

Unit 1 Human cells

1. Division and differentiation in human cells

Stem cells

- Describe the process of differentiation.
- Explain how differentiation is brought about with reference to genes.
- Name the two types of stem cell.
- Describe the role of each type of stem cell.
- State the location where each type of stem cell can be found.

Somatic cells

- State that somatic cells divide by mitosis to produce more somatic cells.
- Describe the fate of somatic cells after differentiation.
- Explain what is meant by the terms tissue and organ.
- Describe the role of epithelial cells.
- Describe the role of connective tissue.
- Describe the role of muscle tissue.
- Describe the role of nerve tissue.
- State that during cell division the nucleus of a somatic cell divides by mitosis to maintain the diploid chromosome number.
- State that diploid cells have 23 pairs of homologous chromosomes.

Germline cells

- State that germline cells divide by mitosis to produce more germline cells or by meiosis to produce haploid gametes.
- Compare the effects of mutation in germline cells compared to somatic cells.

Research and therapeutic uses of stem cells

- Describe some current therapeutic uses of stem cells.
- State that stem cells can also be used as model cells to study how diseases develop or for drug testing.
- State that stem cell research provides information on how cell processes such as cell growth, differentiation and gene regulation work.
- Discuss the benefits and ethical issues of stem cell research.

Cancer cells

- Describe how cancer cells differ from somatic cells.
- Explain why tumours can form.
- Explain what can happen if cancer cells do not attach to one another.

2. Structure and function of DNA

Structure and replication of DNA

- State that DNA is made up of units called nucleotides.
- Describe the composition of a nucleotide.
- State that the DNA backbone is called a sugar phosphate backbone.
- Name the bonds which hold the sugar of one nucleotide to the phosphate of the next.
- Name the four bases of DNA and describe the base pairing rule.
- State that hydrogen bonds hold the bases together.
- State that DNA is a double stranded helix.
- Give the meaning of the term antiparallel.
- Add labels to a diagram of DNA to indicate the 5' and 3' ends of each strand.
- State that all cells store their genetic information in the base sequence of DNA.
- State that the genotype is determined by the sequence of bases.
- Describe the organisation of DNA in human cells.
- State that DNA found in the linear chromosomes of human cells is tightly coiled and packaged with associated proteins.
- Explain the role of DNA polymerase in the replication of DNA.
- Explain the role of primers in the replication of DNA.
- State that DNA polymerase needs a primer to start replication and can only add complementary DNA nucleotides to the deoxyribose (3') end of a DNA strand.
- Explain the role of ligase in the replication of DNA.
- Describe the process of DNA replication in stages.
- Explain the difference between the replication process on the leading and lagging strands.
- State that DNA replication happens at several locations on a DNA molecule at one time.

Gene expression

- State that phenotype is determined by proteins produced as a result of gene expression.
- State that only a fraction of the genes present in a cell are expressed.
- State that gene expression involves two processes; transcription and translation.
- Describe the structure of RNA.
- Describe the role of mRNA.
- Describe the role of rRNA.
- Describe the role of tRNA.
- Describe the process of transcription including the role of RNA polymerase and complementary base pairing.
- Give the meaning of the terms intron and exon.
- Describe the process of RNA splicing.
- Describe the process of translation including the role of tRNA and ribosomes.
- State that codons are found on mRNA and anticodons are found on tRNA.
- Name the bonds which hold amino acids together in a protein.
- Give the meaning of the term post-translational modification.
- State that many proteins arise from one gene due to alternative RNA splicing and post-translational modification.
- Explain how alternative RNA splicing and post-translational modification give rise to many proteins.

Genes and protein in health and disease

- State that polypeptide chains fold to give the final structure of the protein.
- Describe the role of hydrogen bonds and interactions between amino acids in the 3D shape of a protein.
- Describe the functions of proteins to include enzymes, structural, hormones and antibodies.
- Give the meaning of the term mutation.
- Explain what single gene mutations are including substitution, insertion and deletion.
- Describe the effects of substitutions including missense, nonsense and splice-site mutations.
- Describe the effects of insertions and deletions including frame-shift mutations and expansion of a nucleotide sequence repeat.
- Describe chromosome structure mutations including duplication, deletion and translocation.
- State that substantial changes in chromosome mutations often make them lethal.

Human genomics

- State that genomic sequencing allows the sequence of nucleotide bases to be determined for individual genes and entire genomes.
- State that to compare sequence data, computer and statistical analyses (bioinformatics) are required.
- State that computer programs can be used to identify gene sequences by looking for coding sequences similar to known genes, start sequences or sequences lacking stop codons.
- Describe the use of sequence data to study the evolutionary relatedness among groups of organisms.
- Explain the importance of comparing genomes from different species.
- Give the meaning of the term pharmacogenetics.
- Describe the role of personal genomics in health.
- State that analysis of an individual's genome may lead to personalised medicine.
- State that individual genome analysis can allow us to gather information about genetic component of risk of disease and likelihood of success of a particular treatment.
- State that PCR allows the amplification of DNA using complementary primers.
- State that PCR amplifies DNA *in vitro*.
- Describe the stages involved in the PCR process.
- Explain the role of primers in the PCR process.
- Explain the importance of using heat-tolerant DNA polymerase in this process.
- Explain what probes are and how they are detected.
- Describe the use of arrays of DNA probes.
- Explain how DNA profiling is performed and describe its benefits.
- Explain how DNA probes can be used to screen for diseases.

3. Cell metabolism

Metabolic pathways

- Explain what is meant by the term metabolism.
- Define the term anabolic and catabolic.
- State that anabolic and catabolic reactions can have reversible and irreversible steps and alternative routes.
- Describe how metabolic pathways can be controlled by the presence or absence of particular enzymes and through the regulation of the rate of reaction of key enzymes within the pathway.

Metabolic pathways continued

- State that regulation can be controlled by intra- and extracellular signal molecules.
- Explain how enzymes which are constantly produced are controlled.
- State that most metabolic reactions are reversible.
- Explain how reactions are reversible
- Describe the induced fit model of enzyme action.
- Describe the role of the active site of an enzymes.
- Define the term activation energy.
- Describe the effects of substrate and end product concentration on the direction and rate of enzyme reactions.
- State that enzymes often act in groups or as multi-enzyme complexes.
- Explain how metabolic pathways can be controlled by competitive inhibition.
- Explain how metabolic pathways can be controlled by non- competitive inhibition.
- Explain how metabolic pathways can be controlled by feedback inhibition.
- State that competitive inhibition can be reversed by increasing substrate concentration.

Cellular respiration

- Explain what is meant by the term cellular respiration.
- Describe the role of ATP in the transfer of energy.
- Describe the role of ATP in the phosphorylation of other molecules.
- Describe the process of glycolysis.
- Describe the role of the enzyme phosphofructokinase.
- Explain the meaning of the terms energy investment and energy pay-off with reference to ATP.
- Describe the processes which take place during the citric acid cycle.
- Describe the role of dehydrogenase enzymes.
- Describe the role of the coenzymes FAD and NAD.
- Explain what the electron transport chain is and state where it is found.
- Describe the role of hydrogen ions in the generation of ATP.
- Describe the role of high energy electrons in the generation of ATP.
- Describe the role of the enzyme ATP synthase.
- State that the return flow of hydrogen ions rotates part of the membrane protein ATP synthase, catalysing the synthesis of ATP.
- State that the final electron acceptor is oxygen, which combines with hydrogen ions and electrons to form water.

Cellular respiration continued

- Name alternative substrates for respiration.
- State that starch and glycogen are broken down to glucose for use as a respiratory substrate.
- Explain how sugars other than glucose can be used in respiration.
- State that proteins can be broken down to amino acids and converted to intermediates of glycolysis and the citric acid cycle for use as respiratory substrates.
- State that fats can also be broken down to intermediates of glycolysis and the citric acid cycle.
- State that the pathways of cellular respiration are regulated by feedback inhibition.
- Describe how ATP production is regulated by inhibition of phosphofructokinase.
- Explain how the rates of glycolysis and citric acid cycle are synchronised.
- State that the cell conserves its resources by only producing ATP when required.

Energy systems in muscle cells

- Describe the role of creatine phosphate in the release of energy.
- Describe the role of ATP during strenuous muscle activity.
- State that the creatine phosphate system can only support strenuous muscle activity for around 10 seconds, when the creatine phosphate supply runs out.
- Explain how levels of creatine phosphate are restored.
- Describe the process of anaerobic respiration.
- Describe the circumstances under which anaerobic respiration takes place.
- State which stages of respiration cannot take place in the absence of oxygen.
- Describe the fate of pyruvate during anaerobic respiration.
- Describe the role of NAD in anaerobic respiration.
- Give the meaning of the term muscle fatigue.
- Give the meaning of the term oxygen debt and explain how this is repaid.
- Explain what is meant by the term skeletal muscle fibres.
- Describe the role of slow twitch muscle fibres.
- Describe the activities for which these muscle fibres are important.
- Explain how slow twitch muscle fibres generate ATP.
- State that slow twitch muscle fibres have many mitochondria, a large blood supply and a high concentration of the oxygen storing protein myoglobin.
- State that the major storage fuel of slow twitch muscles fibres is fats.
- State that slow twitch (Type 1) muscle fibres contract more slowly, but can sustain contractions for longer.

Energy systems in muscle cells continued

- State that fast twitch (Type 2) muscle fibres contract more quickly, over short periods.
- Describe the activities for which these muscle fibres are important.
- Explain how fast twitch muscle fibres generate ATP.
- State that fast twitch muscle fibres have few mitochondria and a lower blood supply than slow twitch muscle fibres.
- Name the major storage fuels of fast twitch muscles.
- State that most human muscle tissue contains a mixture of both slow and fast twitch muscle fibres.
- State that athletes show distinct patterns of muscle fibres that reflect their sporting activities.