Higher Biology Unit 1 DNA and the Genome
Learning Outcomes

Key Area 1: The Structure of DNA

- Genetic information is inherited
- DNA is a substance that encodes the genetic information of heredity in a chemical language
- DNA is a very long double-stranded molecule in the shape of a double helix
- Each strand is made up from chemical units called nucleotides
- A nucleotide is made up of three parts: a deoxyribose sugar, a phosphate and a base
- Deoxyribose molecules have five carbon atoms, which are numbered 1 to 5
- The phosphate of one nucleotide is joined to carbon 5 (5') of its sugar and linked to carbon 3 (3') of the sugar of the next nucleotide in the strand to form a 3'–5' sugar–phosphate backbone
- There are four different bases called adenine (A), guanine (G), thymine (T) and cytosine (C)
- Genetic information is encoded in the sequence of bases along the length of one of the strands of a DNA molecule. The nucleotides of one strand of DNA are linked to the nucleotides on the second strand through their bases – the bases form pairs joining the strands
- Bases pair in a complementary way – adenine always pairs with thymine and guanine always pairs with cytosine. Base pairs are held together by hydrogen bonds
- Each strand has a sugar–phosphate backbone with a 3' end that starts with a deoxyribose molecule and a 5' end that finishes with a phosphate
- The two strands of a DNA molecule run in opposite directions and are said to be antiparallel to each other. Molecules of DNA can be linear or circular
- Prokaryotic cells do not have a distinct nucleus and their DNA is organised into circular chromosomes and plasmids
- Eukaryotic cells have nuclei containing their DNA in linear chromosomes
- Linear chromosomes in eukaryotic cells have very tightly coiled DNA, which is packaged with associated proteins.
- Yeast is a special example of a eukaryote as it also has plasmids
- Mitochondria and chloroplasts in eukaryotic cells have DNA in circular chromosome

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Key Area 2: Replication of DNA

- Replication is the process by which DNA molecules can direct the synthesis of identical copies of themselves
- DNA molecules replicate prior to cell division
- DNA unwinds and unzips to form two template strands
- Replication starts at several places along the DNA molecule at the same time
- The enzyme DNA polymerase adds complementary DNA nucleotides to the 3' end of a DNA strand
- DNA polymerase requires primers to start replication
- Primers are short complementary sequences of nucleotides that allow binding of DNA polymerase
- The 3'–5' lead strand is replicated continuously in the direction from its 3' end towards its 5' end
- Nucleotides are added as fragments on the lagging strand
- The replicated fragments on the lagging strand are joined together by a ligase enzyme
- The polymerase chain reaction (PCR) is a laboratory technique for the amplification of DNA
- PCR uses primers complementary to specific target sequences at the two ends of a DNA region to be amplified. In PCR, heating separates the two strands of the DNA to be amplified
- The separated strands are cooled to allow primers to bind to target sequences
- Heat-tolerant DNA polymerase (Taq polymerase) replicates the region of DNA that has been primed
- A cycle of PCR doubles the number of copies of a region of DNA
- Repeated cycles of heating and cooling amplify the region of DNA
- Practical applications of PCR include forensics and studies in evolution

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Key Area 3: Gene Expression

- Genes are encoded into DNA and the genetic code is found in all forms of life
- Eukaryotic genes have introns (non-coding regions) and exons (coding regions)
- Genes are transcribed and translated during gene expression
- Gene expression is controlled by the regulation of transcription and translation
- Genes are expressed to produce proteins
- Proteins have a variety of shapes which determines their functions
- Proteins are formed from polypeptides, which are chains of amino acids held together by peptide bonds and folded in various ways
- The polypeptide chains are held in their folded 3D shape by hydrogen bonds and other interactions between individual amino acids
- Protein function depends on the three-dimensional shape of its molecules
- Protein functions include acting as structural components of cells, enzymes, some hormones and antibodies
- Only a fraction of the genes in a cell are expressed
- Transcription and Translation involves three types of RNA (mRNA, tRNA and rRNA). This is similar to DNA but it is single-stranded, its nucleotides contain ribose instead of deoxyribose and the base uracil replaces the thymine found in DNA
- DNA in the nucleus is transcribed to produce messenger RNA (mRNA), which carries a copy of the genetic code
- In transcription, RNA polymerase moves along DNA, unwinding the double helix and breaking the hydrogen bonds between bases. RNA polymerase aligns RNA nucleotides by complementary base pairing to form a primary mRNA transcript
- RNA splicing forms the mature transcript. Introns are removed from the primary transcript and the exons spliced together to form a mature mRNA transcript.
- The order of exons is unchanged during splicing
- Alternative RNA splicing allows different mature mRNAs to be formed from the same primary mRNA transcript depending on which exons are

retained

- Triplets of bases on mRNA are called codons
- Translation of mRNA results in the production of a polypeptide
- Most codons code for specific amino acids
- Translation begins at a start codon and stops at a stop codon
- Ribosomes are made from ribosomal RNA (rRNA) and proteins
- mRNA carries a copy of the DNA code from the nucleus to the ribosomes, where it is translated.
- Transfer RNA (tRNA) folds because of base pairing and forms a triplet anticodon site and an attachment site for a specific amino acid
- Amino acids are carried by specific tRNA molecules
- tRNA anticodons align with their complementary codons on mRNA
- tRNA molecules deliver amino acids in sequence, which are then joined together by peptide bonds to form polypeptides
- Following polypeptide formation, tRNA exits the ribosome to collect further amino acids
- As a result of alternative RNA splicing one gene can express many proteins
- Phenotype is determined by the proteins produced as a result of gene expression. Environmental factors also influence phenotype.

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Key Area 4: Cellular Differentiation

- Cellular differentiation is a process in which a cell develops more specialised functions
- Cellular differentiation is the process by which cells express the genes to produce proteins characteristic of that type of cell
- Meristems are regions of unspecialised cells in plants
- Meristem cells can continue to divide (self-renew) and/or differentiate into specialised cells
- Stem cells are unspecialised cells in animals that can continue to divide (self-renew) and/or differentiate into specialised cells
- In the very early mammal embryo, embryonic stem cells can divide and differentiate into all of the cell types that make up the organism. They are pluripotent
- Tissue (adult) stem cells replenish differentiated cells that need to be replaced following damage or disease
- Tissue stem cells can differentiate into all of the types of cell found in a particular tissue type. They are multipotent. For example blood stem cells located in bone marrow can give rise to all types of blood cell.
- Stem cell research provides information on how cell processes such as cell growth, differentiation and gene regulation work
- Stem cells are used therapeutically in the repair of damaged or diseased organs or tissues (e.g. repair of the cornea in eyes, regeneration of damaged skin)
- Stem cells can be used as model cells to study how diseases develop or for drug testing
- Embryonic stem cell use is regulated and raises ethical issues as the embryo must be destroyed
- Given the right conditions stem cells from embryos can self-renew in the lab

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Key Area 5&6: The Structure of the Genome & Mutations

- The genome of an organism is the genetic information encoded into its DNA that can be inherited by its offspring
- DNA sequences that code for protein are known as genes
- A genome is defined as the genes that code for protein and other DNA sequences that do not code for proteins
- Most of the genome in eukaryotic species consists of non-coding sequences
- Non-coding sequences include those that regulate transcription and those that are transcribed into mRNA but are not translated
- Some non-coding DNA sequences have no known function
- tRNA and rRNA are non-translated forms of RNA
- Mutations are changes in the DNA that can result in no protein or an altered protein being synthesized.
- Single gene mutations affect DNA nucleotide sequences and include deletion, insertion and substitution of nucleotides
- Nucleotide substitutions include missense, nonsense and splice-site mutations
- Missense mutations result in one amino acid being changed for another. This may result in a non-functional protein or have little effect on the protein
- Nonsense mutations result in a premature stop codon being produced which results in a shorter protein
- Splice-site mutation result in some introns being retained and/or some exons not being included in the mature transcript
- Nucleotide insertions or deletions result in frame-shift mutations
- Frame-shift mutations cause all the codons and all the amino acids after the mutation to be changed. This has a major effect on the protein.
- Chromosome mutations that involve alterations to the structure of a chromosome include duplication, deletion, inversion and translocation
- Duplication is where a section of a chromosome is added from its homologous partner
- Deletion is where a section of chromosome is removed

- Inversion is where a section of chromosome is reversed
- Translocation is where a section of a chromosome is added to a chromosome, not its homologous partner
- The substantial changes in chromosome mutations often make them lethal
- Mutations are important in evolution. Mutation is the source of all new alleles
- Duplication is important in evolution as it allows potential beneficial mutations to occur in a duplicated gene whilst the original gene can still be expressed to produce its protein

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Key Area 7: Evolution

- Evolution is the changes in organisms over generations as a result of genomic variations
- In vertical inheritance, genetic sequences pass from parent to offspring as a result of sexual or asexual reproduction
- Natural selection is more rapid in prokaryotes
- Horizontal gene transfer is where genes are transferred between individuals in the same generation.
- Prokaryotes can exchange genetic material by horizontally, resulting in faster evolutionary change than organisms that only use vertical transfer.
- Natural selection is the non-random increase in the frequency of DNA sequences that increase survival and the non-random reduction in the frequency of deleterious sequences
- Changes in phenotype are a result of selection
- Selection can be stabilising, directional or disruptive
- Stabilising selection – an average phenotype is selected for and extreme phenotypes are selected against
- Directional selection – one extreme of the phenotype is selected for
- Disruptive selection – two or more phenotypes are selected for
- Speciation is the generation of new biological species by evolution, as a result of isolation, mutation and natural selection
- A species is a group of organisms capable of interbreeding to produce fertile offspring and which does not normally breed with other groups
- Geographical barriers prevent gene flow in allopatric speciation
- Behavioural and ecological barriers prevent gene flow in sympatric speciation

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Key Area 8: Genomic Sequencing

- In genomic sequencing, the sequence of nucleotide bases can be determined for individual genes and entire genomes.
- Comparison of sequence data requires bioinformatics, which involves computer and statistical analyses.
- Comparison of genomes from different species reveals that many genes are highly conserved across different organisms.
- Many genomes have been sequenced including disease-causing organisms and pest species
- Phylogenetics is the study of evolutionary relatedness among groups of organisms.
- Molecular clocks are used to show when species diverged during evolution.
- Molecular clocks assume a constant mutation rate and show differences in DNA sequences or amino acid sequences between organisms.
- Difference in sequence data between species indicate the time of divergence from a common ancestor
- Evidence from phylogenetics and molecular clocks has been used to determine the main sequence of events in evolution. The sequence data is used together with fossil evidence.
- Comparison of sequence data has provided evidence for three domains of life: the bacteria, the archaea and the eukaryotes.
- Sequence data and fossil evidence have been used to determine the main sequence of events in the evolution of life.
- Main events in the evolution: cells, last common ancestor, prokaryotes, photosynthetic organisms, eukaryotes, multicellular organisms, animals, vertebrates and land plants.
- An individual's genome can be analysed to predict the likelihood of developing certain diseases
- Pharmacogenetics is the use of genome information in the choice of drugs to treat patients.
- Analysis of an individual's genome could be used to select the moist effective drugs and dosage to treat their disease (personalised medicine)