CAIE Biology A-level

Topic 16 - Inherited Change

Definitions and Concepts

Albinism - An autosomal recessive condition due to mutations of the tyrosinase (*TYR*) gene. It is characterised by lack of pigment in the skin, eyes and hair.

Allele - A version of a gene.

Anaphase I - The third stage of meiosis where the chromosomes that make up the bivalent are pulled apart to the poles of the cell by the spindle fibres.

Anaphase II - The seventh stage of meiosis where the chromatids are pulled apart to opposite poles of the cell.

Autosomal linkage - When two or more genes are positioned on the same autosome. They are unlikely to be separated by crossing over during meiosis so are often inherited together.

Chi-squared (X^2) test - A statistical test used to determine whether a pattern of inheritance is statistically significant.

Codominance - When both alleles for a gene in a heterozygous organism equally contribute to the phenotype.

Crossing over - The exchange of genetic material between two chromosomes in a bivalent.

Degrees of freedom (X^2 test) - The number of categories minus one.

Deletion mutation - A form of gene mutation in which one or more nucleotide bases are removed from a DNA sequence. This may lead to a frameshift mutation, changing every successive codon.

DELLA protein repressors - Negative regulators of gibberellin.

Dihybrid inheritance - The determination of a trait by the inheritance of two genes.

Diploid (2n) - Cells with two copies of each chromosome.

Dominant allele - Describes an allele that is always expressed. Dominant alleles are represented by a capital letter in genetic crosses.

Embryo sac (plants) - The female gametophyte in flowering plants. It is formed when the haploid megaspore nucleus divides.

Epistasis - Describes a relationship between genes at different loci, where the allele of one gene affects the expression of a different gene.

F_1 - The first generation of offspring from a genetic cross.

F_2 - The offspring produced when F_1 individuals reproduce with one another.

Fertilisation - The fusion of male and female gametes. This is random and produces genetically different individuals.

Gametes - Sex cells that have a haploid nucleus and are produced through meioisis.

Gametogenesis - The process of producing new gametes that occurs in sex organs.

Gene - A length of DNA on a chromosome that codes for the production of one or more polypeptide chains and functional RNA.

Gene expression - Some genes in cells are 'switched on' so that their corresponding protein is produced, whereas other genes are silenced.

Gene mutation - A change to at least one nucleotide base in DNA or the arrangement of bases. Gene mutations can occur spontaneously during DNA replication and may be beneficial, damaging, or neutral.

Genetic cross - Individuals of a species are bred. The ratio of phenotypes present in their offspring can be linked to the genotype of the parent.

Genetic variation - Differences in the DNA sequence of individuals, including individuals of the same species.

Genotype - An organism's genetic composition. Describes all alleles.

Gibberellins - Plant hormones that control stem elongation, trigger the growth of the pollen tube during fertilisation and stimulate the mobilisation of food storage reserves during seed germination.

Haemophilia - A sex-linked genetic condition caused by a mutation of the factor VIII (*F8*) gene on the X chromosome. This disrupts the body's ability to form clots.

Haploid (n) - Cells with only one copy of each chromosome.

Heterozygous - When someone has two different alleles of a gene e.g. Ff.

Homologous chromosomes - Two chromosomes with similar gene loci but different alleles, one inherited from each parent.

Homozygous - When someone has two identical alleles of a gene e.g. ff.

Huntington's disease - A genetic condition caused by a mutation (CAG trisomy expansion) of the huntingtin (*HTT*) gene. This results in progressive death of nerve cells in the brain.

Independent assortment - A source of variation in meiosis where the bivalent chromosomes can line up either way around on the metaphase plate.

Inducible enzyme - An enzyme which is only expressed when cells are exposed to its specific substrate.

Insertion mutation - A form of gene mutation in which one or more nucleotide bases are added to a DNA sequence. This may lead to a frameshift mutation, changing every successive codon.

***Lac* operon** - A group of three structural genes, lacZ, lacY and lacA, that are required for the metabolism of lactose.

***Le* allele** - A dominant allele that codes for a functional enzyme in the gibberellin synthesis pathway. If a plant possesses the *Le* allele, gibberellin is active and the plant grows tall.

***le* allele** - A recessive allele that codes for a non-functional enzyme in the gibberellin synthesis pathway. If *le* is homologously present the plant remains short.

Locus - A position on a chromosome.

Meiosis - A type of cell division used to produce gametes that produces four genetically different haploid daughter cells from one parent cell.

Metaphase I - The second stage of meiosis where the bivalent chromosomes align along the metaphase plate and independent assortment occurs.

Metaphase II - The sixth stage of meiosis where the recombinant chromosomes align on the metaphase plate.

Monohybrid cross - A genetic cross in which only one gene is studied.

Phenotype - An organism's observable characteristics. Due to interactions of the genotype and the environment.

Pollen grain - A pollen microspore that the male gametophyte arises from.

Prophase I - The first stage of meiosis where the nuclear envelope breaks down, the spindle fibres form and the chromosomes condense and form bivalents. This is the stage of meiosis where crossing over occurs.

Prophase II - The fifth stage of meiosis where the nuclear envelope breaks down, the spindle fibres form and the chromosomes condense.

Punnett square - A square diagram used to predict the possible genotypes and phenotypes of an offspring and the probability of each when two individuals breed.

Recessive allele - Describes an allele that is only expressed in the absence of a dominant allele. Represented by a small letter.

Regulatory gene - A gene that codes for the production of proteins involved in DNA regulation. The expression of regulatory genes is influenced by internal and external stimuli.

Repressible enzyme - Operons for repressible enzymes are switched on under normal conditions but can be repressed in response to a stimulus.

Sex linkage - The presence of a gene on an X or Y chromosome.

Sexual reproduction - Reproduction which involves the fusion of male and female gametes. It introduces genetic variation.

Sickle cell anaemia - A disease characterised by hook-shaped red blood cells. A missense point mutation of GAG to GTG in the HbA (normal) allele for the β-globin polypeptide results in the HbS (sickle cell) allele of the gene.

Structural gene - A gene that codes for the production of proteins or enzymes that are not involved in DNA regulation.

Substitution mutation - A form of gene mutation in which one nucleotide base is exchanged for another. This may change an amino acid or produce the same amino acid (due to the degeneracy of the genetic code).

Telophase I - The fourth stage of meiosis where the nuclear envelopes reform around the separated chromosomes and they uncoil.

Telophase II - The final stage in meiosis where the nuclear envelopes reform around the separated chromatids (now called chromosomes) and they uncoil.

Test cross - An individual with the dominant phenotype is bred with an individual with the recessive phenotype (homozygous recessive genotype). The ratio of phenotypes present in the offspring indicates whether the dominant individual is homozygous or heterozygous for that allele.

Transcription factors - Proteins that help to switch genes 'on' or 'off' by controlling the binding of RNA polymerase to DNA.

Tyrosinase - An enzyme that catalyses the production of melanin from tyrosine by oxidation.