



Genetics

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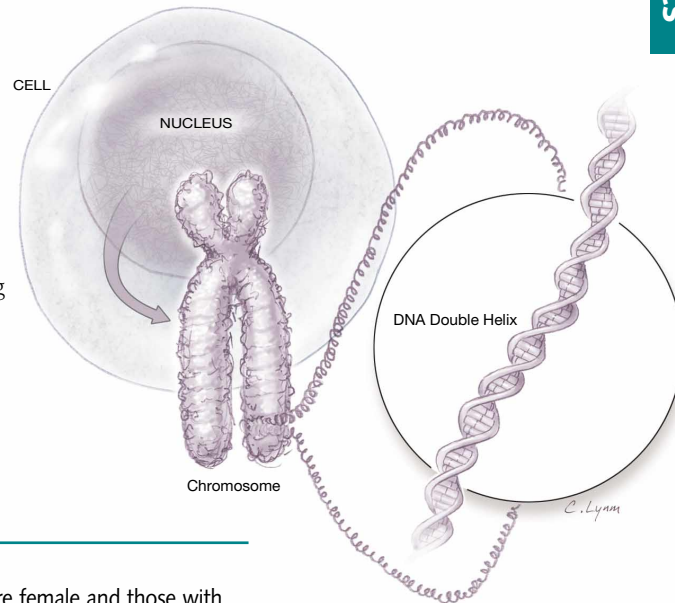
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Genetics

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 approximately 30 000 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 an important role as well. The November 14, 2001, issue of *JAMA* is dedicated to genetics.



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, 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

- Centers for Disease Control and Prevention
800-311-3435
www.cdc.gov/genetics/info.htm
- National Human Genome Research Institute (National Institutes of Health)
www.nhgri.nih.gov/DIR/VIP/Glossary
- Public Health Genetics Unit (University of Cambridge)
www.medinfo.cam.ac.uk/phgu/info_database/glossary.asp

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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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