Glossary of Common Terms in Genetics

Acquired mutations Gene changes that arise within individual cells and accumulate throughout a person's life span.

Alleles One of a group of genes that occur alternatively at a given locus. A single allele is inherited separately from each parent (e.g., at a locus for eye color, the allele might result in blue or brown eyes).

Base pair Two nitrogenous bases (adenine and thymine or guanine and cytosine) held together by weak bonds. Two strands of DNA are held together in the shape of a double helix by the bonds between base pairs.

Carrier A person who has a recessive mutated gene along with its normal allele. Carriers do not usually develop disease caused by the mutation, but they can pass the mutated gene on to their children.

Chromosomes The self-replicating genetic structures of cells containing the cellular DNA that bears the linear array of genes in its nucleotide sequence.

Cloning Using specialized DNA technology to produce multiple, exact copies of a single gene or other segment of DNA to obtain enough material for further study. A second type of cloning exploits the natural process of cell division to make many copies of an entire cell. A third type of cloning produces complete, genetically identical animals, such as Dolly the sheep.

DNA (deoxyribonucleic acid) A double-stranded molecule that encodes genetic information. DNA is held together by weak bonds between base pairs of nucleotides: adenine, guanine, cytosine, and thymine.

Gene The fundamental unit of heredity. A gene is an ordered sequence of nucleotides located in a particular position on a particular chromosome that encodes a specific functional product (i.e., a protein or RNA molecule).

Gene expression The process by which a gene's coded information is converted into the structures present and operating in the cell.

Gene mapping Determination of the relative positions of genes on a DNA molecule and the distance between them.

Gene therapy Insertion of normal DNA directly into cells to correct a genetic defect.

Huntington's disease An adult-onset disease caused by an inherited dominant gene mutation that is characterized by progressive mental and physical deterioration.

Linkage analysis A gene-hunting technique that traces patterns of heredity in large, high-risk families in an attempt to locate a disease-causing gene mutation.

Locus The position on a chromosome of a gene or other marker, or the DNA at that position.

Marker An identifiable physical location on a chromosome whose inheritance can be monitored.

Multiplexing A sequencing approach that uses several pooled samples simultaneously, greatly increasing sequencing speed.

Mutation Any heritable change in DNA sequence.

Nucleotide A subunit of DNA or RNA consisting of a nitrogenous base, a phosphate molecule, and a sugar molecule. Thousands of nucleotides are linked to form a DNA or RNA molecule.

Oncogene One or more forms of a gene associated with cancer.

Polygenic disorders Genetic disorders resulting from the combined action of alleles of more than one gene (e.g., heart disease, diabetes, and some cancers). Although such disorders are inherited, they depend on the simultaneous presence of several alleles; thus the hereditary patterns are usually more complex than single-gene disorders.

Recombination The process by which children derive a combination of genes different from that of either parent.

Single-gene disorder Hereditary disorder caused by a mutant allele of a single gene (e.g., Duchenne muscular dystrophy, retinoblastoma, sickle cell disease).

Tay-Sachs disease An inherited disease caused by a recessive gene mutation that appears in infancy and is characterized by profound mental retardation and early death.

Definitions adapted from The Human Genome Project, http://www.ornl.gov/hgmis/publicat/glossary.html.

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