

Glossary of Terms

(adapted from GeneTests Illustrated Glossary
and National Human Genome Research Institute Talking Glossary of Genetic Terms)

A C D E F G H J L M O P

A

allele: (ă-lēl') One of the variant forms of a gene at a particular locus on a chromosome. Different alleles produce variation in inherited characteristics such as hair color or blood type. In an individual, one form of the allele (the dominant one) may be expressed more than another form (the recessive one).

ambiguous: (am-big'ū-ŭs) uncertain, having more than one interpretation.

amino acids: (ă-mē'nō as'ids) a group of 20 different kinds of small molecules that link together in long chains to form proteins. Often referred to as the "building blocks" of proteins. Examples include methionine, tryptophan, threonine, lysine, phenylalanine; branch chain amino acids: valine, isoleucine, leucine.

autosomal dominant: (aw-tō-sō'măl dom'i-nant) Describes a trait or disorder in which the phenotype is expressed in those who have inherited only one copy of a particular gene mutation (heterozygotes); specifically refers to a gene on one of the 22 pairs of autosomes (non-sex chromosomes) Examples of autosomal dominant diseases include Huntington's disease, neurofibromatosis, and polycystic kidney disease.

autosomal recessive: (aw-tō-sō'măl ri-'ses-iv) Describes a trait or disorder requiring the presence of two copies of a gene mutation at a particular locus (location) in order to express observable phenotype; specifically refers to genes on one of the 22 pairs of autosomes (non-sex chromosomes); When both parents are carriers, there is a one-in-four (or 25%) chance that both will pass the changed gene on to a child, causing the child to be born with the condition. There also is a one-in-four (or 25%) chance that they will each pass on a normal gene, and the child will be free of the condition. There is a two-in-four (or 50%) chance that a child will inherit a changed gene from one parent and a normal gene from the other, making it a carrier like its parents. These chances are the same in each pregnancy with the same parents. Examples of autosomal recessive conditions include sickle cell anemia, phenylketonuria, and galactosemia.

C

carrier: (kăr'ē ěr) an individual who has a recessive, disease-causing gene mutation at a particular locus on one chromosome of a pair and a normal allele at that locus (location) on the other chromosome; may also refer to an individual with a balanced chromosome rearrangement. Although carriers are not affected by the disease, two carriers can produce a child who has the disease.

cataracts: (kat'ă-rakts) complete or partial cloudy area on the eye lens.

chromosome: (krō'mō-sōm) A physical structure in a cell consisting of DNA and supporting proteins called chromatin. Human cells normally contain 46 chromosomes identified as 23 pairs; 22 pairs are autosomes and one pair are the sex chromosomes.

congenital: (kon-jen'i-tāl) Any trait or condition that exists from birth, but not necessarily genetic.

constipation: (kon-sti-pā'shūn) A condition in which bowel movements are infrequent or incomplete.

D

DNA: synonym deoxyribonucleic acid (dē-oks'ē-rībō-nū-klē'ik as'id); the molecule which encodes the genes responsible for the structure and function of an organism and allows for transmission of genetic information from one generation to the next.

E

enzyme: (en'zīm) a protein that controls a biochemical reaction, usually speeding it up. Organisms could not function if they had no enzymes.

F

fatty acids: (fat' ē as'ids) any acid derived from fats through a chemical reaction called hydrolysis, they accumulate in certain disorders.

fetal: (fē'tāl) relating to a fetus; stage of development after the eighth week of pregnancy.

G

gene: (jēn) the basic unit of heredity, consisting of a segment of DNA arranged in a linear manner along a chromosome, which codes for a specific protein or segment of protein leading to a particular characteristic or function.

genotype: (jēn-ō-tīp) The genetic identity of an individual that does not show as outward characteristics; also refers to the specific set of alleles inherited at a locus.

H

hormones: (hōr'mōns) chemical substances formed in one organ or part of the body and carried in blood to another organ or part of the body; depending on the specific type and function, can alter function. Examples include thyroid hormones, adrenal hormones, pituitary hormones.

J

jaundice: (jawn'dis) a condition of increased levels of bilirubin (a byproduct of the normal breakdown of red blood cells by the liver) which gives a yellowish tinge to the skin and whites of the eyes.

L

locus: (lo'kūs) the place on a chromosome where a specific gene is located, a kind of address for the gene. The plural is "loci," not "locuses".

M

metabolite: (mě-tab'ō-līt) Any product (intermediate, waste product) of a chemical change or break down.

mutation: (mū-tā'-shŭn) Any alteration in a gene from its natural state; may be benign (commonly referred to as a "polymorphism"), pathogenic (able to cause disease), or of unknown significance.

O

organic acids: (ōr-gan'ik as'ids) non amino acids containing the chemical element carbon.

P

phenotype: (fē'nō-tīp) The observable physical and/or biochemical characteristics of the expression of a gene; the clinical presentation of an individual with a particular genotype.

pigmentation: (pig-men- tā'-shŭn) coloration of the skin or tissue.

protein: (prō' tēn) a large complex molecule made up of one or more chains of amino acids. Proteins perform a wide variety of activities in the cell. All structure and functions in the body depend on protein.