# OCR (B) Biology A-level

## Topic 5.1 - Genetics in the Twenty First Century

### Definitions and Concepts

## 5.1.1 Patterns of inheritance

**Allele** - A version of a gene.

**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.

**Blood group** - Classification of blood type as A, B or O based on surface antigens present on red blood cells and the presence of certain antibodies in the plasma.

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

**Chromosome mutation** - A change in the number or structure of chromosomes.

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

**Cystic fibrosis** - An autosomal recessive disease which increases mucus production.

**Degrees of freedom** - The number of categories in a chi squared test 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.

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

**Dominant allele** - Describes an allele that is always expressed. Represented by a capital letter in genetic crosses.

**Down's syndrome** - A condition associated with physical and intellectual disability caused by a chromosomal non-disjunction mutation in which three copies of chromosome 21 are inherited.

***Drosophila melanogaster*** - A species of fruit fly often used for studying patterns of inheritance.

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

**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 counselling** - A service that provides information and advice to people affected by or at risk of genetic diseases. This helps individuals and families to make informed decisions.

**Genetic testing** - Cell samples can be screened for genetic conditions.

**Gene variant** - See 'Allele'.

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

**Haemophilia** - A sex-linked genetic condition caused by a gene mutation on the X chromosome. It disrupts the body's ability to form clots.

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

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

**Human Leukocyte Antigens (HLA)** - Cell surface proteins coded for by the major histocompatibility complex (MHC). They vary significantly between individuals and exhibit codominance.

**Huntington's disease** - A genetic condition caused by a CAG trisomy expansion which causes progressive death of nerve cells in the brain.

**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.

**Klinefelter's syndrome** - A condition associated with infertility in males caused by a chromosomal mutation in which males inherit two copies of the X chromosome and one copy of the Y chromosome.

**Locus** - The position of a gene on a chromosome.

**Monogenic inheritance** - See 'Monohybrid inheritance'.

**Monohybrid inheritance** - The determination of a trait by the inheritance of a single gene.

**Nail patella syndrome** - A genetic disorder inherited in an autosomal dominant pattern. It commonly results in malformation of fingernails, kneecaps and elbows.

**Non-disjunction (mutation)** - A chromosomal mutation in which homologous chromosomes or sister chromatids do not separate properly.

**Pedigree analysis** - A diagram showing the phenotypes in several generations of a family can be used to work out which alleles are dominant or recessive.

**Phenotype** - The observable physical characteristics of an organism which is based on the interactions of genotype and the environment.

**Phenylketonuria (PKU)** - An autosomal recessive disorder caused by homozygous inheritance of a mutated PAH gene. Individuals cannot metabolise the amino acid phenylalanine so it accumulates in the brain, which can cause seizures and learning difficulties.

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

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

**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.

**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).

**Translocation mutation** - A chromosomal mutation in which part of a chromosome is moved to another locus.

**Turner's syndrome** - A genetic condition where the second X chromosome in females is either partially or fully missing.

## 5.1.2 Population genetics and epigenetics

**Allele frequency** - The number of times an allele appears at a particular locus in a population, expressed as a proportion or a percentage.

**Allopatric speciation** - The development of new species over time following the geographic isolation of populations of a species.

**Behavioural isolation** - The reproductive isolation of two populations due to differences in their behaviour, e.g. different mating rituals.

**Directional selection** - A type of selection that favours one extreme phenotype and selects against all other phenotypes.

**Disruptive selection** - A type of selection that favours individuals with extreme phenotypes and selects against those with phenotypes close to the mean.

**DNA methylation** - The addition of methyl groups to DNA molecules as a method of epigenetic modification.

**Dutch Hunger Winter study** - A study of pregnant women, the first generation of children and the second generation of children after a famine in the Netherlands in 1944-45 provided evidence for epigenetic modification.

**Ellis-van Creveld syndrome** - An autosomal recessive condition which causes polydactyly, congenital heart defects, short stature nail and teeth defects. Due to the founder effect, it is unusually common in the Amish population.

**Epigenetics** - Heritable changes to gene expression brought about without changing the original sequence of bases that compose the DNA molecule.

**Evolution** - The gradual change in the allele frequencies within a population over time. Occurs due to natural selection.

**Founder effect** - A type of genetic drift in which a few individuals of a species break off from the population and form a new colony. This results in smaller gene pools and an increased frequency of rare alleles.

**Gene expression** - The transcription (and often subsequent translation) of genes.

**Genetic biodiversity** - A measure of the variety of genes that make up a species.

**Genetic bottleneck** - A drastic reduction in population size leading to reduced genetic diversity within a population.

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

**Geographical isolation** - When two populations of a species are separated by a physical barrier, e.g. a mountain or river.

**Hardy-Weinberg principle** - A model that predicts that the ratio of dominant and recessive alleles in a population will remain constant between generations if the following five conditions are met: no new mutations; no natural selection; no migration; large population; and random mating. It provides a formula for calculating the frequencies of alleles:

$$p^2 + 2pq + q^2 = 1.0$$

where p is the frequency of the dominant allele, and q is the frequency of the recessive allele.

**Histone acetylation** - The addition of negative acetyl groups to histones which loosens their binding to DNA, allowing for transcription.

**Histone deacetylation** - The removal of negative acetyl groups from histones which means the histone proteins can bind more tightly to DNA which restricts transcription.

**Histone proteins** - Positively charged proteins which bind to and package DNA and can be a target of epigenetic modification.

**Malaria** - A disease caused by the protoctista *Plasmodium* that lives within two hosts, mosquitoes and humans. It causes recurrent episodes of fever and can be fatal.

**Morphological isolation** - The reproductive isolation of two populations due to the incompatibility of their reproductive systems.

**Natural selection** - The process by which the frequency of 'advantageous' alleles gradually increases in a population's gene pool over time.

**Norrbotten studies** - A series of studies conducted on pregnant women, the first generation of children and the second generation of children after a famine in Sweden provided evidence that environmental factors can cause epigenetic changes after only one or two generations.

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

**Reproductive isolation** - The inability of two populations of the same species to interbreed due to behavioural, morphological or seasonal barriers.

**Seasonal isolation** - The reproductive isolation of two populations due to differences in their breeding seasons.

**Selection pressures** - Environmental factors that drive evolution by natural selection and limit population sizes e.g. competition, predation and disease.

**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.

**Speciation** - The formation of new species due to the evolution of two reproductively separated populations. Two forms: allopatric and sympatric speciation.

**Sympatric speciation** - A form of speciation that occurs when two populations within the same area become reproductively isolated.

**Tertiary structure** - The way that the whole protein folds to make a three dimensional structure.

## 5.1.3 Gene technologies

**Agarose** - A polysaccharide used to make an agar gel matrix.

**Antibiotic resistance** - Some bacteria have favourable mutations which allow them to survive in the presence of an antibiotic. These bacteria reproduce rapidly to form resistant strains.

**Clinical trial** - Testing a new drug on healthy volunteers then a small group of patients for safety and efficacy before testing it on a larger group of patients.

**Cystic fibrosis** - An autosomal recessive disease which increases mucus production.

**Disease predisposition testing** - Genetic screening to determine if a person has alleles associated with increased likelihood of a disease.

**DNA amplification** - The production of many copies of DNA from a small starting amount.

**DNA ligase** - An enzyme that joins the sugar-phosphate backbone of two DNA segments.

**DNA primers** - Short nucleotide sequences, complementary to one end of each of the DNA fragments.

**Exon** - A sequence of DNA that codes for an amino acid sequence.

**Forensic analysis** - The use of DNA analysis to provide evidence for civil or criminal cases e.g. for paternity testing.

**Gel electrophoresis** - A type of chromatography that separates nucleic acid fragments or proteins by size using electric current.

**Genetically modified crops** - A crop that has had its genome altered.

**Genetic engineering** - The modification of the genome of an organism by the insertion of a desired gene from another organism. This enables the formation of organisms with beneficial characteristics.

**Germ line therapy** - A type of gene therapy in which a faulty allele is replaced with a functional allele in germ cells or a very early embryo. The effects of this are permanent and can be inherited.

**Haplotypes** - Alleles or SNPs on a chromosome that are statistically likely to be inherited together.

**Insulin** - A globular protein hormone that is made in the pancreas in response to detection of high glucose levels in the blood.

**Intron** - A non-coding sequence of DNA.

**Knockout mice** - Laboratory mice which have had gene(s) removed or inactivated and are used in studies to identify the function of the knockout genes and the effect of harmful mutations in those genes.

**Log scale** - A nonlinear scale with intervals that increase by powers of 10 used to represent data with a wide range on the same graph.

**Mature mRNA** - The final mRNA product that has had introns removed, as well as having undergone other post-transcriptional changes.

**Messenger RNA (mRNA)** - A type of RNA that carries genetic information from the DNA in the nucleus to the ribosomes for translation.

**Micro RNA (miRNA)** - Short fragments of RNA that are sometimes involved in RNA interference.

**Palindromic** - The triplet code is the same whether read forwards or backwards.

**Paternity testing** - The use of genetic fingerprinting to establish paternity.

**Plasmid** - A small ring of DNA in prokaryotes that carries non-essential genes and can be exchanged between bacterial cells via conjugation.

**Polymerase Chain Reaction (PCR)** - An *in vitro* technique used to rapidly amplify fragments of DNA.

**Post transcriptional modification** - Exons can be rejoined in a different order after pre-mRNA splicing. This means that different mRNA molecules can be produced from the same gene.

**Pre-mRNA** - The product of transcription before any post-transcriptional regulation.

**Recognition sequences** - Short, palindromic sequences of DNA recognised by restriction enzymes.

**Reporter genes** - Genes that can be inserted into plasmids at the same time as DNA fragments to identify transformed cells.

**Restriction enzymes** - Enzymes that cut DNA molecules at recognition sequences, creating sticky ends.

**Reverse transcriptase** - An enzyme which produces cDNA from an RNA template.

**RNA interference** - siRNAs and miRNAs act to inhibit gene expression, usually by destroying mRNA so that it cannot be translated.

**Severe combined immunodeficiency disease (SCID)** - A group of polygenic diseases which disrupt the development of B and T lymphocytes, resulting in persistent infection.

**Small interfering RNA (siRNA)** - A short sequence of double-stranded RNA produced when the eukaryotic enzyme dicer hydrolyses double-stranded viral RNA.

**Single nucleotide polymorphisms (SNPs)** - A single nucleotide change in the base sequence.

**Somatic therapy** - A type of gene therapy in which a faulty allele is replaced with a functional allele in affected somatic cells. The effects of this are temporary and cannot be inherited.

**Splicing** - Introns are removed from eukaryotic pre-mRNA to form mature mRNA that can be translated.

**Transcription** - The process of synthesising a new mRNA strand from a molecule of DNA. The process of synthesising a new mRNA strand from a molecule of DNA.

**Transformation** - The reinsertion of plasmids back into bacterial cells to form transgenic bacteria. This involves mixing the plasmids and bacterial cells.

**Taq polymerase** - A DNA polymerase from a bacterial species which is adapted to live in extreme heat. It is used in PCR technology to produce fragments.

**Variable number tandem repeats (VNTRs)** - Repeated DNA sequences in introns.

**Vector** - A carrier used to transfer a gene from one organism to another, e.g. plasmid.