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Understanding Genetics and Your Health

Humans have between 20,000 and 25,000 genes. Most genes are the same in all people. But small differences in these genes give you a one-of-a-kind look and contribute to your personality and talents. Genes also can affect your health. To understand how, it's helpful to learn what genes do.

Genes: Your body's blueprint

Genes, which are made up of DNA, contain the instructions your body's cells need to function. Genes are located on structures called chromosomes. Information from your genes is used to make proteins. Each cell contains thousands of proteins. Each protein has a specific job to do at a specific time for the cell to work properly.

Sometimes, a gene can have something wrong with it. This is called a gene mutation or a mutated gene. A mutation causes the gene to give the wrong instructions for making a protein, so that the protein works improperly or is missing. If the mutation affects a protein that plays an important role in the body, a medical problem could result. Most gene mutations have no effect on health or development.

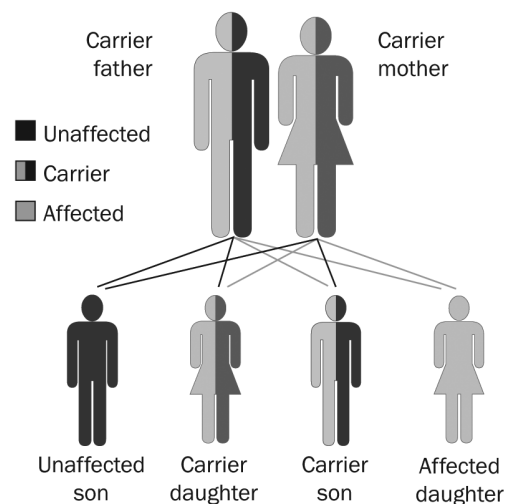
Genetic disorders: The basics

The genes you are born with can affect your health in these ways:

- **Single gene disorders are caused by a mutation in one gene.** There is a pattern to the way these genetic disorders

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How sickle cell anemia runs in families



Two unaffected parents each carry one copy of a gene mutation for sickle cell anemia. They have one child with sickle cell anemia and three unaffected children. Two of the unaffected children inherit one copy of the gene mutation — as carriers, they can pass the sickle cell trait on to their children.

We are learning more and more about the role specific genes play in our health. For example, you may have heard about "breast cancer genes," called BRCA genes. All people have BRCA genes. But only women and men born with mutated BRCA genes are at higher risk of breast cancer. Still, some women born with mutated BRCA genes don't get breast cancer. And most women who get breast cancer are born with normal BRCA genes. Keep in mind that genes themselves do not cause disease, but that mutated genes may cause health problems.



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ders show up in families. Even though the mutated gene is passed down from parent to child, not all family members are affected. Some members are “carriers” of the mutated gene. Examples of single-gene disorders are cystic fibrosis (SISS-tik-feye-BROH-suhss) and sickle cell anemia (uh-NEE-mee-uh).

- **Chromosome disorders occur when all or part of a chromosome is missing or extra, or if the structure of one or more chromosomes is not normal.** This can affect some of the genes. Most chromosome disorders involving whole chromosomes do not run in families. Genes can influence your risk of getting some diseases, such as breast cancer, heart disease, diabetes, and thyroid conditions. But other factors, such as lifestyle and environment, also play a role in developing these conditions. Rarely, single genes are responsible for these diseases; however, most of the time they are due to a combination of genes and environment. The role genes play in developing these conditions often is not known, but our understanding of this continues to grow through research.

Some companies offer genetic tests that you can do yourself through the mail. These tests may not provide true or meaningful information. These tests might even provide harmful information to consumers. Talk to your doctor before using this type of test.

Genetic counseling: What it is and who needs it

Genetic counseling gives information and support to people who have, or may be at risk of, genetic disorders. Some reasons a family might seek genetic counseling are:

- a family history of a genetic condition, birth defect, chromosomal disorder, or cancer
- two or more pregnancy losses, a stillbirth, or a baby who died
- a child with a known inherited disorder, birth defect, mental retardation, or developmental delay
- a woman who is pregnant or plans to become pregnant at 35 years or older
- test results that suggest a genetic condition is present
- increased risk of getting or passing on a genetic disorder because of one’s ethnic background
- people related by blood who want to have children together

Your doctor can help you find a genetic professional if you might benefit from this service. During a consultation, the genetics professional meets with a person or family to discuss genetic risks or to diagnose, confirm, or rule out a genetic condition. Sometimes, a family chooses to have genetic testing. Most of the time, testing is used to find changes that are linked to genetic disorders. The results can confirm or rule out a condition. Tests also can help to know the chances that a person will get or pass on a genetic disorder. The genetics professional can help a family decide if genetic testing is the right choice for them. ■