# Genetic Terms and Definitions

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World Health Organization Glossary : http://www.who.int/genomics/glossary/en/index.html
National Human Genome Research Institute "Talking Glossary": http://www.genome.gov/glossary/index.cfm

| Term | Definition | Expansion |
|---|---|---|
| **Allele** | One of two or more versions of a gene. | An individual inherits two alleles for each gene, one from each parent. If the two alleles are the same, the individual is homozygou for that gene. If the alleles are different, the individual is heterozygous. |
| **$BRCA_1$/$BRCA_2$** | BRCA1 and BRCA2 are the first two genes found to be associated with inherited forms of breast cancer. | Both genes normally act as tumor suppressors, meaning that they help regulate repair errors in copying genes. When these genes are rendered inactive due to mutation, errors are not corrected, which can lead to breast cancer. |
| **Chromosome** | An organized package of DNA found in the nucleus of the cell. | Humans have ...22 pairs of numbered chromosomes... and one pair of sex chromosomes, X and Y. Each parent contributes one chromosome to each pair so that offspring get half of their chromosomes from their mother and half from their father. |
| **Cloning** | The process of making identical copies of an organism, cell, or DNA sequence. | A process by which scientists amplify a desired DNA sequence. The target sequence is inserted into another DNA molecule and introduced into a host cell. Each time the host cell divides, it replicates the foreign DNA sequence along with its own DNA. |
| **Deletion** | A type of mutation involving the loss of genetic material. | (Deletions) can be small, involving a single missing DNA base pair, or large, involving a piece of a chromosome. Just by having a deletion of one base pair, you can have the most severe birth defect, and sometimes by missing an entire chromosome, you don't even see all that much. |
| **DNA** | Deoxyribonucleic Acid - the chemical name for the molecule that carries genetic instructions in all living things. | Consists of two strands that wind around one another to form a shape known as a double helix. Each strand has a backbone made of alternating sugar (deoxyribose) and phosphate groups. Attached to each sugar is one of four bases--adenine (A), cytosine (C), guanine (G), and thymine (T). |
| **DNA Sequencing** | A laboratory technique used to determine the exact sequence of bases (A, C, G, and T) in a DNA molecule. | Sequence(s) carr(y) the information a cell needs to assemble protein and RNA molecules. DNA sequence information is important t scientists investigating the functions of genes. The technology was made faster and less expensive as a part of the Human Genome Project. |
| **Gene** | The basic physical unit of inheritance. | Genes are passed from parents to offspring and contain the information needed to specify traits. Genes are arranged, one after another, on structures called chromosomes. |
| **Genome** | The entire set of genetic instructions found in a cell. | In humans, the genome consists of 23 pairs of chromosomes, found in the nucleus, as well as a small chromosome found in the cells' mitochondria. These chromosomes contain approximately 3.1 billion bases of DNA sequence. |
| **Genetic Marker** | A DNA sequence with a known physical location on a chromosome. | Genetic markers can help link an inherited disease with the responsible gene. DNA segments close to each other on a chromosome tend to be inherited together. (They) are used to track the inheritance of a nearby gene that has not yet been identified, but whos approximate location is known. |
| **Pharmacogenomics** | A branch of pharmacology concerned with using DNA and amino acid sequence data to inform drug development and testing. | An important application of pharmacogenomics is correlating individual genetic variation with drug responses. |
| **Single nucleotide polymorphisms (SNPs)** | A type of polymorphism involving variation of a single base pair. | Scientists are studying how single nucleotide polymorphisms, or SNPs (pronounced "snips"), in the human genome SNPs correlate with disease, drug response, and other phenotypes. |