



Genetics Terminology List

The Basics

Allele – an alternate form of a gene. Example: the gene that determines whether or not a horse will gray comes in two forms; there is an allele which causes graying and another allele which does not result in graying.

Autosome - any chromosomes not involved in sex determination, as opposed to the sex chromosomes.

Autosomal – a gene which is located on one of the non-sex chromosomes.

Base pair - two chemical bases held together by weak bonds. The bases have specific pairings: adenine (A) pairs with thymine (T) and cytosine (C) pairs with guanine (G). Two strands of DNA are held together in the shape of a double helix by the bonds between base pairs.

Carrier - an individual who carries and is capable of passing on a genetic mutation associated with a disease, but does not display disease signs.

Chromosome - package of DNA that carries genetic information.

Cloning - the process of making a genetic copy of an individual.

Co-dominance - situation in which two different alleles for a trait are both expressed. Example: In blood typing people who have an AB blood type; these individuals have characteristics of both type A and type B blood.

DNA (Deoxyribonucleic acid) - the chemical name for the molecule that carries genetic instructions in all living things. The DNA molecule consists of two strands that wind around one another to form a shape known as a double helix.

DNA sequence - the relative order of base pairs.

Dominant – an allele that is physically expressed if present. Example: the allele for graying (G) is dominant to the allele for non-graying (g), so as long as a horse possesses at least one (G) allele, the horse will be gray.

Gamete - mature male or female reproductive cell (sperm or ovum) with a haploid set (half the number) of chromosomes. In horses, each gamete has 32 chromosomes; after fertilization, the sperm and egg combine their 32 chromosomes so the embryo has a full set of 64 chromosomes.

Gene - section of a chromosome that carries the information for a specific trait.

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Gene locus - the specific location of a gene on a chromosome.

Genotype -the genetic makeup of an individual.

Genetic code - the instructions in a gene that tell the cell how to function.

Genetic marker - a gene or other identifiable portion of DNA whose inheritance can be followed.

Heterozygote - an individual having two different alleles of a particular gene; the individual can pass either allele to their offspring. Example: a horse that is a “carrier” for Cerebellar Abiotrophy is a heterozygote; the horse possesses one copy of the mutated allele and one copy of the normal allele.

Heterozygous – possessing differing forms of an allele for a particular trait.

Homozygote - an individual having two copies of the same allele of a particular gene; the individual can pass only that allele to their offspring. Example: a horse that is chestnut is a homozygote; since chestnut (e) is a recessive trait, all chestnut horses are “ee”.

Homozygous – possessing two copies of the same allele for a particular trait.

Incomplete dominance – a form of intermediate inheritance where an allele for a specific trait is not completely dominant over the other allele; the result is an intermediate phenotype.

Incomplete penetrance – expression of an allele is determined by the presence of other factors.

Inherited – the passing of genetic material from parents to offspring.

Meiosis - the process of cell division in sexually reproducing organisms that reduces the number of chromosomes in reproductive cells from diploid (a full set) to haploid (a half set), leading to the production of gametes (sperm and eggs).

Mendelian genetics -inheritance patterns explained by simple rules of dominant and recessive genes.

Mitochondrial DNA (mtDNA) – a type of DNA located in the cell’s mitochondria and passed along from the female line to offspring.

Mitosis - a cellular process that replicates chromosomes and produces two identical nuclei in preparation for cell division.

Mutation – a change in the DNA sequence.

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Nuclear DNA – a type of DNA located in the cell's nucleus and passed along equally from both parents to offspring.

Phenotype – the physical appearance of an individual.

Protein – a large molecule composed of one or more chains of amino acids in a specific order; the order is determined by the base sequence of nucleotides in the gene that codes for the protein. Proteins are required for the structure, function, and regulation of the body's cells, tissues, and organs; and each protein has unique functions. Examples are hormones, enzymes, and antibodies.

Recessive – an allele that is expressed (displayed phenotypically) in the homozygous state but is not expressed in the presence of a dominant allele.

RNA (Ribonucleic acid) - a chemical found in the nucleus and cytoplasm of cells which plays an important role in protein synthesis and other chemical activities of the cell. The structure of RNA is similar to that of DNA, except that the amino acid thymine (T) is replaced with uracil (U).

Sex chromosome - chromosome that participates in sex determination.

Somatic - any cell in the body except sperm and egg cells.

Variable expression – variation in the type and severity of clinical signs between individuals affected by the same genetic disorder.

X-chromosome – one of the sex chromosomes. Female possess two X chromosomes, while males possess one X chromosome and one Y chromosome.

Y-chromosome – one of the sex chromosomes; this chromosome is responsible for determining if an individual is male. If the Y chromosome is present, the individual is male; females lack the Y chromosome and instead possess two X chromosomes.

Y-chromosome DNA (Y-DNA) – a type of DNA located on the Y chromosome and passed along from the male line to male offspring.

Beyond Basics

Conserved sequence - DNA sequence that has remained essentially unchanged throughout evolution.

Crossing over – during cell division, the breaking of one maternal and one paternal chromosome, the exchange of corresponding sections of DNA, and the rejoining of the chromosomes. This process can result in an exchange of alleles between chromosomes.

Exon - the protein-coding DNA sequence of a gene.

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Fluorescence in situ hybridization (FISH) - is a laboratory technique for detecting and locating a specific DNA sequence on a chromosome. The technique relies on exposing chromosomes to a small DNA sequence called a probe that has a fluorescent molecule attached to it. The probe sequence binds to its corresponding sequence on the chromosome.

Frame-shift mutation – a mutation involving the insertion or deletion of a nucleotide which causes the entire DNA sequence following the mutation to be read incorrectly; resulting in the production of an altered gene product.

Gene family - group of closely related genes that make similar products.

Gene mapping - determination of the relative positions of genes on a DNA molecule (chromosome or plasmid) and of the distance, in linkage units or physical units, between them.

Gene product - the biochemical material resulting from expression of a gene.

Intron – a DNA sequence that interrupts the protein-coding sequence of a gene; an intron is transcribed into RNA but is cut out of the message before it is translated into protein.

Linkage - the proximity of genes or other DNA sequences on the same chromosome.

Linkage map - A map of the relative positions of genetic loci on a chromosome, determined on the basis of how often the loci are inherited together.

Physical map - a map of the locations of identifiable landmarks on DNA (e.g., restriction-enzyme cutting sites, genes), regardless of inheritance. Distance is measured in base pairs.

Point mutation – alteration in a single base pair of a DNA sequence which may or may not alter the gene product.

Polymorphism – natural variations in a gene or DNA sequence that occur frequently in the general population and do not cause an adverse effect.

Promoter – a site or DNA sequence on a chromosome that is needed to turn a gene on or off. The process of transcription is initiated at the promoter and is usually found near the beginning of a gene.

Transcription – formation of an RNA molecule by ‘reading’ the DNA template sequence.

Translation - formation of a polypeptide chain in the specific amino acid sequence directed by the genetic information. The generation of a protein from the transcribed messenger RNA template.

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Telomere - a telomere is the end of a chromosome. Telomeres are made of repetitive sequences of non-coding DNA that protect the chromosome from damage.

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