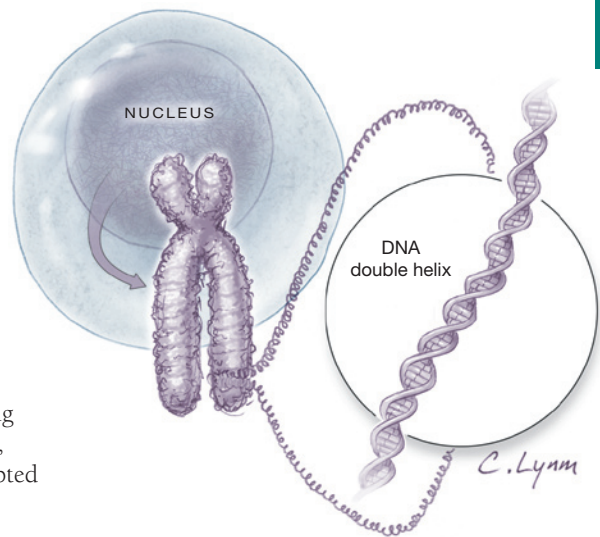


Genetics: the Basics

Genetics includes the study of how human characteristics are inherited from one's parents. It explains how traits as simple as eye color or as complex as susceptibility to diseases run in families. **Genes** are units of heredity passed from parents to offspring and are contained in a person's cells—every human cell contains about 20,000 to 25,000 genes. Genes vary greatly from person to person and influence personality, intelligence, physical appearance, and other traits to a certain extent, but learning and environment play important roles as well. The March 19, 2008, issue of *JAMA* is a theme issue on genetics. This Patient Page is adapted from one published in the November 14, 2001, issue of *JAMA*.



GENETICS TERMINOLOGY

- **DNA** (deoxyribonucleic acid) is the chemical inside a gene that carries genetic instructions for making living things. DNA is made of 2 long, twisting molecules called the double helix.
- **Chromosomes** are packets of genes in a cell. Humans have 23 pairs (46 total). One member of each pair of chromosomes is inherited from the mother and the other from the father. Two of the 46 chromosomes (X and Y) are sex chromosomes that determine whether offspring will be male or female. Individuals with a pair of X chromosomes are female and those with an X chromosome and a Y chromosome are male.
- A **genome** is an entire system of genes. **Genomics** is the study of how genes interact and influence the biology and physical characteristics of living things.
- **Mutations** are changes in DNA. These changes are sometimes passed on to offspring.
- **Genetic disorders** are diseases or disorders caused by gene mutations or chromosomal defects.

GENETIC DISORDERS AND INHERITANCE

Heritability is the degree to which a characteristic is determined by a person's genes. Many diseases have a genetic cause. Types of genetic disorders are described below.

- **Familial disorders** are those that affect more than 1 person in a family. There are times, however, when a child is born with an unexpected genetic disorder without a known family history of the disorder.
- **Chromosome abnormalities** can cause some genetic disorders, which occur when a child is born with an abnormal number of chromosomes or extra or missing pieces of chromosomes.
- **Unifactorial diseases** can occur when certain defects in a gene or a pair of genes are present. Sickle cell anemia and cystic fibrosis are 2 examples of unifactorial diseases.
- **Multifactorial diseases** are those that involve a number of different genes that, combined with environmental factors, can cause disorders such as asthma and diabetes.

If you or your partner is concerned about a disorder that seems to run in either of your families, you may want to see a genetic counselor before having children. You can often be tested to see if your or your partner's genes carry certain disorders.

FOR MORE INFORMATION

- Human Genome Project information genomics.energy.gov
- Centers for Disease Control and Prevention www.cdc.gov/genetics/info.htm

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To find this and previous JAMA Patient Pages, go to the Patient Page link on JAMA's Web site at www.jama.com.

Sources: American Medical Association Encyclopedia of Medicine, American Medical Association Family Medical Guide, Centers for Disease Control and Prevention, Human Genetics Programme (World Health Organization), National Human Genome Research Institute, Public Health Genetics Unit

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