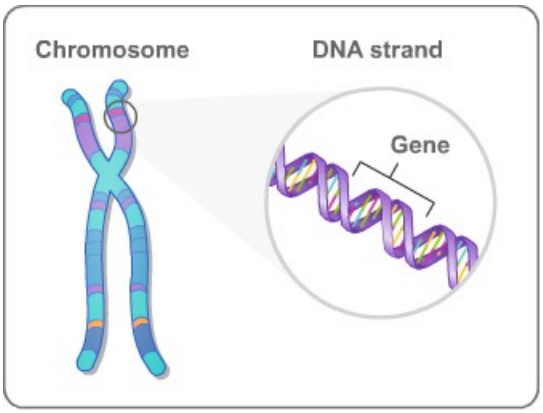
Understanding Genetics: Part 1 of 2

Most people know that genetics has to do with heredity: the act of passing on individual or family characteristics to the next generation. However, exactly how this happens in the body and how this process is influenced by the environment are not as commonly understood.

To help interpret genetic information, we are presenting a two-part feature that explains the basic mechanisms of genetic transmission. This first article in the series focuses on mechanisms, the second, on the relation of genetic irregularities to disease and conditions that affect children.

Part One: Genes, chromosomes, heredity, and the influence of the environment.

What are Genes?



Chromosomes are made up of tightly packed strands of DNA and other proteins. A gene is a portion of a DNA strand.

What is genetics?

Genetics is the area of biology that studies heredity and spontaneous changes in genetic material. Heredity refers to the passing of features, such as eye colour, from parent to child.

In health care, genetics is important because some diseases, or the risk of getting some diseases, can be passed from parent to child or are caused by a spontaneous change in genetic material.

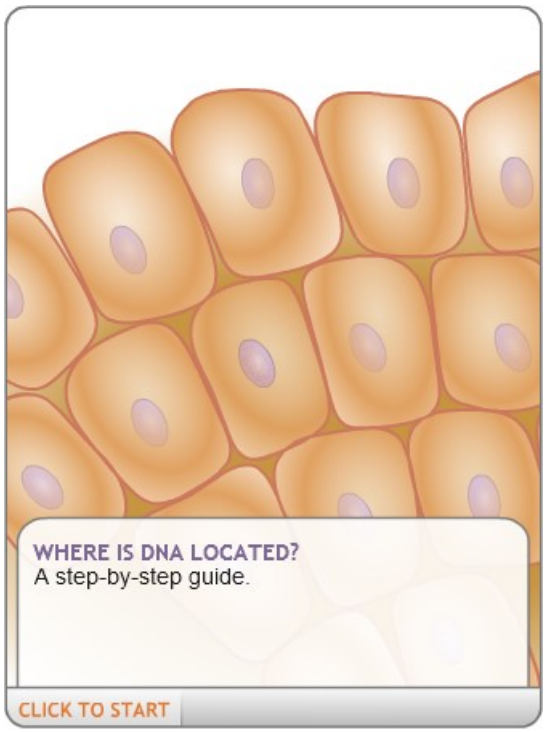
As far back as Aristotle in Ancient Greece, scientists understood that children were like their parents because certain features were passed from generation to generation. In 1860, Gregor Mendel discovered rules about how some features

were passed from generation to generation by studying pea plants. The rules he discovered are still useful for understanding how features that are controlled by single genes are passed from parent to child.

Today, genetic research holds the promise of finding a cure for diseases that are caused or influenced by genes.

What are genes?

Where is DNA located?



Heredity is controlled by genes. Genes are the basic units of heredity, and are made of lengths of DNA, which stands for deoxyribonucleic acid. DNA is a complex molecule that carries the information, or genetic code, that will determine many features of a living thing.

There are about 30,000 genes in humans. This entire collection of genes is known as the human genome. The Human Genome Project, a major research project begun in 1990 and funded by the U.S. government, succeeded in creating a map of all the human genes in 2001. Every person has a unique set of genes. This is known as the *genotype* of that person. *Phenotype* refers to the features and qualities of a living thing that come from the genotype influenced by the environment.

DNA is organized in chromosomes, structures that are found in every cell in our body. Each chromosome has a single long DNA molecule. Cells are the basic building blocks of living things. Within each cell, there are 23 chromosome pairs. One chromosome in each pair comes from the mother, and the other comes from the father. Twenty-two of these chromosome pairs, numbered from one to 22 based on their features, including size, are the same for males and females.

The 23 rd pair of chromosomes is called sex chromosomes. These are different in females and males. Females have two X chromosomes (XX). Males have an X chromosome and a Y chromosome (XY). Except for the sex chromosomes in males, both chromosomes in a pair contain genes with the same function in the same order, but one copy of each gene comes from the mother and one comes from the father.

Another structure in each cell, the mitochondria, also contains DNA. Mitochondria are structures that make energy for the cells by way of a

chemical called adenosine triphosphate (ATP). Genes in the mitochondria, as well as in the nucleus of the cell, direct the cells to make the enzymes needed for ATP production. The DNA in mitochondria is inherited only from the mother. Mutations in this DNA have been related to some hereditary disorders.

What do genes do?

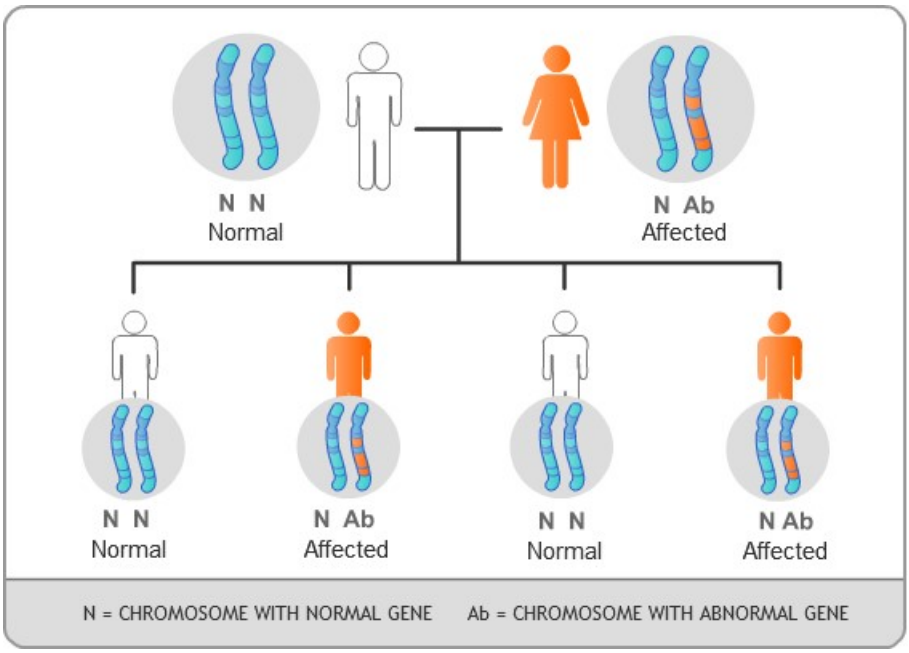
Genes control the making of a protein or help control the activity of other genes. Proteins are very important molecules for living things. There are many types of proteins that do many things. Some form the parts of the body, while some help control functions of the body.

The activity of a gene is affected by the environment around it. When a baby is growing during pregnancy, the protein controlled by a gene can control a particular feature. Some features are controlled by one gene, but many features are controlled by a number of genes acting together under the influence of the environment at many levels.

Genes are always found in pairs that control the making of the same protein, or the same feature. In some cases, one gene of the pair will control the feature. In this case the gene that controls the feature is called the dominant gene, and the other gene in the pair is called the recessive gene. If there are two copies of the recessive gene, the feature is controlled by the recessive gene.

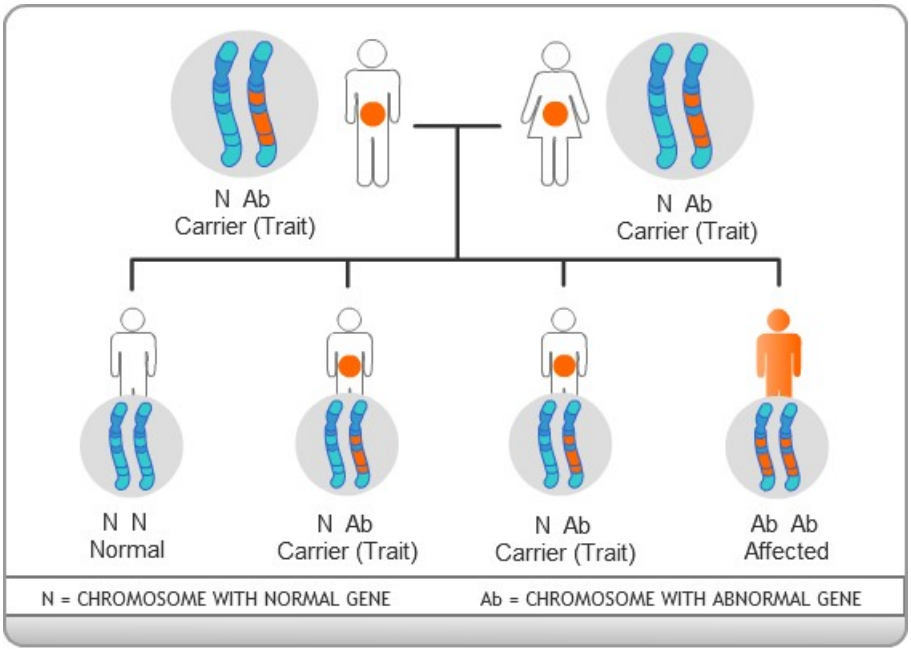
There are more than two types, or alleles of many genes. Most of these alleles are normal, and do not cause problems. Some types that have come from a change called mutation can cause disease. Many of the genes that cause disease are recessive. If a baby has only one copy of a recessive gene that causes a disease, she will not have the disease. But if the disease is caused by a dominant gene, one copy is sufficient to produce the disease.

Dominant Inheritance



Some genes that cause disease are dominant. In this example, only one parent carries the dominant gene (Ab) for the disease. A baby will only have to inherit one copy of the dominant gene in order to be affected by the disease.

Recessive Inheritance



Many genes that cause disease are recessive. In this example, both parents are carriers of the recessive gene (Ab) for the disease . If a baby inherits only one copy of the the recessive gene, she will not have the disease. If a baby inherits two copies of the recessive gene, she will be affected by the disease. If a baby inherits neither copy of the recessive gene, she will not be affected and will not be a carrier of the recessive disease.

Genes and the environment

It is important to realize that just as genes influence physical characteristics and behaviour, characteristics of the individual, the environment, and the way the

individual behaves influence and regulate the expression of genes. With very few exceptions, most physical traits, behaviours, and susceptibilities to disease are influenced by not just one, but a number of genes.

But genes by themselves are not destiny. Their ultimate effect is the consequence of a complex series of interactions between genes and the environment, as noted above. The idea of hereditability, expressed as a ratio or a percentage, captures this idea. Among humans, height is a highly heritable characteristic. Around seventy-six per cent of the variation in height is due to genetic factors. The balance of the variation in height, 24%, is related to environmental factors and their interaction with genes.

Please read about mutation, birth defects, and genetic testing and counselling in the second article of this two-part feature.

1/17/2011

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