

## Glossary

### Activator gene:

A gene that codes for an activator protein. The activator protein stimulates the transcription of a target gene - it turns the gene on. See Repressor gene. (Lecture 3G)

### Allele:

One of the versions of a gene, or a version of a DNA sequence, even if it's not a gene. (Lecture 1I).

Expanded definition: "Allele" is a relationship term. A gene/DNA sequence can't just be an allele.

Instead, genes (DNA sequences) are alleles of a locus or of each other if they are different versions of the same locus (if they occur at homologous positions in their chromosomes). If they occur at different loci, then they are alleles of different genes, but not alleles of each other, even if they are members of the same gene family.

### Centromere:

The region of a chromosome whose sequence specifies the location of the kinetochore, where fibers attach and pull chromosomes apart during cell division. (Lecture 1I)

### Chaperone proteins:

A broad class of proteins that function to assist folding/unfolding and assembly/disassembly of proteins and protein complexes, respectively. (Lecture 3B)

### Chaperonin:

A subclass of chaperone proteins that help nascent proteins fold. (Lecture 3B)

### Chromosome:

A very long molecule of double-stranded DNA, typically coiled and complexed with proteins. The DNA carries an organism's genetic information. (Lecture 1A, Lecture 1C)

### Codon:

A sequence of three bases in a DNA or RNA molecule, specifying a specific amino acid during protein synthesis, or the end of protein synthesis ("stop codon"). (Lecture 1E, Lecture 1F)

### Daughter cells

The cells that result from the division of a cell (the parent cell)

### Diploid:

A diploid cell or organism contains two non-identical sets of chromosomes. The majority of higher plants and animals (including humans) have diploid somatic cells. See Haploid, Polyploid. (Lecture 1N)

### Gene dose:

The number of copies of a gene in the genome. In principle "gene dose" *could* refer to having no functional copies due to mutation, deletion, or absence of the Y chromosome, but this is not how the term is used in genetics.

### Exon:

A segment of bases within a gene that is copied from DNA to RNA and remains in the mature mRNA after the splicing process. Most exon sequences are translated to make protein. See Intron. (Lecture 1G)

**Enhancer:**

Short DNA sequences that help regulate gene expression by binding transcription factors, often a long distance (up to 1 Mbp) from the gene that is being regulated.

**Gamete:**

A mature haploid male or female germ cell that is able to unite with another of the opposite sex in sexual reproduction (Lecture 7C).

**Gene:**

Usually, a segment of DNA specifying a protein or functional RNA. (Often used where 'allele' or 'locus' would be clearer.) (Lecture 1A, Lecture 1E)

**Genetic Code:**

The rules by which DNA and mRNA (nucleic acid) sequences specify protein (amino acid) sequences. See "Codon."

**Genome:**

The complete set of genes or genetic material present in a cell or organism

**Genotype:**

The genotype is the specific alleles that distinguish one individual from another. Usually only the alleles relevant to a particular concern are specified.

**Haploid:**

A haploid cell contains one full set of chromosomes. Organisms whose bodies consist of haploid cells include many protists, algae and fungi, mosses for the main part of their life cycle and male bees. (Lecture 1N)

**Haplotype:**

The genotype of a haploid gamete, or of a single chromosome or chromosomal segment. A summary of DNA sequence differences between two individuals, usually within a single unit (e.g. a chromosome or mtDNA). (Lecture 6J).

**Heritable:**

A characteristic (e.g., a trait or DNA sequence) that is transmissible from parent (e.g. an organism or cell) to offspring (e.g. an organism or daughter cells).

**Homology:**

Similarity due to shared ancestry. Homologous genes in different species have similar sequences and functions. Homologous genes within a species usually have the same arrangement on the chromosome. (Lecture 1L)

**Indel:**

A genetic difference created by insertion or deletion of a base pair or a longer DNA segment. (Lecture 1O)

**Intron:**

A segment of bases within a gene that is initially copied from DNA to RNA but is then removed (spliced out) of the RNA before the mRNA sequence is translated to make protein. See Exon. (Lecture 1G)

**Knock-out:**

A mutation that makes a gene in an organism inoperative or unexpressed; e.g. a “leptin receptor knock-out mouse” does not express any functional leptin receptor.

**Kinetochores:**

A protein structure located at the centromere of a chromosome. The kinetochore is the site of spindle fibre attachment (Lecture 7B Part 2).

**Locus:**

The location of a gene or other DNA sequence on a chromosome. (refers to any/all alleles of that gene) (Lecture 1I)

**Meiosis:**

The process of cell division that produces haploid cells from a diploid cell. In plants and animals this usually produces the male and female gametes. This results in four daughter cells (males) or one daughter cell (female; not including polar bodies, each with half the number of chromosomes of the parent cell (Lecture 7C).

**Mitosis:**

The process of cell division that results in two daughter cells with identical chromosomes to the parent cell. (Lecture 7A).

**Monosomy:**

The state of lacking the homologous partner for one chromosome in a diploid organism.

**Trisomy:**

The state of gaining an extra homologous partner for one chromosome in a diploid organism.

**Mutation:**

A heritable change in the DNA sequence. (Lecture 2A)

**Open Reading Frame:**

A sequence of DNA bases starting with ATG and ending with the first in-frame stop codon. (TAA, TAG or TGA.) The equivalent mRNA bases are AUG for the start codon and UAA, UAG and UGA for the stop codon. (Lecture 1F, Lecture 1G)

**Phenotype:**

The observable properties of an organism, including molecular properties detectable by blood tests etc. (Lecture 1A)

**Polymorphism:**

A genetic difference present in  $\geq 1\%$  of the population. (Lecture 1O)

**Polyploid:**

A polyploid cell contains more than two full sets of chromosomes. There are naturally occurring polyploid plants and animals and also polyploid crops produced by plant breeding. Our normal bread wheat is hexaploid (6 sets of chromosomes).

**Promoter:**

A DNA sequence where RNA polymerase binds in order to start transcription. (Lecture 1E)

**Rare variant:**

A genetic difference present in < 1% of the population. (Lecture 1O)

Rate-limiting step:

The slowest step in a biochemical pathway. The other steps are fast compared to this step.

Replication of DNA:

The process that produces two identical DNA molecules from a single molecule. The two strands of DNA separate and each strand acts as a template to make a new complementary strand of DNA. (Lecture 1D)

Repressor gene:

A gene that codes for a repressor protein. The repressor protein prevents the transcription of a target gene - it turns the gene off. See Activator gene.

SNP:

A single-nucleotide polymorphism ('snip'); a SNV present in  $\geq 1\%$  of the population. (Lecture 1O)

SNV:

Single-nucleotide variant; the presence of different nucleotides at homologous positions in two DNA sequences. (Lecture 1O)

Somatic:

Variations in somatic structure that are not hereditary.

Start Codon:

The three-nucleotide sequence (usually AUG) in a mRNA where the ribosome starts translating the codons into the amino acids of a protein. (Lecture 1E).

Stop Codon:

The three-nucleotide sequence (UAA, UAG or UGA) in a mRNA where the ribosome stops translating the codons, and therefore stops making a protein. (Lecture 1E).

Telomere:

The end region of a chromosome. Usually a special repeat sequence that permits the ends of the DNA to be replicated (Lecture 1I).

Terminator:

A region of DNA that gives RNA polymerase the signal to stop transcription. (Lecture 1E)

Tetraploid:

A tetraploid cell contains four full sets of chromosomes. Some commercially grown plants are tetraploids and they are usually fertile when crossed among themselves.

Transcription:

The process in which a segment of DNA is copied using the mechanism of base pairing to produce RNA. (Lecture 1B, Lecture 1E, Lecture 1G, Lecture 1H)

Translation:

The process where the ribosome uses the sequence of bases in mRNA to determine the order of amino acids that it assembles into a protein. (Lecture 1E, Lecture 1F)

**Triploid:**

A triploid cell contains three full sets of chromosomes. Triploidy is fatal in humans. Triploid plants may grow well but are usually infertile. This feature is used to produce seedless fruit in some crops, for example watermelons and bananas.

\*\* A table of useful prefixes and suffixes can be found at  
<http://www.biologyjunction.com/prefixes%20and%20suffixes.pdf>