The Basics on Genes and Genetic Disorders

Have people ever said to you, "It's in your genes"? They were probably talking about a physical characteristic, personality trait, or talent that you share with other members of your family.

We know that genes play an important role in shaping how we look and act and even whether we get sick. Now scientists are trying to use that knowledge in exciting new ways, such as treating health problems.

What Is a Gene?

To understand how **genes** work, let's review some biology basics. Most living organisms are made up of cells that contain a substance called deoxyribonucleic (pronounced: dee-AHK-see-rye-bow-noo-klee-ik) acid (DNA).

DNA contains four chemicals (adenine, thymine, cytosine, and guanine — called A, T, C, and G for short) that are strung in patterns on extremely thin, coiled strands in the cell. How thin? Cells are tiny — invisible to the naked eye — and each cell in your body contains about 6 feet of DNA thread, for a total of about 3 billion miles of DNA inside you!

So where do genes come in? Genes are made of DNA, and different patterns of A, T, G, and C code for the instructions for making things your body needs to function (like the enzymes to digest food or the pigment that gives your eyes their color). As your cells duplicate, they pass this genetic information to the new cells.

DNA is wrapped together to form structures called **chromosomes**. Most cells in the human body have 23 pairs of chromosomes, making a total of 46. Individual sperm and egg cells, however, have just 23 unpaired chromosomes. You received half of your chromosomes from your mother's egg and the other half from your father's sperm cell. A male child receives an X chromosome from his mother and a Y chromosome from his father; females get an X chromosome from each parent.

Genes are sections or segments of DNA that are carried on the chromosomes and determine specific human characteristics, such as height or hair color. Because you have a pair of each chromosome, you have two copies of every gene (except for some of the genes on the X and Y chromosomes in boys, because boys have only one of each).

Some characteristics come from a single gene, whereas others come from gene combinations. Because every person has about 25,000 different genes, there is an almost endless number of possible combinations!

Genes and Heredity

Heredity is the passing of genes from one generation to the next. You inherit your parents' genes. Heredity helps to make you the person you are today: short or tall, with black hair or blond, with brown eyes or blue.

Can your genes determine whether you'll be a straight-A student or a great athlete? Heredity plays an important role, but your environment (including things like the foods you eat and the people you interact with) also influences your abilities and interests.

A person can have changes (or mutations) in a gene that can cause many issues for them. Sometimes changes cause little differences, like hair color. Other changes in genes can cause health problems.

Mutations in a gene usually end up causing that particular gene copy to not do its job the way it normally should. Since we have two copies of every gene, typically there's still a "normal" working copy of the gene. In these cases, usually nothing out of the ordinary happens since the body can still do the jobs it needs to do. This is an example of an **autosomal recessive trait**.

For someone to have a recessive disease or characteristic, the person must have a gene mutation in *both* copies of the gene pair, causing the body to not have working copies of that particular gene.

Genes can be either **dominant** or **recessive**. Dominant genes show their effect even if there is just one mutation in one copy of that gene pair; the one mutation "dominates" the normal back-up copy of the gene, and the characteristic shows itself.

A person can be born with gene mutations, or they can happen over a lifetime. Mutations can occur when cells are aging or have been exposed to certain chemicals or radiation. Fortunately, cells usually recognize these types of mutations and repair them by themselves. Other times, however, they can cause illnesses, such as some types of cancer.

If the gene mutation exists in egg or sperm cells, children can inherit the gene mutation from their parents. When the mutation is in every cell of the body (meaning a child was born with it), the body is not able to "repair" the gene change.

What Are Genetic Disorders?

Researchers have identified more than 4,000 diseases that are caused by mutations. But having a genetic mutation that may cause a disease or condition doesn't always mean that a person will actually develop that disease or condition.

On average, people probably carry from 5 to 10 genes with mutations in each of their cells. Problems happen when the particular gene is dominant or when a mutation is present in both copies of a recessive gene pair. Problems can also happen when several variant genes interact with each other — or with the environment — to increase susceptibility to diseases.

If a person has a change in a dominant gene that is associated with a particular condition, he or she will usually have features of that condition. And, each of the person's children will have a 1 in 2 (50%) chance of inheriting the gene and developing the same features. Diseases and conditions caused by a dominant gene include achondroplasia (pronounced: ay-kon-druh-PLAY-zhuh, a form of dwarfism), Marfan syndrome (a connective tissue disorder), and Huntington disease (a degenerative disease of the nervous system).

People who have a change in just one copy of a recessive gene are called "carriers." They don't usually have the disease because they have a normal gene copy of that pair that can do the job. When two carriers have a child together, however, the child has a 1 in 4 (25%) chance of getting a gene with a mutation from both parents, which would result in the child having the disease. Cystic fibrosis (a lung disease), sickle cell anemia (a blood disorder), and Tay-Sachs disease (which causes nervous system problems) are caused by recessive mutations from both parents coming together in a child.

With recessive gene mutations on the X chromosome, usually only guys can develop the disease because they have only one X chromosome. Girls have two X chromosomes — since they have a back-up copy of another X chromosome, they don't always show features of X-linked conditions. These include the bleeding disorder hemophilia (pronounced: hee-muh-FIL-ee-uh) and color blindness.

Sometimes when an egg and sperm unite, the new cell gets too many or too few chromosomes, which can cause issues for the child. For example, most children born with Down_syndrome have an extra chromosome number 21.

In some cases, people who are concerned that they might carry certain variant genes can have genetic testing so they can learn their children's chances of inheriting a disease. Pregnant women can also have tests done to see if the fetus they are carrying might have certain genetic illnesses. Genetic testing usually involves taking a sample of someone's blood, skin, or amniotic fluid and checking it for genetic changes.

Changing Genes

Sometimes scientists alter genes on purpose. For many years, researchers have altered the genes in plants to produce other plants with special characteristics, such as an increased resistance to disease and pests or the ability to grow in difficult environments. We call this **genetic engineering**.

Gene therapy is a promising new field of medical research. In gene therapy, researchers try to supply copies of healthy genes to cells with variant or missing genes so that the "good" genes will take over. Viruses are often used to carry the healthy genes into the targeted cells because many viruses can insert their own DNA into targeted cells.

But there are problems with gene therapy. Scientists still don't quite know what every gene in the human body does. Huge scientific efforts like The Human Genome Project and related projects have completed a map of the entire human genome (all of the genetic material on a living thing's chromosomes), but it will take many more years to find out what each gene does and how they interact with one another. For most diseases, scientists don't know if and how genes play a role. Plus, there are major difficulties inserting the normal genes into the proper cells without causing problems for the rest of the body.

There are also concerns that people might try changing genes for ethically troubling reasons, such as to make smarter or more athletic children. No one knows what the long-term effects of that kind of change would be.

Still, for many people who have genetic diseases, gene therapy holds the hope that they — or their children — will be able to live better, healthier lives.

Reviewed by: Angela L. Duker, MS, CGC
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