Genetics 101

Good Start Genetics Customer Care
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What are genes?

Our bodies are made up of billions of cells. Inside each cell are chromosomes made up of DNA, which provides the instructions for how our bodies grow and develop. Our DNA, in turn, is packaged into small segments called genes, and each gene plays a unique role in how our bodies develop and function.

How are genes inherited?

Everyone has two copies of each chromosome an, therefore, two copies of each gene. With each pregnancy, one copy of each chromosome is passed on to the child from the mom (egg) and the other copy comes from the dad (sperm).

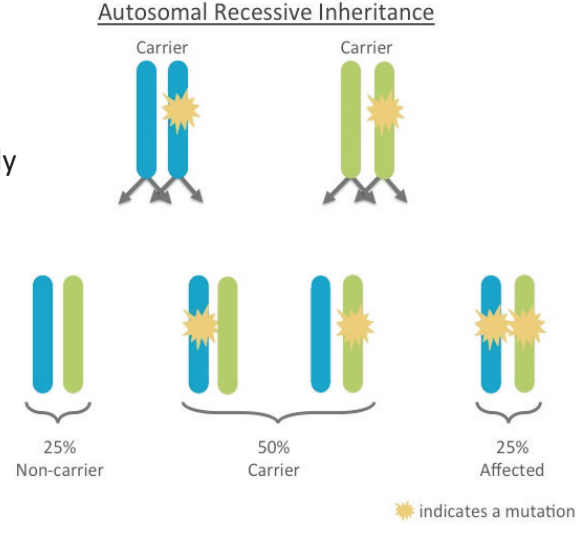
What is a mutation?

Mutations are changes in a gene that cause it not to work properly. Everyone has some genes that have mutations in them; however, for autosomal recessive disorders – the type of disorders carrier screening is recommended for – carrying a mutation in only one copy of a gene typically does not cause disease because the other copy of the gene still works properly.

What is a carrier?

A carrier is someone who is known to have a mutation in one of their genes. Carriers typically do not have symptoms of the disorder, so they do not know they are a carrier until after they have carrier screening.

When two carriers of the same disorder have a child together, there is a 25% chance the child will inherit the mutation from BOTH parents and therefore have that disorder. This is shown in the graphic to the right.



Carrier Screening Basics

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What is carrier screening?

Carrier screening is a blood test to see if a person is a carrier of a severe genetic disorder that could affect their children.

Why is carrier screening recommended?

Though everyone carries mutations in some of their genes, there are certain genetic disorders that are common in certain populations, and it is recommended that people in these populations be screened for these disorders. Two professional organizations, the American Congress of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG), make recommendations for which disorders should be screened.

How is carrier screening done?

The first step is to determine which disorders are appropriate to test. This is typically based on a persons ethnic background as well as their family history. Your physician will draw one or two tubes of blood to send to the laboratory for testing. It typically takes 2 weeks to get the results of your carrier screen(s).

What are the possible results?

Most likely, the results will be negative, meaning that the likelihood of being a carrier is greatly reduced. However no carrier screen detects all mutations so there is always a small residual risk of being a carrier, even after a negative test.

A positive test result means you are a carrier. This typically does not affect your individual health. However this does mean that you are at increased risk of having a child with the disorder for which you are a carrier. The next step is to test your reproductive partner. Most likely, they will not be a carrier of the same disorder and the chance of having a child with that disorder will be reduced. If both reproductive partners are carriers of the same disorder, they are at risk of having an affected child and can discuss reproductive options with a genetic counselor or qualified health care provider.