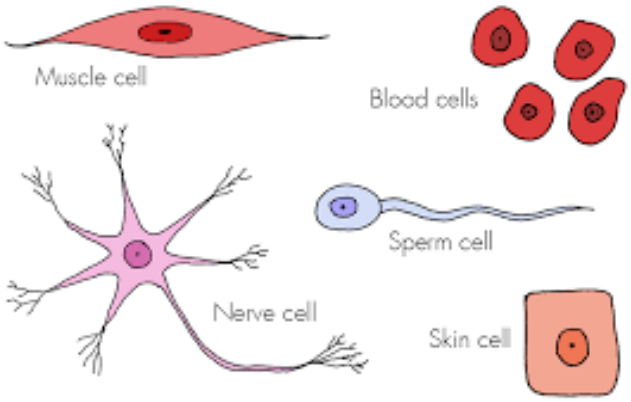
Higher Human Biology [2018IM]

Name _____

Unit 1 - Human Cells



Learning Outcome Checklists and Glossaries

HHB Unit 1.1 DIVISION AND DIFFERENTIATION LOs

	..by the end of the lesson(s) I should know…
Division in Somatic and Germline	☐ Somatic cells are any cell in the body other than cells involved in reproduction ☐ Germline cells are gametes (sperm and ova) and the stem cells that divide to form gametes ☐ Somatic stem cells divide by mitosis to form more somatic cells. ☐ Germline stem cells divide by mitosis to produce more germline stem cells ☐ Germline stem cells divide by meiosis to produce haploid gametes ☐ Mitosis maintains the diploid chromosome number of 23 pairs of homologous chromosomes ☐ In meiosis the nucleus divides twice. 1st division separates the homologous chromosomes. 2nd division separates the chromatids. ☐ Meiosis creates haploid gametes containing 23 single chromosomes ☐ The nucleus of germline cells have 23 pairs of chromosomes and are said to be diploid.
Cellular Differentiation	☐ Cellular differentiation is the process by which a cell expresses certain genes to produce proteins characteristic for that type of cell. ☐ Differentiation allows a cell to carry out specialised functions ☐ Embryonic stem cells are in the very early embryo ☐ All the genes in embryonic stem cells can be switched on, so these cells can differentiate into any cell type and so are pluripotent ☐ Tissue stem cells are involved in growth, repair and renewal of the cells found in that tissue. ☐ Tissue stem cells can differentiate into all the types of cell found in a particular tissue type and so are multipotent ☐ Blood stem cells in the bone marrow are an example of tissue stem cells, they can differentiate into red blood cells, platelets, phagocytes and lymphocytes
Therapeutic and Research use of Stem cells	☐ Stem cells can be used therapeutically for the repair of damaged or diseased organs or tissues ☐ Therapeutic uses of stem cells include corneal repair and regeneration of damaged skin ☐ Stem cell research provides information on how cell processes such as cell growth, differentiation and gene regulation work. ☐ Stem cells can also be used as model cells to study how diseases develop or for drug testing. ☐ Embryonic stem cells can self-renew, under the right conditions, in the lab ☐ Embryonic stem cells can be used to treat disease of injury ☐ Using Embryonic stem cells involves destroying embryos and so causes ethical issues
Cancer cells	☐ Cancer cells divide excessively to produce a mass of abnormal cells (a tumour) that do not respond to regulatory signals and may fail to attach to each other. ☐ If the cancer cells fail to attach to each other they can spread through the body to form secondary tumours.

HHB Unit 1.2 STRUCTURE AND REPLICATION OF DNA LOs

	..by the end of the lesson(s) I should know…
DNA structure	☐ DNA is a long double stranded molecule ☐ the two strands of DNA are twisted in to a double helix ☐ the sub units of DNA are nucleotides ☐ each nucleotide contains deoxyribose sugar, phosphate & base. ☐ DNA has a 3'-5' sugar–phosphate backbone, where the phosphate joined to carbon 5' on one nucleotide is joined to the carbon 3' of the next nucleotide ☐ there are four different bases – (A) adenine, (T) thymine, (G) guanine and (C) cytosine. The base sequence of DNA forms the genetic code ☐ complementary base pairing means A and T pair & C and G pair ☐ two DNA strands are held together by hydrogen bonds between the bases ☐ the two DNA strands run in opposite directions = antiparallel
DNA replication	☐ Replication of DNA occurs before cell division (mitosis & meiosis) to make identical copies of the chromosomes ☐ an enzyme called DNA polymerase adds complementary DNA nucleotides to the deoxyribose (3') end of a DNA strand. ☐ DNA polymerase requires a primer to start replication ☐ primers are short complimentary sequences of DNA that allow the binding of DNA polymerase ☐ DNA is unwound and the hydrogen bonds between bases are broken to form two template strands. ☐ the 3'-5' strand is called the lead strand and is replicated continuously from the 3' end to the 5' end ☐ the 5'-3' strand is called the lagging strand and is replicated in fragments which are joined together by the enzyme ligase ☐ ATP is required to provide the energy for replication
PCR	☐ Polymerase Chain Reaction is a process which amplifies DNA ☐ Primers are short strands of DNA and are used in PCR to target base sequences at both ends of the region of DNA to be amplified ☐ PCR involves repeated cycles of heating and cooling ☐ Heating the DNA to 92-98oC to separate the two strands of DNA ☐ Separated strands are then cooled to 50-60oC to allow primers to bind to target sequences ☐ It is then heated to 70-80oC and heat tolerant DNA polymerase replicate the region of DNA ☐ PCR can be used to amplify DNA to- help solve crime, settle paternity suits, diagnose genetic disorders

HHB Unit 1.3 GENE EXPRESSION LOs

	..by the end of the lesson(s) I should know…
gene expression	Gene expression involves the transcription and translation of DNA sequences only a fraction of the genes in a cell are expressed gene expression involves three kinds of ribonucleic acid - RNA. RNA is single stranded and is composed of nucleotides containing ribose sugar, phosphate and one of four bases : cytosine, guanine, adenine and uracil. messenger RNA (mRNA) carries a copy of the DNA code from the nucleus to the ribosome, it is transcribed from DNA in the nucleus and translated into proteins by ribosomes in the cytoplasm each triplet of bases on the mRNA molecule is called a codon and codes for a specific amino acid ribosomal RNA (rRNA) and proteins form the ribosome. transfer RNA (tRNA) folds due to complementary base pairing and carries a specific amino acid to the ribosome a tRNA molecule has an anti-codon, an exposed triplet of bases, at one end and a site for amino acid attachment at the other end
Transcription	in transcription, DNA is transcribed to into primary mRNA transcripts in the nucleus RNA polymerase unwinds the double helix, breaks the hydrogen bonds between the bases and aligns RNA nucleotides by complementary base pairing (C-G& A-U/T-A) the primary transcript includes introns (non-coding DNA sequence) and exons (coding DNA sequence) during RNA splicing the introns of the primary transcript of mRNA are removed and the exons are joined together to form mature transcript. alternative RNA splicing allows different mature transcripts to be formed Different proteins can be expressed from one gene because of alternative RNA splicing
Translation	in translation, the mRNA code is turned into a polypeptide by tRNA at the ribosome. Translation begins at a start codon and ends at a stop codon Anticodons bond to codons by complementary base pairing, translating the genetic code into the correct sequence of amino acids. adjacent amino acids are joined by a peptide bond to form a polypeptide tRNA exits from the ribosome to collect more amino acids.
Protein	an organism's phenotype is determined by the proteins produced as the result of gene expression environmental factors also affect phenotype amino acids are linked by peptide bonds to form polypeptides polypeptide chains fold to form the three-dimensional shape of a protein, held together by hydrogen bonds and other interactions between individual amino acids proteins have a large variety of shapes which determines their functions

HHB Unit 1.4 MUTATIONS LOs

	..by the end of the lesson(s) I should know…
mutations	▯mutations are random changes in genetic information ▯mutations can alter genes or chromosomes ▯mutations result in no protein or a faulty protein being expressed.
single gene mutations	▯Single gene mutations involve the alteration of a DNA nucleotide sequence ▯single gene mutations include the substitution, insertion or deletion of nucleotides. ▯single-nucleotide substitutions include: missense, nonsense and splice-site mutations ▯missense mutations result in one amino acid being changed for another ▯missense mutations may result in a non-functional protein or have little effect on the protein ▯nonsense mutations results in a premature stop codon being produced ▯nonsense mutations result in the production of a shorter protein ▯splice site mutations change the bases at the point where intron and exon meet ▯splice site mutations result in some introns being retained and/or some exons being removed ▯insertions or deletions result in frame-shift mutations ▯frame shift mutations cause all the amino acids after mutation to be affected, causing a major effect on structure of the protein produced
chromosome mutations	▯deletion – a section of a chromosome is removed ▯duplication – a section of a chromosome is added from its homologous (matching) chromosome ▯inversion – a section of chromosome is reversed ▯translocation – a section of a chromosome is added to a chromosome that is not its homologous partner (ie not its matching chromosome) ▯the substantial changes in chromosome mutations often make them lethal.

HHB Unit 1.5 HUMAN GENOMICS LOs

Lesson(s)	..by the end of the lesson(s) I should know…
Genome	□ the genome of an organism is its entire hereditary information encoded in DNA □ a genome is made up of genes that code for proteins and other DNA sequences that do not code for proteins □ in genomic sequencing the sequence of nucleotide bases can be determined for individual genes and entire genomes □ computer programs can be used to identify base sequences by looking for sequences similar to known genes □ Bioinformatics is the use of computer technology to identify DNA sequences.
Personalised medicine	□ An individual's genome can be analysed to predict the likelihood of developing certain diseases □ Pharmacogenetics is the use of the genome information in the choice of drug treatment □ personalised medicine is based on an individual's genome □ an individual's personal genome sequence can be used to select the most effective drugs and dosage to treat their disease

HHB Unit 1.6 METABOLIIC PATHWAYS LOs

Lesson(s)	..by the end of the lesson(s) I should know…
Types of metabolic pathways	□Metabolic pathways are integrated and controlled pathways of enzyme-catalysed reactions within a cell □Metabolic pathways can involve anabolic reactions or catabolic reactions □Anabolic reactions build up small molecules into large molecules and require energy □Catabolic reactions breakdown large molecules into smaller molecules and release energy □metabolic pathways can have reversible and irreversible steps and alternative routes.
Enzymes	□metabolic pathways are controlled by the presence or absence of particular enzymes □ metabolic pathways are controlled by the regulation of the rate of reaction of key enzymes □substrate molecules have an affinity for the active site of an enzyme □the enzyme is flexible, and the substrate can induce the active site to change shape by induced fit to fit the substrate better □the active site can orientate the substrate molecules so that they fit more closely □Activation energy is the energy required to initiate a chemical reaction. Enzymes lower the activation energy. □products of reactions have a low affinity for the active site and are released □ some metabolic reactions are reversible, the concentrations of substrate and end product concentration affect the direction and rate of an enzyme reaction
Control of metabolic pathways	□competitive inhibitors are molecules that resemble the substrate and compete with it for the active site of the enzyme □competitive inhibition can be reversed by increasing substrate concentration □non-competitive inhibitors are molecules that bind to the enzyme away from the active site but change the shape of the active site preventing the substrate from binding □non-competitive inhibition cannot be reversed by increasing substrate concentration □feedback inhibition occurs when the end-product in the metabolic pathway reaches a critical concentration □in feedback inhibition the end-product inhibits an earlier enzyme blocking the pathway and preventing further synthesis of the end-product.

HHB Unit 1.7 CELLULAR RESPIRATION LOs

	..by the end of the lesson(s) I should know…
glycolysis	□ cellular respiration is the release of energy from food molecules in cells □ glycolysis is the breakdown of glucose to pyruvate in the cytoplasm □ ATP is required for the phosphorylation of glucose and intermediates during the energy investment phase of glycolysis □ More ATP is generated in the energy pay-off phase, leading to a net gain of ATP □ Dehydrogenase enzymes remove hydrogen ions and electrons and pass them to the coenzyme NAD, forming NADH □ In aerobic conditions (when oxygen is present) pyruvate is broken down to an acetyl group. □ The acetyl group combines with coenzyme A to form acetyl coenzyme A
citric acid cycle	□ the citric acid cycle happens in the matrix of the mitochondria □ the acetyl group from acetyl coenzyme A combines with oxaloacetate to form citrate □ during a series of enzyme-controlled steps, citrate is gradually converted back into oxaloacetate □ ATP is generated □ carbon dioxide is released □ Dehydrogenase enzymes remove hydrogen ions and electrons and pass them to the coenzyme NAD, forming NADH
electron transport chain	□ The electron transport chain is a series of carrier proteins attached to the inner membrane of the mitochondria □ Hydrogen ions and electrons are passed to the electron transport chain by from NADH □ the electrons are passed along the electron transport chain releasing energy □ this energy is used to pump hydrogen ions across the inner mitochondrial membrane □ return flow of hydrogen ions, through the ATP synthase protein on the inner mitochondrial membrane, synthesises ATP □ most ATP is generated from the electron transport chain □ the final acceptor is oxygen, which combines with hydrogen ions and electrons to from water
ATP	□ ATP is used to transfer energy to cellular processes which require energy □ cellular processes which require energy include DNA replication, protein synthesis, muscle contraction

HHB Unit 1.8 ENERGY SYSTEMS IN MUSCLE CELLS LOs

	..by the end of the lesson(s) I should know…
lactate metabolism	□ during vigorous exercise the muscle cells do not get enough oxygen to support the electron transport chain □ in oxygen-deficient conditions pyruvate from glycolysis is converted to lactate □ hydrogen from the NADH formed during glycolysis is transferred to the pyruvate to produce lactate □ this regenerates NAD needed to maintain ATP production through glycolysis □ lactate accumulates in muscle cells when oxygen is in short supply during strenuous exercise □ lactate build-up in muscle cells can cause muscle fatigue & oxygen debt □ oxygen debt is repaid in a recovery period following strenuous exercise □ during the recovery time lactate is converted back into pyruvate and glucose in the liver
skeletal muscle fibres	□ skeletal muscle contracts to bring about the movements of the body □ skeletal muscle has slow-twitch fibres and fast-twitch fibres □ slow-twitch fibres contract more slowly but can sustain contractions for longer □ slow twitch muscle fibres - are useful for endurance activities e.g. long-distance running, cycling, cross country skiing - rely on aerobic respiration to generate ATP - have many mitochondria - have a large blood supply - have high concentration of oxygen storing protein called myoglobin - mainly store fuel as fat □ fast-twitch fibres contract more quickly over short periods and are effective for short bursts of strenuous activity e.g. sprinting, weight lifting □ fast twitch muscle fibres - can generate ATP through glycolysis only - have fewer mitochondria - have a lower blood supply - mainly store fuel as glycogen □ most human muscle tissue contains a mixture of slow & fast twitch fibres □ athletes show distinct patterns of muscle fibres that reflect their sporting activity