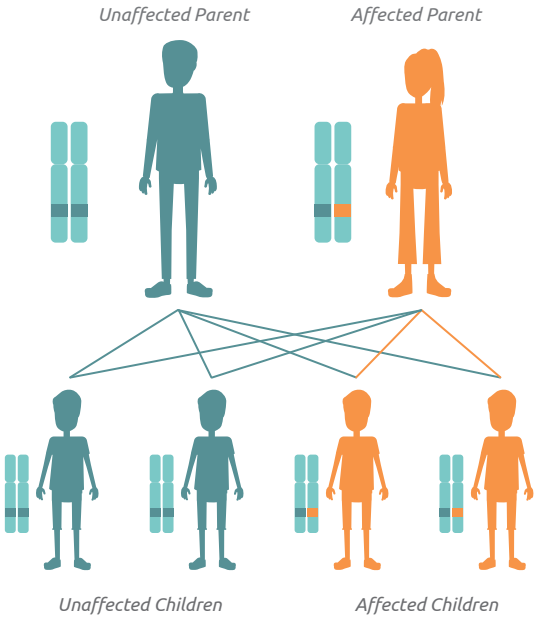
Everyone has small differences in their genes, and most of these differences don't cause problems. But sometimes, these changes—called **mutations** or **pathogenic variants**—can stop a gene from working properly. When this happens in one of our heart genes, it can affect how our heart works.

Inheritance

Genes can be passed down from parents to children. In fact, we typically have two copies of each gene, one from each parent. Most commonly, genetic risks for heart disease are passed down through autosomal dominant and recessive inheritance patterns *(see below)*. However, sometimes mutations are not passed down from a parent, but are new in a child.

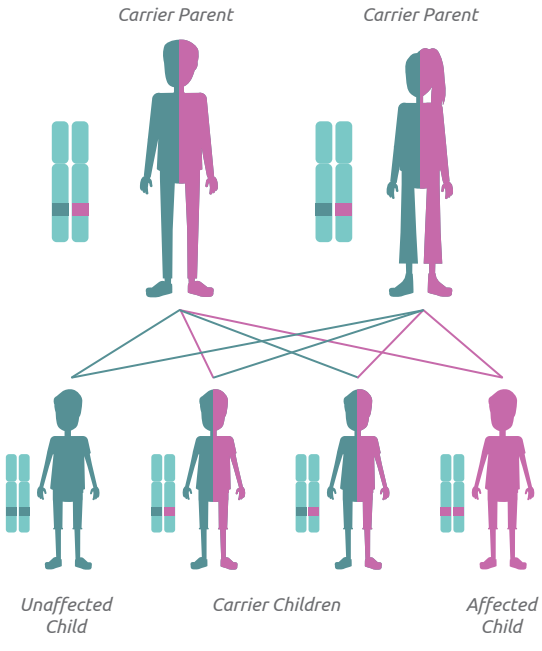
AUTOSOMAL DOMINANT
Inheritance

A mutation in **one copy of a gene** may increase the risk of heart disease.



AUTOSOMAL RECESSIVE
Inheritance

Mutations in **both copies of the gene** are needed to increase the risk of heart disease.



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Genetic Testing

Genetic testing can be done through a cheek swab, saliva sample, or blood test. A laboratory studies your DNA to see if there are any mutations that could lead to heart disease. Genetic testing can help your doctors better understand your heart.

Once a mutation is found in someone with heart problems, family mutation testing can be used to see who else in the family may be at risk. Family members who have the same mutation can have heart screening before symptoms develop. This can help diagnosis heart problems earlier and keep the heart muscle healthy.

When considering genetic testing, it is important to know the types of results you can get. See below.

+	VUS	−
POSITIVE ***Pathogenic or disease causing*** A mutation was found, which may explain the heart disease in you and/or your family.	**VARIANT OF UNCERTAIN SIGNIFICANCE (VUS)** ***Not enough evidence or data*** A small difference, or variant was found, but the relationship of the change to your heart disease is unclear. More information is needed to know if a VUS does, or does not, increase risk for heart disease. Remember, not all genetic changes are harmful.	**NEGATIVE** ***Benign or not disease causing*** No mutations were found in the studied genes.

Genetic Counseling

Genetic Counselors are healthcare providers that specialize in genetics and work closely with your Cardiologist. They can talk to you and your family about genetic testing options, types of results, and questions or concerns you may have. Genetic counselors will also ensure you understand what the results mean for you and your family.

When meeting with a Genetic Counselor, there is no obligation to get genetic testing. They will walk you through the benefits and limitations and help you make the best decision for your family.

Please ask your Cardiologist if you would like to meet with a Genetic Counselor.

Genetic Information Nondiscrimination Act (GINA)

GINA is a federal law that helps protect you from being treated unfairly because of your genetic test results. GINA makes it illegal for most employers and health insurance companies to discriminate based on genetic information. However, it doesn't cover life insurance, long-term care insurance, or disability insurance.

For people who have been diagnosed with heart disease, genetic testing is unlikely to affect insurance decisions. But for healthy family members, it might be something to think about before deciding to take a genetic test