The Genetic Science Glossary

Adenine (A) One of the four bases, or building blocks, found in DNA. A d e n i n e Analogy: Adenine is one of the letters in the four-letter DNA alphabet. Adenine (A), Guanine (G), Thymidine (T), and Cytidine (C) are the four different molecules base molecules that compose DNA. The smallest unit of DNA consists of one base molecule, one sugar molecule, and one phosphate molecule. Since the DNA molecule is a double helix, each base must be paired with one other base to form the "rungs of the ladder" that makes up the helix. Adenine is always paired with Thymidine and Guanine with Cytidine. When a cell prepares to divide, the DNA unwinds its helix and each strand of DNA is seen as a linear series of codons (ex: ACC; GTC; AAT, etc). Each codon consists of three base molecules that form the code linked to a particular amino acid (see definition of codon below). The sequence of different combinations of codons on the chromosomes determines what amino acids will be incorporated into specific proteins according to a specific order. Thus, the DNA determines the way proteins are built up for that person's cells.

Allele Alternate forms of a gene. We receive two copies of each gene, one copy from each parent. Some genes have more than one form of a trait (e.g., brown eyes or blue eyes). These alternative forms of a gene are called alleles. Allele analogy: Genes are like recipes for proteins. An allele is like a variation of that recipe. There could be a recipe for oatmeal cookies that has two variations: one with raisins and one without. A gene for skin colour could have two variations: one with freckles and one without.

Amino Acid There are 20 amino acids. Each amino acid is one of 20 kinds of building blocks that form proteins. Protein shape and function are determined by the combination of the amino acids. The order of bases in DNA, the genetic code, determines which amino acids make up each protein and in what order. If the nucleotide bases are the letters of the DNA alphabet, then each codon (three nucleotide combination) is a word. The amino acid is the meaning of the word. All the words together make a sentence, like all the amino acids together make a protein.

Anticipation	A genetic disorder tending to increase in severity and with earlier onset as passed on through generations.
Apoptosis	Programmed cell death
Artificial Selection	Evolution caused by humans through choosing and breeding specific organisms based on the expression of a desired trait. Example: If a farmer wants to grow bigger tomatoes, selecting and then planting only the seeds of the biggest tomatoes the farmer grows might select the tomatoes with the genes that cause them to be larger. This could result in a crop of tomatoes that are bigger.
Autosomal Dominant	A gene on the autosomes (non-sex chromosomes) that is always expressed even if only one copy is present. Example: The brown eye allele is autosomal dominant. If you have a gene for brown eyes and a gene for blue eyes, brown eyes will be expressed.
Autosome	Humans have 23 pairs of chromosomes: 46 chromosomes in total. An autosome is one of the 44 chromosomes that contain genetic information that does not determine sex. There are a total of 46 chromosomes, 44 autosomal plus two sex chromosomes (X and Y).
Base	One of the molecules, or building blocks, that form DNA and RNA. The bases consist of adenine, guanine, cytosine, and thymine. Base Analogy: If a genome is the encyclopedia containing all the information necessary to produce an organism, DNA is the alphabet in which the encyclopedia is written. DNA is a four-letter alphabet, and each base is one of those letters.
Base Pair	Two of the four bases that are held together along the double helix DNA molecule. The four bases are: adenine (A), thymine (T), cytosine (C), and guanine (G). A always binds to, or pairs with, T and C always binds to C.
Behavioural Genetics	The study of how genes may influence behavior.
Bioinformatics	Use of advanced computing techniques to analyze genomic data.
Blastocyst	A blastocyst is an embryo that has not yet implanted, containing about 150 cells. The blastocyst is a hollow ball consisting of an outer layer of cells that will specialize to become extra-embryonic tissues

such as the placenta, umbilical cord and amniotic sac. It also consists of an inner cell mass that will specialize to become all the cells of the developing fetus. It is from the inner cell mass of a blastocyst that embryonic stem cells are derived.

Blastomere A blastomere is any of the cells that form an early embryo, after the first division of a fertilized egg. For example, at the 8-cell stage, a single cell, a blastomere, can be removed for pre-implantation diagnosis. Additionally, blastomeres can he separated at an early stage and each blastomere may develop into a separate embryo.

Cancer Diseases involving the unregulated division of abnormal cells within the body. All cancer is genetic in the sense that mutations in the genes that regulate cell division are the cause of cancer.

Carrier Someone who has an unexpressed recessive genetic trait. Example: A person with brown eyes may be a carrier of the trait for blue eyes. Two brown-eyed parents who are carriers of blue eyes may have a child with blue eyes.

Catalyst A catalyst is something that speeds up a chemical reaction. Often enzymes catalyze chemical reactions.

Chimera An organism that has cells with different genetic makeup. These organisms may be transgenic, that is carrying gene sequences from more than one species.

Chromosome The cellular structure containing the DNA molecule carrying genes. There are 46 chromosomes in the human genome. Each person receives 23 chromosomes from each parent: 22 autosomal chromosomes plus an X chromosome from the mother and either an X or a Y sex-determining chromosome from the father. Each chromosome has two arms. The shorter arm is referred to as "p," the longer arm as "q." Chromosome Analogy: The genetic material is organized into structures called chromosomes. If the genetic material of an individual is a set of encyclopedias, each chromosome is a volume of that set. Each chromosome contains information that is different from the information contained by the other chromosomes.

Clone An exact (or nearly exact) copy made of a DNA segment, a whole cell, or a complete organism. Cloning of mammals has been accomplished through the process of somatic cell nuclear transfer. In nature, cloning occurs through blastomere separation, when, at a very early stage of development the blastomere separates and forms two distinct and developing pre-embryos. We refer to these clones as identical twins.

Codominance Where two different alleles for a genetic trait are both expressed. This is the case for the alleles for type A and type B blood. The person is said to be AB because both the A and B allele are expressed.

Codon Sequences of three nucleotides (bases) code for amino acids. These triplets are called *codons*. Example: the nucleotide triplet ACC codes for the amino acid serine. The amino acids in turn make up the proteins, which in turn do the work in developing and maintaining a particular organism according to its genetic code. Codon Analogy: If nucleotides are the letters in the DNA alphabet, then codons are the words. Each word has a specific meaning, being an amino acid. Some words are synonyms, coding for the same amino acid.

Complex Trait Many traits and their development involve the expression of more than one gene. In addition many genes interact with the environment to create a trait. These traits are considered "complex."

Conserved Sequence A base sequence in a DNA molecule that has remained essentially unchanged throughout evolution. A conserved sequence could he found in one organism, such as a snail, and another organism, such as a human, and would be the same or very similar.

Cytosine (C) One of the four bases, or building blocks, in DNA.

Deletion A loss of a part of the DNA from a chromosome. This can lead to a disease, or to an abnormality leading to disability.

DNA Deoxyribonucleic acid. The molecule that encodes genetic information. DNA is a double stranded molecule held together by hoods between base pairs of nucleotides. There are four bases in DNA: adenine (A), guanine (G), cytosine (C) and thymine (T). Generally, A only bonds to T, and C to G. DNA Analogy: DNA is the

genetic material. It contains a recipe for the characteristics of a human being. If all genetic material can he considered a set of encyclopedias, the DNA is the words on each page. There are only four "letters" in the DNA alphabet but, just like the 26 letters of the English alphabet, the DNA letters can be put together to form words. Each volume of the set could he considered a chromosome.

DNA repair genes Genes that code for proteins that function to correct errors in DNA base sequences.

Dominant allele An allele that is always expressed, even if only one copy is present. For example, the Huntington allele is dominant. That is, you will get the disease even if only one of the two alleles has the defective gene.

Embryonic Stem Cells A cell found in embryos that can replicate indefinitely and transform itself into other types of cells.

Enzyme An enzyme is a protein that catalyzes reactions. Many of the functions in the human body are chemical reactions (digestion, growth, transmitting signals along nerves). These chemical reactions happen slowly (some reactions would take years) unless an enzyme is present to speed up, or catalyze, the reaction. For example, the enzyme called acetylcholinesterase catalyzes (speeds up) the breakdown of the neurotransmitter acetylcholine. Acetylcholine is released by nerve cells and received by muscle cells, causing the muscle cells to contract. If acetylcholinesterase did not speed up the breakdown of acetylcholine, all your muscles would contract continuously.

Epigenetics The study of how environmental factors change gene function without changing gene sequence.

Epistasis Genes from one location interacting with genes at another location, affecting their expression. For example, if a dog has the gene for brown hair, but does not have the gene for expressing hair colour, the brown hair will not be expressed.

Eugenics The manipulation of the gene pool through artificial selection or genetic engineering with the purpose of improving a species.

Fingerprinting	Every genome is unique because of the accumulation of mutations over time. Fingerprinting in genetics refers to flapping a set of variations due to mutation in someone to uniquely identify them. This process is useful in establishing the presence of a suspect at a crime, establishing paternity, and identifying accident victims.
Gamete	These are mature male or female reproductive cells with a full complement of chromosomes (23).
Gene	The fundamental unit of heredity. An ordered sequence of bases (nucleotides) that encode for specific proteins and the functions those proteins will carry out.
Gene Expression	The process whereby the cellular machinery converts a gene's encoded instructions into the structures and operations of a cell.
Genetic Discrimination	Prejudice against those who have or are likely to develop a genetic disorder.
Genetic Predisposition	A genetic trait that leads to susceptibility to certain diseases. These diseases may or may not actually occur.
Genome	All the genetic material in an individual cell of an organism.
Genomics	The study of genes, their origins, and their functions
Genotype	The genetic constitution of an organism. This includes the traits an organism carries and the traits an organism expresses. This term contrasts with phenotype, which is an organism's measurable traits, or only the traits an organism expresses.
Germ Cell	Sperm and egg cells and their precursors. These are the only cells that contain 23 rather than 46 chromosomes.
Germ Line	The continuation of genetic information from one generation to the next.
Green fluorescent protein	Green fluorescent protein (GFP) is a protein produced by a jellyfish. CFI fluoresces (glows) bluish-green. The gene for GFP has been isolated and inserted into the cells of other organisms, including mice, pigs and monkeys. The expression of certain genes in these transgenic animals can he monitored by examining the pattern of GFP fluorescence.

Huntington Disease	Huntington Disease (HD) is a fatal, progressive, neurodegenerative disease that usually appears in adults between 35 and 50 years of age. HD is inherited as an autosomal dominant trait. An individual who has the HD allele has a 50% chance of transmitting that allele to a child. The HD allele arises as the result of an expansion of a 3-base repeat (CAC) in the gene. Normal individuals have up to 26 repeats. Individuals with over 40 repeats will likely develop HD). A higher number of repeats correlates with earlier onset of symptoms.
Imprinting	A non-permanent alteration of a gene that varies depending upon whether the alteration takes place in a male or a female. In some cases, the particular disease one inherits depends on whether the allele is inherited from the mother or father. For instance an offspring will get either Prader-Willi or Angelman syndrome depending on whether the missing portion of chromosome 15 is inherited from the mother or father.
Junk DNA	A better term for "non-coding" DNA. These are vast stretches of DNA that do not code for the expression of amino acids. They may have regulatory functions, structural functions, or functions we have yet to discover.
Locus	The position of a specific gene on a chromosome.
Messenger RNA (mRNA)	RNA that functions as a blueprint for manufacturing specific sequences of amino acids to produce proteins.
Mitochondrial DNA	Genetic material found in mitochondria. Mitochondria are involved in the production of energy in a cell. Mitochondria (and, therefore, mitochondrial DNA) are inherited only from one's mother.
Monogenic Disorder	A disorder caused by a mutation of a single gene.
Mutation	Any heritable change in DNA sequence.

Nematod Nematodes are microscopic worms generally found in the soil. These structurally simple organisms (adult nematodes are comprised of fewer than 1000 cells) are useful model organisms for scientists studying the genetics of development.

Nuclear Transfer A procedure in which the nucleus of a cell is removed and placed within an oocyte, which then uses the new genetic information in the development of a new organism. This is how cloning in mammals was accomplished.

Nucleotide A subunit of DNA or RNA consisting of a base (A, C, T or C), a phosphate molecule and sugar molecule. These units link to form the DNA or RNA molecule.

Oncogene A gene that, when mutated, is associated with the onset of cancer. Many oncogenes are involved in the control of the rate of cell growth.

Oocyte A female gamete before it matures.

Pedigree A genetic family tree that shows how a particular genetic trait or disease has been transmitted.

Penetrance The probability of a gene or genetic trait being expressed. Complete penetrance means that a particular genotype always results in a particular phenotype. Incomplete penetrance means that a particular genotype (such as polydactyly) is expressed in only a portion of those individuals with that genotype.

Peptide Two or more amino acids joined together.

Pharmacogenomics The study of the interaction between a person's genetic profile and their interaction with specific drugs.

Phenotype A measurable characteristic (blood type, height) that is determined by, or influenced by, expression of a particular gene(s). An organism may have the genotype for blue and brown eyes, but only brown eyes are expressed (brown eyes are autosomal dominant). In this case, the phenotype is for brown eyes.

Pluripotent The potential of a stem cell to develop into more than one type of mature cell depending on environment.

Polygenic	A phenotypic trait created through the interaction of two or more genes.
Polygenic Disorder	Genetic disorders that rely on the combined action of alleles of more than one gene. Although these diseases arc inherited their actual expression is more complex than in monogenic disorders.
Promoter	A site on the DNA strand to which RNA polymerase will bind and begin the process of transcription, the first step of gene expression.
Protein	Large molecules made up of amino acids in specific sequences as determined by the corresponding gene. Proteins provide the structure, function, and regulation of cells, tissues and organs.
Recessive	A gene that will only be expressed if there are two copies of the same allele. (Note: only one copy is required for males on the sex chromosomes.) A recessive allele will only be expressed if the organism does not also have a dominant allele.
Recombinant DNA technology	A procedure for splicing genes from different organisms outside the structure of the cell and then inserting the altered sequence into a cell where it can replicate.
Regulatory Sequence	A DNA sequence that controls gene expression.
Ribonucleic Acid (RNA)	There are several types of RNA. RNA is the blueprint taken from the template DNA, which is used in the construction of amino acids, which in turn determine the shape and function of proteins.
Sex Chromosome	The X or Y chromosome in human beings that determines the sex of an individual. Females have two X chromosomes while males have both an X and a Y chromosome. These sex chromosomes comprise the 23rd chromosomal pair.
Somatic Cell	Any cell in the body except gametes and their precursors.
Stem Cell	Undifferentiated cell. They are found in embryos, placental tissue and bone marrow. Since the most useful lines of stem cells are derived from embryos, research on them is controversial.
Suppressor Gene	A gene that can suppress the action of another gene.

Telomere The end of a chromosome. Telomeres are involved in the replication and stability of DNA molecules. They are therefore thought to the involved in the process of aging.

Thymine (T) One of the four bases, or building blocks, in the DNA sequence.

Totipotent In mammals, totipotent cells have the potential to differentiate into all the cells of an adult organism as well as all the cells of the extra-embryonic membranes.

Transcription The creation of an RNA copy of DNA that may then he used to direct the binding of amino acids to one another, thus creating a protein.

Transfer RNA (tRNA) RNA that uses the information from mRNA (messenger RNA) to position amino acids in a particular order, allowing them to be bound together to create proteins.

Transgenic An experimentally produced organism in which foreign genetic sequences have been added to the germline. ANDi is a good example of a transgenic organism.

Zygote A zygote is the result of fertilization of an egg cell with a sperm cell