Module 5: Heredity

Reproduction

IQ1: How does reproduction ensure the continuity of a species?

★ Mechanisms of reproduction

- Reproduction = formation of an offspring by asexual or sexual processes

- Asexual and sexual reproduction

- Asexual reproduction = one parent and no gametes, resulting in offspring that are genetically identical to each other and the parent
- Sexual reproduction = two parents, who produce offspring that contain a mix of the parent's genes and differ from each other and from the parent

Type of reproduction	Advantages	Disadvantages
Sexual reproduction	• ↑ genetic variation of offspring - ↑ species survival and continuity • Offspring are more likely to be born with favourable traits under environmental selective pressures	• ↑ time and energy expenditure in finding a mate • Production of offspring is slower than asexual • Produces fewer offspring
Asexual reproduction	• Only requires a single organism to produce an offspring • Less energy intensive than sexual reproduction - population can increase rapidly and exploit suitable habitats quickly • Allows organisms to rapidly populate an environment	• Lack of genetic diversity - affect survival and continuity of species • Increases the susceptibility of species to new diseases

★ Advantages of internal and external fertilisation

- Sexual reproduction - internal and external fertilisation
- Fertilisation = union of male and female gametes and takes place externally or internally
- Internal fertilisation = involves male attraction and copulation (sexual intercourse)
    - Occurs in some invertebrates eg. insects and most vertebrates eg. mammals
    - The internally fertilised egg may develop a shell and be laid in the external environment (oviparous) to complete its development (in reptiles and birds) or it may continue to develop inside the female's body
- External fertilisation = occurs in aquatic or most terrestrial environments where both eggs and sperm are released into the water eg. amphibians and fish
    - Females release the eggs and the male releases the sperm which swim to the eggs in the environment - after the sperm reaches the egg, fertilisation occurs

- Release of reproductive material may be triggered by water temperature or the length of daylight
- Occurs in most invertebrates and some vertebrates eg. amphibians
- Mostly happens during spawning - one or several females release their eggs and the males release sperm in the same area at the same time

Types of fertilisation	Advantages	Disadvantages
Internal fertilisation	• Increased likelihood of fertilisation as egg and sperm as in close proximity - higher survival rates of offspring • Embryo is protected and more likely to survive in female reproductive tract	• Greater energy expenditure in locating a mate • Less offspring produced • More energy required to support offspring in gestation - mother becomes vulnerable in pregnancy • There are a higher risk of sexually transmitted infections passing between organisms
External fertilisation	• Little energy required to mate • Large number of offspring produced • Offspring can be spread widely, reducing competition with parent organisms • Fewer offspring are produced	• Zygote is exposed to the environment rather than protected inside the mother's body for internal fertilisation - limited defence capabilities of the zygote (eg. against predators), it is more susceptible to death • Requires a watery environment - more environmental factors such as harsh aquatic conditions

★ Plants: Asexual and sexual reproduction

- Plants - eukaryotic, multicellular autotrophic organisms
- Sexual reproduction - pollination
- Asexual reproduction - vegetative propagation
- Some plants can reproduce by asexual haploid spores - produces haploid offspring plant

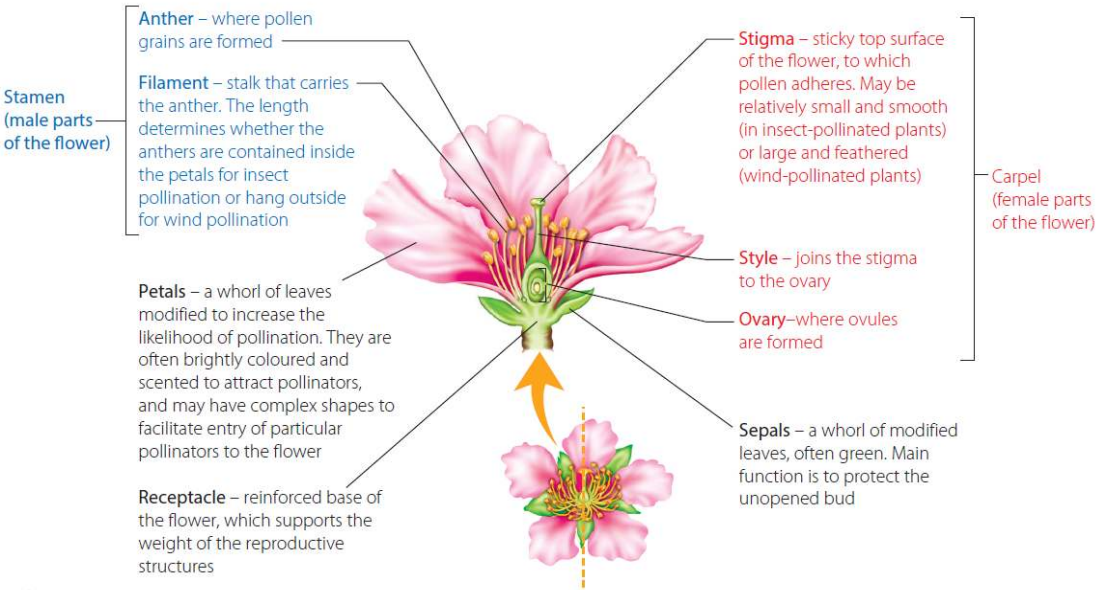
- Asexual reproduction

- Plants that reproduce asexually clone new individuals from portions of the root, stem, leaves or ovules of adult individuals eg. runners, rhizomes and suckers
- Common in harsh environments - organisms are so specific that there is little benefit in having variation within the population
- Eg. Runners - modified surface stem that forms a new plant eg. spinifex grass

Advantages	Disadvantages
• Enables organisms to reproduce quickly without having to find a mating partner • Being genetically identical may give an organism a competitive advantage in an environment to which they are well adapted • Economically advantageous for farmers to ensure consistency in their crops	• Pathogens may spread easily from parent to offspring • Reduction in genetic diversity increases the susceptibility of species to new diseases • Little or no variation in the population - whole group is particularly vulnerable to sudden changes in the environment

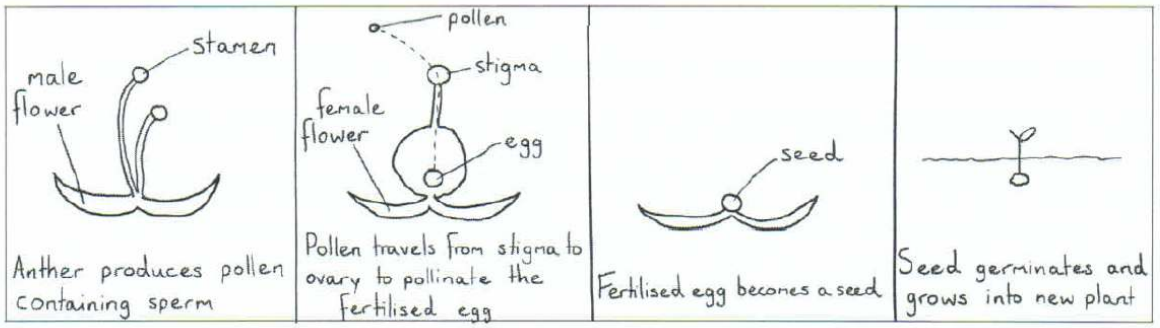
- Sexual reproduction

- **Flowers** = sexual reproductive organs of plants
- **Stamen** - male reproductive part
  - Each stamen consists of an anther at the top of a stalk called the filament
  - Anthers produce pollen which contain the male sex cells of a plant
  - Filament carries the anther - length determines whether the anthers are contained inside the petals for insect pollination or hang outside for wind pollination
- **Carpel** - female reproductive part
  - Pollinators help transfer pollen to the carpel
  - Has ovary at the bottom, a stalk called the style and a stigma at the top
  - Stigma - sticky surface where pollen adheres
  - Style - joins the stigma to the ovary
  - Ovary - eggs are fertilised by pollen will develop into the fruit of the plant



- Pollination

1) Male gametes inside the pollen must be carried from the anthers to the stigma
2) Once pollen has been deposited on the stigma, a pollen tube germinates and grows down the style, carrying inside it the male gamete
3) The pollen tube fuses with the egg cell in the ovule, which is contained in the ovary
4) The fertilised ovule develops into an embryo, which is now a seed
5) The ovary grows to become a fruit



Self-pollination	Cross-pollination
• Plant pollinates itself • Pollen matures and the anthers split releasing the pollen which is usually deposed on its own stigma • Desirable to ensure survival if reproductive partners are scarce	• Pollen is transferred from the anther of one plant to the stigma of another plant • Transfer occurs by wind or by vector, eg. animals like birds, insects or mammals • Advantageous to a species - increases genetic variation and ensures survival is a sudden environmental change eg. disease or drought

- Seed dispersal

- Advantage for seeds to be dispersed over a wide distance - prevents overcrowding and competition for light, water and soil nutrients
- Seed dispersal relies on the type of fruit in which the seed occur, matching the type of dispersal agent available in that environment
- Dry fruits often have inbuilt ‘explosive’ mechanisms for dispersal by air, wind or water
- They are usually light so they can float on air or water
- Fleshy fruits often rely on insects, birds or mammals for dispersal (biotic agent) - the animals eat the fruit, move along and then egest the seeds, usually some distance away from the parent plant

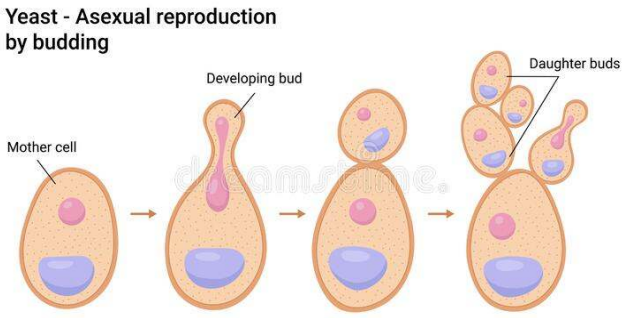
★ Fungi: Budding, spores

- Unicellular fungi - budding
- Multicellular fungi - spore formation

- Budding

- Primarily asexual, some instances of sexual
- An adult organism gives rise to a small bud, which separates from the parent and grows into a new individual
- Creates daughter cells of unequal sizes
- Eg. Yeast
    - Microscopic unicellular organisms, classified as fungi
    - When environmental conditions are favourable, a small bud develops on the parent cell, which enlarges, leading to the parent cell replicating its DNA
    - The nucleus divides and one part goes into daughter cell

- When the daughter cell reaches a certain size, it detaches from the parent cell and continues to grow, until it buds in turn
- Lack of nutrients in yeast cells - use sexual reproduction instead

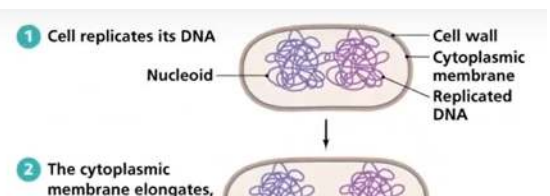


- Spores

- Asexual spore formation - release of haploid spores - haploid spore germinates into a haploid fungi
- Sexual spore formation - hyphae of two different fungi combining haploid spores - creating a diploid fungal zygote - diploid zygote produces haploid spores via meiosis
- Fungi usually produce spores inside a sporangium - when this disintegrates, the spores are released and dispersed through the wind
- When the spore lands in a suitable environment, it germinates and forms a new fungus
- Each spore has several nuclei and some cytoplasm, surrounded by a wall
- Under favourable environmental conditions, fungal spores germinate, absorbing water through the wall, which activates the cytoplasm to grow
- Nuclear division occur, more cytoplasm grows and the spore grows into a new mycelium - enables fungi to reproduce rapidly, colonise a wide area and ensure continuity

★ Bacteria: Binary fission

- Bacteria - unicellular, prokaryotic, asexual
- Involves a single cells splitting into two identical daughter cells
- No nucleus in bacteria - no splitting of cell nucleus
- Different to mitosis - mitosis mainly occurs in eukaryotes (eg. plant and animal cells)
  - Purpose is different - mitosis (repair, maintenance and growth)
  - Bacteria reproduce by binary fission



★ Protists: Binary fission, Budding

- Unicellular, eukaryotic, microscopic organisms
- Reproduce by binary fission - asexual
- Reproduce by budding - generally asexual

- Binary fission

- Different to bacteria - DNA is stored in the nucleus (no nucleus in bacteria)
- Involves mitosis and the formation of a spindle within the cytoplasm of a cell to distribute chromosomes equally
- Reproduce sexually by conjugation - temporary union of two individuals to exchange haploid pronuclear to form a zygote nucleus
- Effects:
    - Rapid population growth in a short period of time in adverse conditions
    - No genetic diversity (introduced only through conjugation)

- Budding

- The parent protozoan producing a bud which is a daughter nucleus that is created based on the replicate of a nucleus DNA, followed by equal nucleus division but unequal separation of the parent protozoan's cytoplasm
- The bud is smaller than the parent
- Over time, the daughter nucleus undergoes further cell division via mitosis to grow and mature, resulting in a protists that is genetically ideal to parent

★ Features of fertilisation

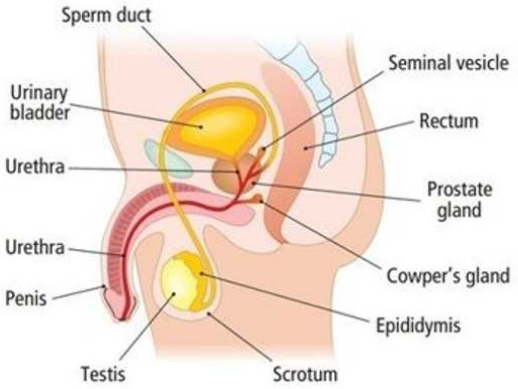
- **Fertilisation** = fusion of two haploid gametes to form a diploid zygote
- Male inserts his penis in the female's vagina and ejaculation pushes semen from his urethra into her vagina
- From the vagina, sperm swim using their flagella to propel forward, through the cervix into the uterus and into an oviduct until one sperm reaches the egg

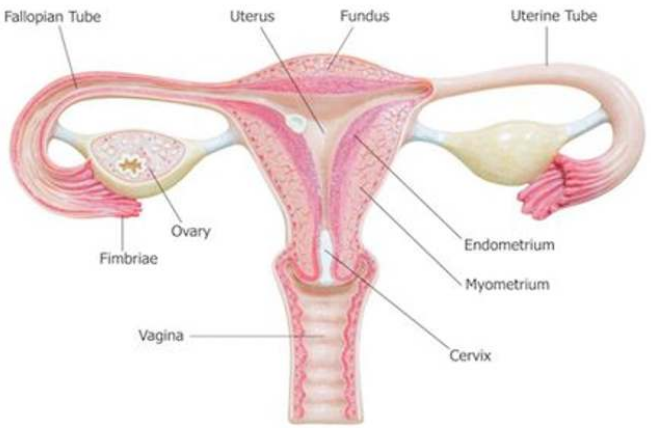
- Gametes

Sperm	Ovum

• Male gamete can move by the motion of a flagellum • Tip of its head contains a bag of digestive enzymes, called the acrosome • Acrosome helps to break down the protective layer of the ovum - the zona pellucida - sperm can reach the ovum's cell membrane	• Female gamete • When released, it is still surrounded by some follicular cells, as well as a thick protein layer called the zona pellucida - facilitates sperm binding and prevents polyspermy (more than one sperm fertilising the egg)

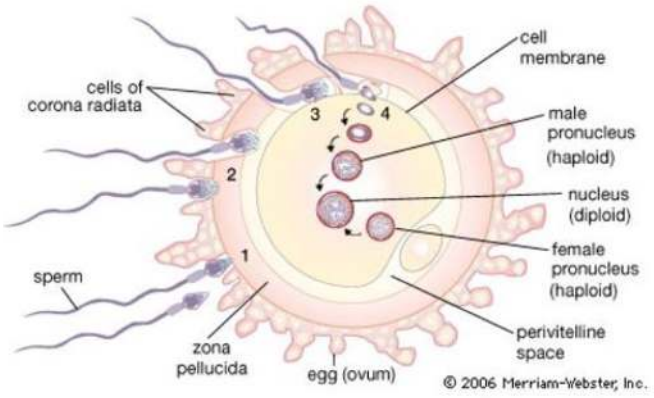
- Male and female reproductive system

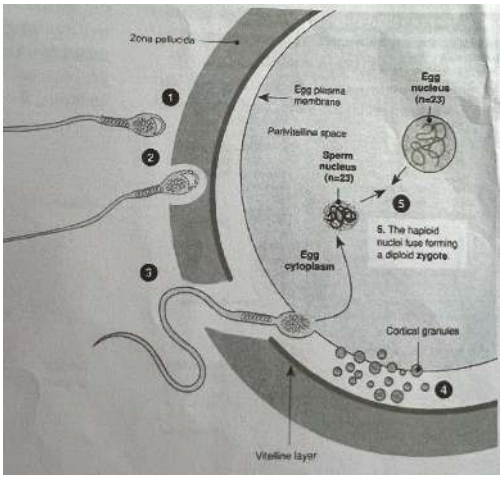




- Stages of fertilisation

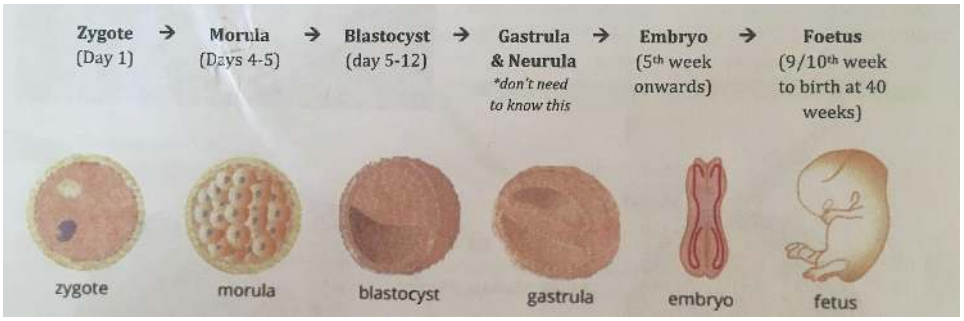
1) Membrane of the sperm cell undergoes changes so that it can fertilise an egg
2) Sperm binds to the zona pellucida and enzymes from the acrosome are released to breakdown a pathway through the zona pellucida - allows the sperm to reach the cell membrane of the ovum
3) Cell membrane of the sperm and ovum fuse and the nucleus of the sperm enters the ovum cytoplasm
4) Fusion of two cell membranes causes a permanent change in the ovum surface that prevents the entry of multiple sperm
5) Fusion of the haploid egg and sperm results in a diploid zygote cell (fertilised egg)

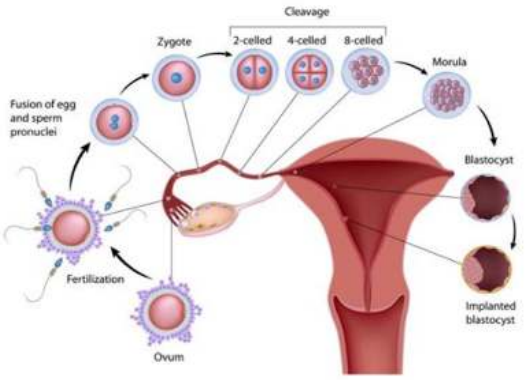


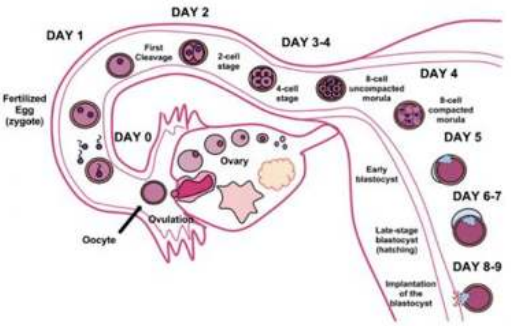


★ Implantation in mammals

- After fertilisation, the zygote travels down the fallopian tube towards the uterus
- A week after fertilisation - implantation occurs
- **Implantation** = attachment of the blastocyst to the endometrium
- The endometrium provides nutrients and oxygen to the embryo







- Placenta

- **Placenta** - embedded into the uterine wall and is connected to the foetus by the umbilical cord
- Role - transport oxygen and nutrients from mother to foetus and to remove wastes such as carbon dioxide and urea from the foetus and pass them to the mother's blood for excretion
- 10 weeks - placenta takes over progesterone production and the corpus luteum deteriorates
- At the end of pregnancy, the placenta loses competency and progesterone levels falls

★ Hormonal control of pregnancy

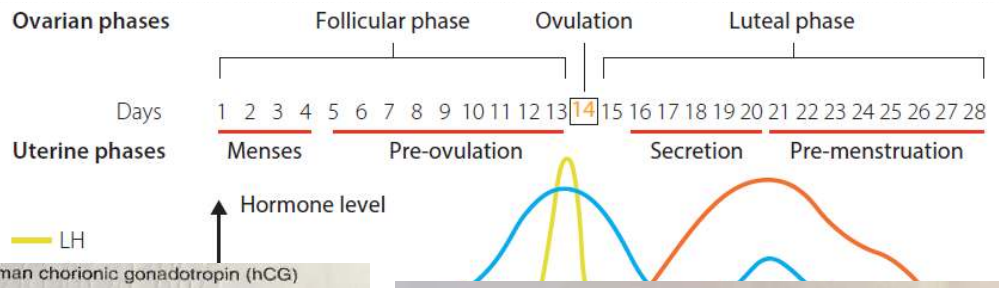
Name	Origin	Effect

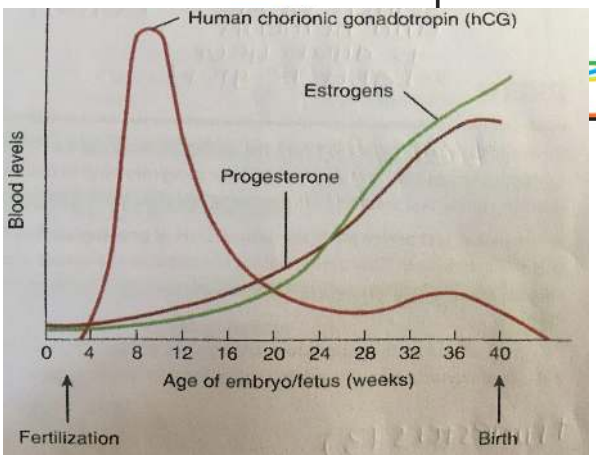
hCG	Corpus luteum	• Produced by the placenta after implantation • Responsible for sustaining the secretion of progesterone and oestrogen by the corpus luteum • Causes pituitary gland to secrete FSH and LH - travel to ovaries • 1st trimester - hCG ↑ rapidly to maintain the corpus luteum • 2nd trimester - hCG ↓ and the corpus luteum deteriorates which stops it from producing progesterone and oestrogen • The placenta takes over and produces oestrogen and progesterone
Luteinizing hormone (LH)	Pituitary gland	• Maintains the corpus luteum, preventing its breakdown in later half of ovarian cycle • Maintenance of corpus luteum is taken over by hCG following implantation • Surge at day 14 triggers ovulation • LH levels ↓ once progesterone and oestrogen levels rise
Follicle stimulating hormone (FSH)	Pituitary gland	• Promotes follicle development and secretion of oestrogen
Oestrogen Increase steadily	Ovary	• Stimulates the growth of the endometrial lining • Promotes organ development in the foetus and the growth of breast tissue for milk production
Progesterone Decreases slightly prior to birth	Ovary	• Maintains endometrial lining • Prevents uterine contractions

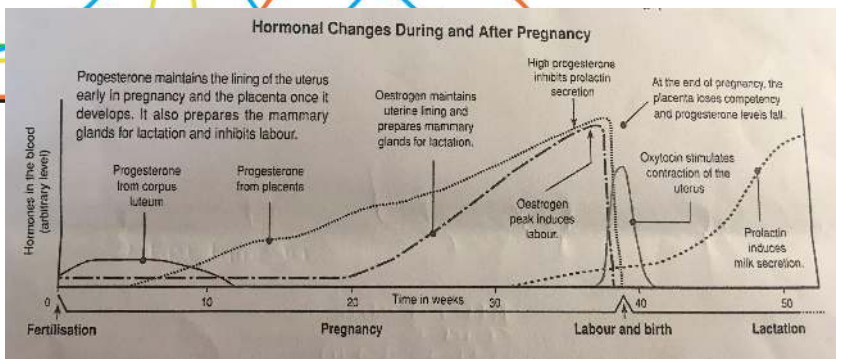
- Female reproductive graph

- 1 - first day of menstruation
    - Bleeding that occurs after ovulation
    - Caused by a fall in the levels of oestrogen and progesterone - decreased the blood supply to the endometrium and causes shedding
- Day 14 - Oestrogen is produced, causing a surge in LH
    - LH is responsible for ovulation
- Corpus luteum responsible for the secretion of oestrogen and progesterone

- Day 21 - progesterone decreases as the corpus luteum degenerates
- Drop in oestrogen and progesterone - no implantation has occurred
- Eventually causes bleeding - marks the next reproductive cycle







★ Birth

- Towards the end of the third trimester, increased oestrogen is released
- Placenta releases prostaglandins - initiates labour and stimulates contractions
- Progesterone levels decline during labour
- Oxytocin causes contractions of the uterus and softening of the cervix - released from pituitary gland
- Positive feedback loop:

1) Head of foetus pushes against cervix
2) Nerve impulses from cervix transmitted to brain
3) Brain stimulates pituitary gland to secrete oxytocin
4) Oxytocin carried in bloodstream to uterus
5) Oxytocin stimulates uterine contractions and pushes foetus toward cervix

★ Manipulation of plant and animal reproduction in agriculture

- Growth of the agricultural industry has been driven by two factors: increasing the number of offspring in each generation and controlling the passing of favourable traits within each generation
- Agriculture must become sustainable for the long-term survival of humans and the environment

- Plant reproduction: Artificial pollination

- Transfer of pollen from the anther of a flower on one plant to the stigma of a flower on the same or another plant to ensure desirable characteristics
- Able to cross-breed with favourable traits

- Ensures pollination of all plants, resulting in high crop yields

- Animal reproduction: Artificial insemination

- Method by which two animals may be forcibly bred to produce favourable offspring
- Involves collecting semen from the male and inserting it into the female with an insemination gun
- Ensures genetic material containing favourable traits is passed onto the next generation, increasing the quality of agricultural products
- Increases the likelihood of successful pregnancy as semen may be screened and the process can be timed with females' fertility cycles
- Birth may be synchronised in a population, which is useful on a large scale

CELL REPLICATION

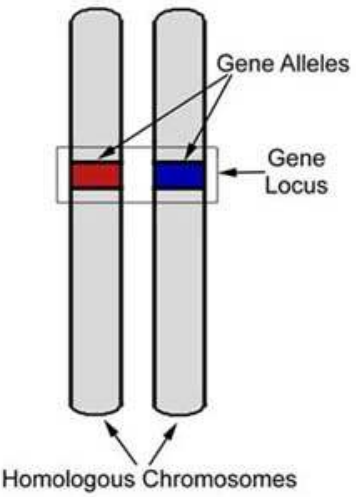
IQ2: How important is it for genetic material to be replicated exactly?

★ Modelling the processes: Mitosis

- Occurs in all body tissues by somatic cells
- Produces two genetically identical daughter cells
- Some organisms can use mitosis to reproduce asexually
- Purpose - growth, repair and maintenance of somatic cells

- Cellular Structures

- **Homologous chromosome** = pair of each chromosomes, maternal and paternal
    - Similar in length, same genes in same order and location along the chromosome
    - Not identical - alleles (different forms of the same gene) for the genes may differ
    - Eg. gene for hair and the allele would be black or brown
- **Genetic locus** = location of a particular gene on a chromosome
    - At each genetic locus, an individual has two alleles, one on each homologous chromosome
- **Chromatin** = material - consists of DNA and histones - condenses to form chromosomes
- **Chromosomes** = carry genetic information in the form of genes
    - When chromosomes separate - chromatids
- **Chromatid** = one half of a replicated chromosome
    - When the DNA replicates, sister chromatids are attached via the centromere
- Count the number of chromosomes by the **centromere**

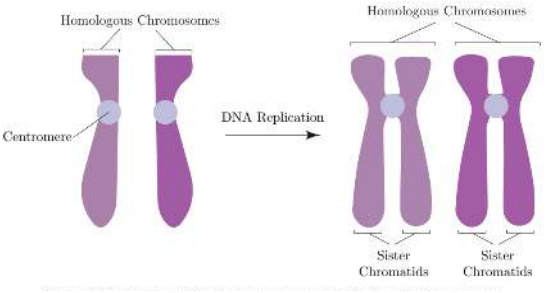


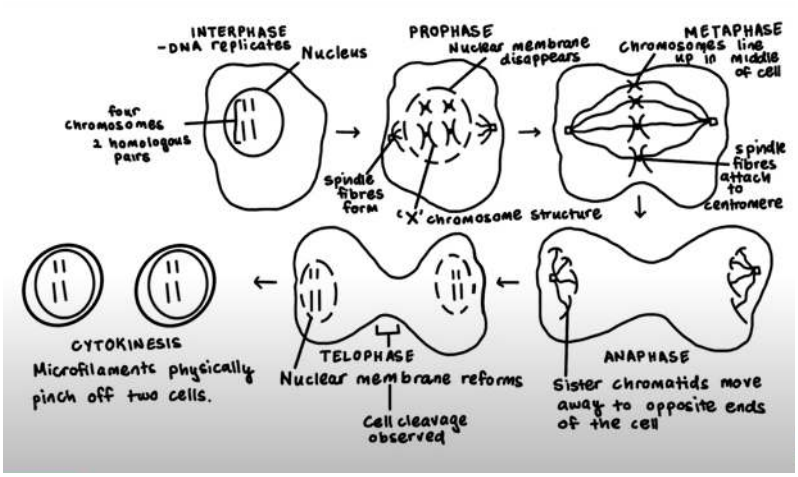
- Cell Cycle

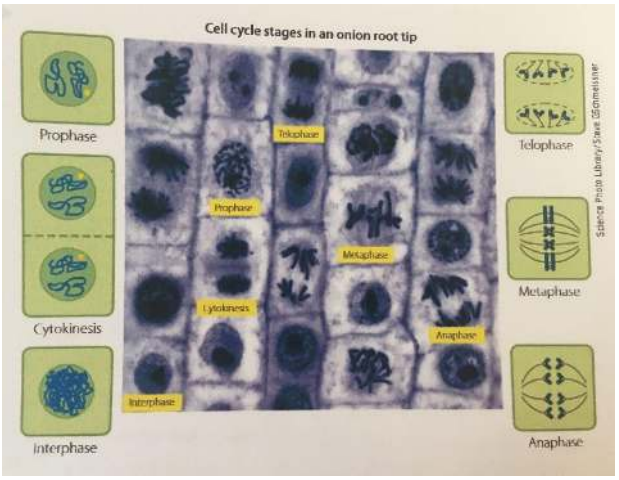
- Majority of the cell's life is in interphase - preparing for division
- S phase - DNA replication

- Stages of Mitosis (PMAT)

- Mitosis occurs after interphase and before cytokinesis

Interphase - DNA replication	• Occurs before mitosis • DNA replication occurs • Chromosomes are not visible - DNA is in the form of chromatin 
Prophase - Pair up	• Chromatin condenses into chromosomes - visible • Centrioles move to opposite ends of the cell and form spindle fibres • Spindle attaches to the centromeres of chromosomes • Nuclear membrane disappears
Metaphase - Middle	• Sister chromosomes line up along the equator of the cell • Each is attached to the spindle fibres by a centromere
Anaphase - Away	• Centromeres split and sister chromatids separate and move to opposite poles of the cell
Telophase - Tear away - Opposite of prophase	• The cleavage furrow forms as the cytoplasm begins to divide into two cells • Nuclear membrane reforms • Spindle fibre disappears • Chromosomes decondenses to becomes chromatin • Results in two identical nuclei
Cytokinesis - Create new cells	• Cytoplasm divides and cells split into two • Results in two identical daughter cells - 46 chromosomes each





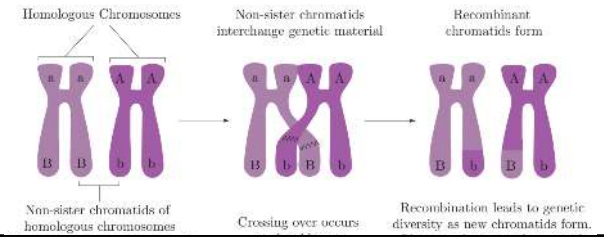
★ Modelling the processes: Meiosis

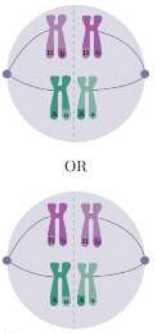
- Produces four daughter cells that are genetically unique with haploid (n) of chromosomes - when two gametes combine via fertilisation, they form a diploid zygote with the full complement of chromosomes
- Occurs during the production of gametes in the gonads:
    - Female - haploid egg cell
    - Male - haploid sperm cell
- Source of genetic variation - induce a change in the genes of an individual, resulting in genetically identical offspring with a varied combination of traits

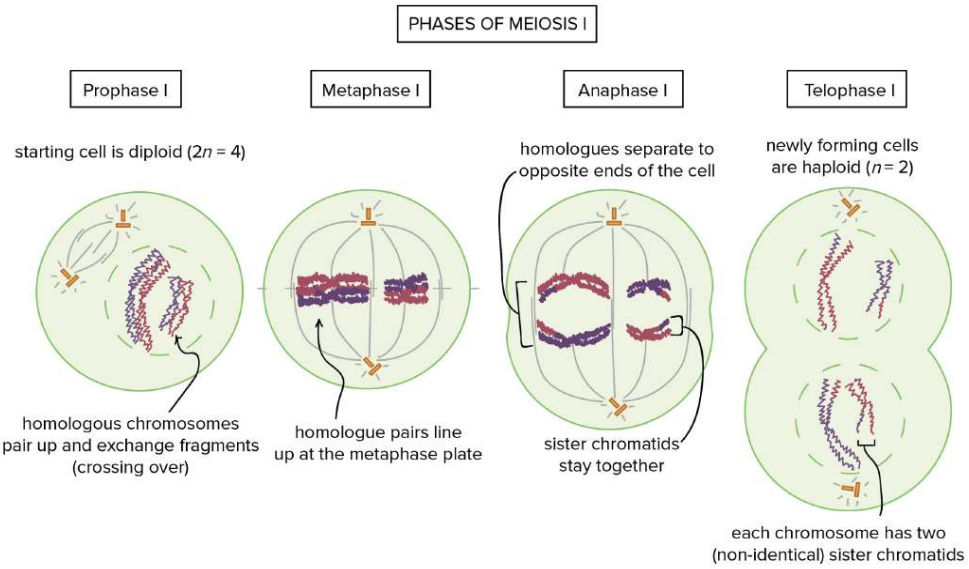
1) Crossing over - homologous chromosomes exchange genetic material during meiosis
2) Independent assortment - meiosis randomly separates the homologous chromosomes
3) Fertilisation - 2 parents contribute half of the genetic material to the offspring
4) Mutation of chromosomes and genes creates new genes and new traits

- Meiosis I

- Separates pairs of homologous chromosomes into two separate cells

Interphase	• Occurs before meiosis • Same as mitosis - DNA replicates
Prophase I	• Chromosomes form - nuclear membrane breaks down • Homologous chromosomes pair up in tetrads • **Crossing** may occur - introduces genetic variation in offspring • Non-sister chromatids in a homologous pair exchange pieces of DNA to form recombinant chromatids • The point of contact - chiasma • At the chiasma, the chromatids can exchange genetic information • Two new recombinant chromatids are formed 

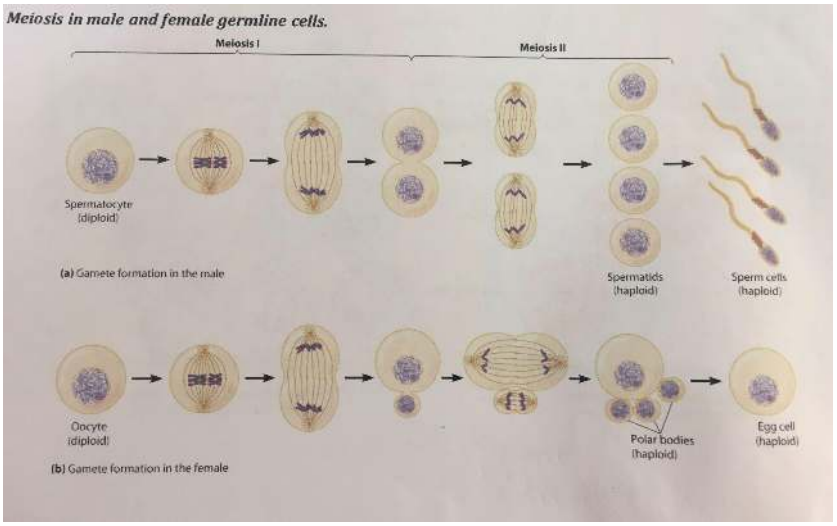
Metaphase I	• Homologous chromosomes pair align in the middle of the cell - arrangement of each pair of chromosomes occur randomly and is unaffected by the positioning of any other pair • **Independent assortment** occurs - each pair lines up independently of the other pairs i.e. the maternal and paternal chromosomes do not line up on the same side of the midline • The random different positions of chromosomes along the midline results in gametes with different combinations of maternal and paternal chromosomes • Spindle fibres then attach to the centromere of the aligned chromosome  OR Figure 4.5: Independent Assortment Page 30 of
Anaphase I - **Identical to mitosis**	• Homologous chromosomes pairs - may or may not have undergone crossing over - separate to opposite ends of the cell • The centromeres do not split - pairs that separate, not the chromatids - random segregation • The law of segregation - two alleles of a single trait will separate randomly - each gamete receives only one allele of that gene • Produces different combinations of genes in different gametes
Telophase I - **Identical to mitosis**	• The nuclear membrane begins to form around each set of separated chromosomes • Division of the cytoplasm begins to take place
Cytokinesis I - **Identical to mitosis**	• First division of the cytoplasm occurs - two daughter cells form • Each daughter cell contains only one of each chromosome pair - the chromosomes are still made up of two chromatids • The chromosome combinations in the daughter cells differ (combination of maternal and paternal chromosome)

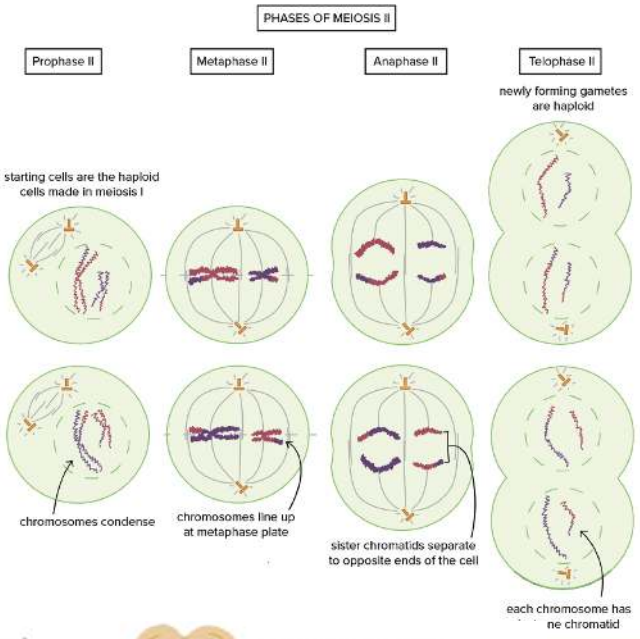


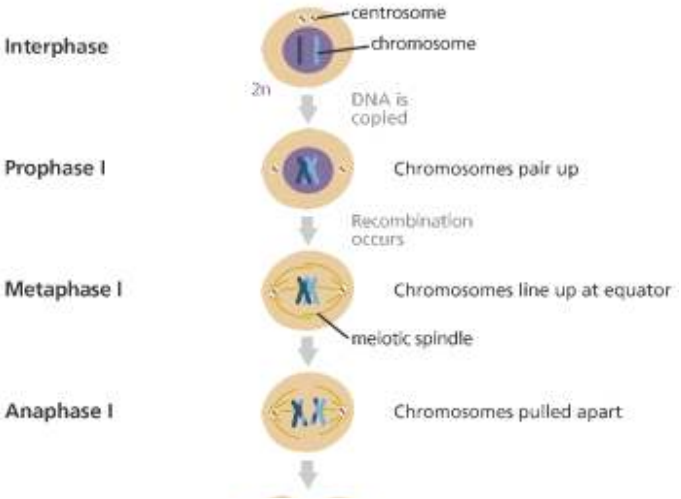
- Meiosis II

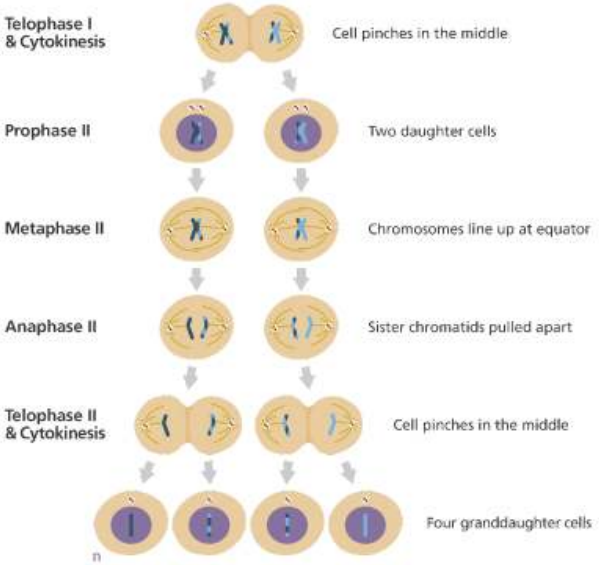
- DNA replication does not occur again
- Chromosomes line up once more in the centre of the cell and the spindle fibres separate the sister chromatids

Prophase II	• Nuclear membrane breaks down and spindle fibres begin to form
Metaphase II	• Chromosomes randomly line up at the equator • Chromosomes - no longer paired • Sister chromatids are still attached via centromere
Anaphase II	• Spindle fibres attach to the centromeres and pull the sister chromatids apart to opposite ends of the cell • Separation of the chromatids occurs randomly
Telophase II	• The nuclear membrane forms around each set of chromatids • The division of the cytoplasm begins to take place
Cytokinesis II	• Second division of the cytoplasm takes place, producing 4 genetically different haploid gametes • The chromatids uncoil









- Difference between mitosis and meiosis

	Mitosis	**Meiosis**
Daughter cells produced	2	4
Haploid or diploid	Diploid	Haploid
Identical or different	Identical	Different
Where the process occurs	Somatic cells	Gonads (ovaries and testes)
Number of divisions	1	2
Chromosome arrangement/ behaviour	Chromosomes do not cross over and independent assortment has no effect	Homologous chromosomes pair up in Prophase I and crossing over occurs. Independent assortment occurs in Metaphase 1

★ DNA replication using the Watson and Crick DNA model

- During resting stages of mitosis or meiosis, genetic material duplicates itself for cell division
- **DNA replication** = process by which an identical copy of DNA is made
- Splitting a DNA molecule into 2 separate strands - each strand can be used to build a new, complementary strand - ensures that the genetic material is copied exactly
- Two strands of DNA run in opposite directions - one runs 5' → 3' while other runs 3' → 5'
    - Important for replication as free nucleotides can only be added in one direction only - from 5' → 3' - are added onto the free 3' end
    - DNA polymerase can only add nucleotides to the 3' end

- Nucleotide composition

- Each nucleotide consists of three parts - phosphate, sugar (deoxyribose sugar in DNA, ribose sugar in RNA) and a nitrogen base
- Four types of nitrogen bases - adenine, thymine, guanine or cytosine nucleotides
- Each DNA molecule is made up of two chains or strands that have an antiparallel arrangement
- Each strand is made up of a sequence of nucleotides and held together by weak hydrogen bonds between the bases in the centre of the DNA molecule
- Advantage of weak hydrogen bonds - little effort is required to pull the bases apart so that DNA can replicate or be decoded to form proteins

- Watson and Crick DNA model

- Watson and Crick's DNA model showed all the requirements expected of hereditary material
- DNA can carry, in coded form, all the instructions for the formation and functioning of cells
- The structure of DNA allows for its own replication - to copy DNA, each strand can serve as a template for enzymes to synthesise a new complementary DNA strand
- DNA can be transferred from one generation to the next, packaged in the form of chromosomes and carried by gametes - DNA is coiled around histones and keeps DNA 'neatly packaged'
- Histones expose sections of DNA so that genes can be expressed

- Process of DNA replication

1) DNA double helix unwinds

- Enzyme helicase causes the DNA helix to unwind and the strands to separate by breaking the bonds between the nitrogen base pairs
- Two parent DNA strands separates, exposing the nucleotide bases
- Creates a replication fork where the double stranded DNA is being separated

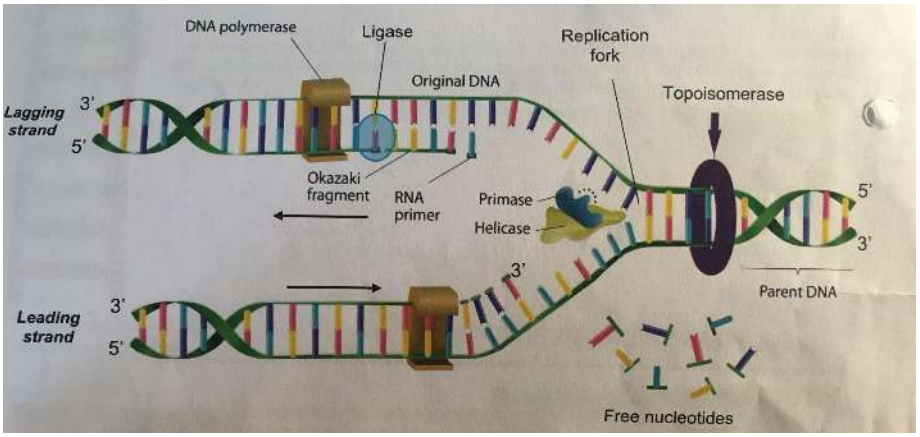
2) Nucleotides are added to the exposed single strand

- RNA known as primer needs to be made and attached to the DNA
- DNA polymerase III adds free nucleotides floating in the nucleus to the exposed bases on the template strand according to the nitrogen base pairings - A-T, C-G
- DNA polymerase acts by joining nucleotides to the 3' end
- On the template strand that runs 3' - 5', nucleotides are added continuously in the 5' - 3' direction in a long, chain growing in the same direction as the replication fork opens up
- Newly synthesised strand is called the leading strand

- On the other template strand (runs from 5’ - 3’), nucleotides are added discontinuously in short sections known as Okazaki fragments - lagging strand
- Replication along the lagging strand is backwards from the replication fork
- Enzyme ligase - connects and seals Okazaki fragments

3) Replication errors are identified and corrected

- DNA polymerase reaches the end of the DNA molecules and two identical daughter strands are produced
- DNA polymerase I - recognises and repairs base pairing errors by cutting out the incorrect base and inserting the correct one - removes any RNA primers and replaces them with correct nucleotides
- The new double-stranded DNA molecule rewind into helix conformation
- Each contains one strand of the original DNA molecule and a newly synthesised strand
- DNA replication is termed semi-conservative



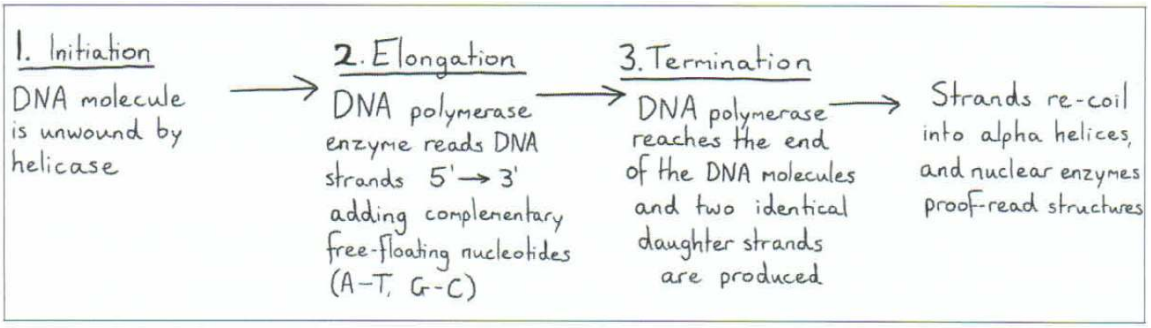
- Consequences of unrepaired DNA replication errors

- If there is minor damage, such as DNA breakages or base mismatches, these will be corrected by enzymes before the cell continues its progress through the cell cycle
- Without repairs to any faulty DNA, the cell may not be able to make the correct proteins - cell may malfunction or it could become a cancerous cell
- Mutations of DNA may affect a single gene or multiple genes - result in the formation of a non-functional protein or a new allele
- Despite this very high rate of accuracy, mistakes or damage to a cell’s DNA can occur during DNA replication
- If a mutation occurs during DNA replication or cell division that is not identified by the checkpoints and repaired, the uncorrected mutation will be replicated in successive divisions and is transmitted to each new daughter cell each time the cell divides
- Meiosis - uncorrected mutations will be passed on to later generations of individuals

★ Effect of cell replication processes on the continuity of species

- Continuity of the species - ongoing survival of species as a result of characteristics being passed from parents to offspring in a continuous lineage
- Accurate DNA replication brings about genetic stability and the consistent passing on of genetic information from parent to offspring
- Mutation results in genetic variation
- Variation is important for the evolution of species - genetic stability is important for the survival of the individual

- Genetic continuity relies on:
- Consistent replication of genetic information that is passed from a parent cell to daughter cells, resulting in continuity in the traits being passed from parent to offspring
    - Ensures that new cells or organisms have all the genes they need to survive
- The effect of natural selection and evolution on the gene pool as a result of:
    - Introduction of variation during sexual reproduction
    - Random errors arising by mutation, being replicated and passed onto offspring
- Random variation that confers an advantage may be selected over those that confer no advantage or are harmful



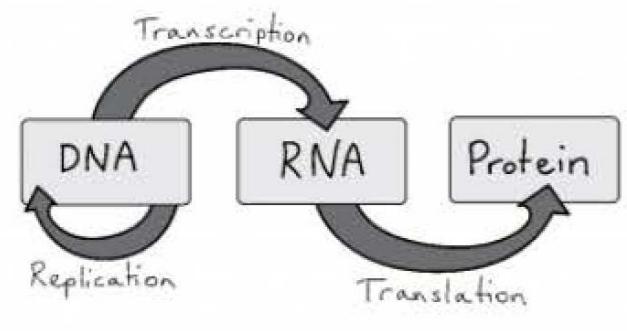
DNA AND POLYPEPTIDE SYNTHESIS

IQ3: Why is polypeptide synthesis important?

★ Comparing the Forms of DNA: Prokaryotic and Eukaryotic

Prokaryotic DNA	Eukaryotic DNA
• DNA is circular and double-stranded • The DNA does not have the histone protein framework of eukaryotic cells • Have non-chromosomal DNA in the form of plasmids (rings of DNA) floating in the cytoplasm	• Has a nucleus where DNA is stored • Each chromosome is larger and more complex • Linear, double-stranded helix • Also have non-nuclear DNA • Mitochondria and chloroplasts also contain their own DNA - inherited independently of nuclear DNA

★ The process of polypeptide synthesis



- Structures involved in polypeptide synthesis

- **RNA** - instead of deoxyribose as the sugar, it has ribose sugar
    - Single stranded
    - Instead of thymine, there is uracil that bonds to adenine
- Two forms of RNA: mRNA and tRNA
- DNA does not leave the nucleus - molecules are too large to pass through the pores in the nuclear membrane ∴ mRNA is created
    - **mRNA** - carries a transcribed copy of the relevant instructions from the nucleus to the ribosomes in the cytoplasm
    - Small enough to pass through nuclear pores and carries the genetic code outside the nucleus, into the cytoplasm, where it can be read by ribosomes
    - RNA polymerase controls the formation of mRNA
    - **Ribosomes** translates the message carried by the mRNA into a polypeptide
- **Transfer RNA (tRNA)** carries the amino acids to the ribosomes to link and form a polypeptide chain
    - Different type for every amino acid
    - At the bottom of every tRNA molecule - anti-codon that binds to the codon on the mRNA strand - how the amino acid is linked to the codon
- **Ribosomes** = found in the cytoplasm, active site for protein synthesis
    - Made up of protein and RNA molecules
    - Can accommodate 2 tRNA at a time
- Set of 3 bases on the mRNA - **triplet code** or **codon**
    - Every codon codes for one amino acid - 20 different amino acids
    - One amino acid can have more than one triplet code

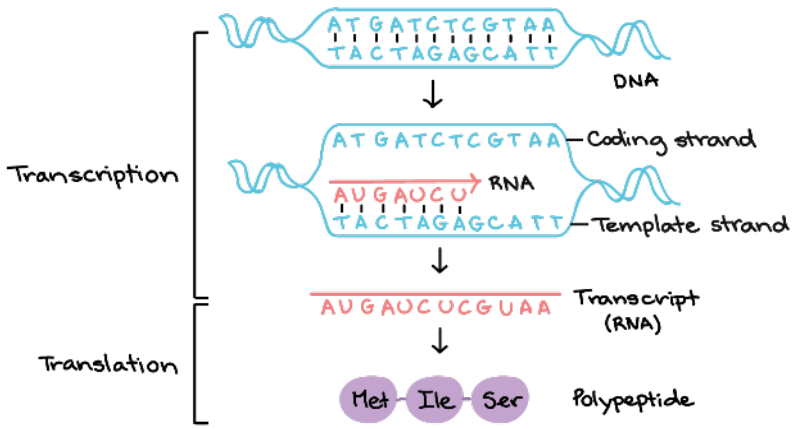
★ Transcription

- Occurs in the nucleus
- Turns DNA into mRNA
- RNA polymerase binds to the promoter region and the DNA ‘unzips’
- RNA polymerase moves along the DNA template strand (3’ - 5’), creating a complementary mRNA strand (5’ - 3’) according to the pairings A-U and C-G
- Transcription stops when the RNA polymerase reaches the stop codon
- New RNA strand is called the primary RNA transcript
- Both exons and introns are transcribed to produce a long primary RNA transcript
- DNA then re-binds
- Introns are removed by splicing from the primary RNA transcript to form a mature messenger RNA
- mRNA strand exits the nucleus and enters the cytoplasm

★ Translation

- Occurs in the cytoplasm
- Translates mRNA into a polypeptide chain
- Codon = group of three bases on the mRNA
- The sequence of ribonucleotides in the mRNA acts as a template for a corresponding tRNA anticodon
- Ribosomes bind to the mRNA molecule and matches the mRNA codons to complementary anticodons on tRNA

- The amino acid on each matching tRNA is added to a chain to form a polypeptide
- When a ‘stop’ codon is reached, the polypeptide chain detaches from the ribosome and is released into the cytoplasm



★ Importance of mRNA and tRNA in transcription and translation

- mRNA - transcription

- Transcribes the genetic instructions in DNA for making a polypeptide
- Bases in mRNA is determined by the complementary sequence on the template strand
- mRNA transports message from the nucleus to ribosomes where polypeptide synthesis occurs
- Each codon on the mRNA molecule attracts a tRNA molecule, showing the anticodon and carrying a specific amino acid
- Sequence of codons on the mRNA molecule determine the sequence of amino acids in the resulting polypeptide

- tRNA

- Carries anticodons, as well as a corresponding amino acid
- The tRNA anticodons bind to the complementary base pairs in the mRNA codon to ensure the correct amino acid is added to the polypeptide being formed

★ Function and importance of polypeptide synthesis

- Allows for the genetic code of organisms, stored as DNA to be read and expressed as proteins - ensures that cells function effectively
- Also produces proteins which are the functional and structural framework of cells
- Important for cell functioning as it enables cells to carry out the functions necessary for life
- Particular base sequences of each DNA molecule determine which polypeptides are formed in the cells of organisms
- Any changes to the DNA base sequences as a result of mutations can affect protein production
- Cell activity is also affected - non-functional protein is produced as a result of mutation
- Changed base sequence can lead to genetic disorders and inherited diseases

★ Genes and environment affecting phenotypic expression

- **Genotype** = combination of genes an organism has

- **Phenotype** = physical appearance of an organism, as determined by its structure, behaviour and physiology

- Gene expression and phenotype

- **Gene expression** = switching on and off of genes to make the required proteins and other end products in particular cell types
- Products determine the physical and chemical features typical of each cell type and the overall phenotype of an organism
- **Phenotypic expression** - result of gene expression - the structural, physiological and behaviour of an individual as a result of genes that have been expressed
- Phenotype depends upon:
    - Factors controlling transcription and translation during protein synthesis
    - Product of other genes
    - The environment
- **'Epigenetic' modifications** = chemical modification of DNA that does not involve a change in the sequence of nucleotides
- May be the mechanism by which some environmental factors bring about variation
- Result is a change in phenotype without a change in genotype
- Epigenetic changes shows links to disease, including cancers and metabolic diseases

- Environmental effects on gene expression and phenotype

- Phenotype depends on both genotype (genes) as well as the environment
- Identical twins have identical genotypes - any phenotypic differences can be attributed to environmental influences
- Eg of variation brought about by the environment - difference in colours of flowers in hydrangeas
    - The acidity or alkalinity of the soil influences the colour of the flower
    - Hydrangeas growing in acidic soil develop blue flowers
    - Those grown in alkaline soil develop pink flowers

★ Structure and function of proteins

- Structure

- Made up of one or more long chains of amino acids
- Each chain is called a polypeptide
- Proteins made up of a single polypeptide chain have primary, secondary, tertiary structure
    - Simple - single chain of amino acid
    - Secondary - 3D arrangement of chain
    - Tertiary - polypeptide folded into complex 3D shape
    - Quaternary - 2 or more linked polypeptide chains

- Made up of one or more polypeptide chains which are composed of amino acids
- Protein structure is crucial to the functioning of the protein as the protein's shape enables its function

- The sequence of the amino acids in a protein's primary structure determines the folding pattern of the polypeptide chain in its secondary structure and in turn, its three-dimensional folding at the teritary level of structure

- Some proteins have a quaternary structure - multiple polypetide chains are joined together and operate as a single functional unit
- Any changes to the amino acid sequence will alter the protein's 3D structure at the secondary and teritrary level, thus affecting its function

- Function

- Provides structure and support for cells, forming the basis of the cytoskeleton which gives cell their shape and allows movement
- Enzymes - type of protein that catalyse biological reactions and are essential to ensure metabolic reactions occur quickly enough to sustain life
- Storage - Used to help store substances for later use

GENETIC VARIATION

IQ4: How can the genetic similarities and differences within and between species be compared?

★ Practicals to predict variations in genotypes of offspring

1) **Mutation** - permanent change in the genetic information
- Can be point mutations with a single nucleotide pair substitution, insertions or deletions, or large scale chromosomal mutations with duplication and alteration of chromosome structure

2) **Meiosis** - type of cell division in sexually reproducing organisms that leads to the formation of haploid daughter cells
- Crossing over and random segregation of chromsomes - processes that lead to variation
- Law of segregation - two alleles for each gene separate during meiosis and ends up in different gametes

3) **Fertilisation** - union of two haploid gametes to form a diploid zygote

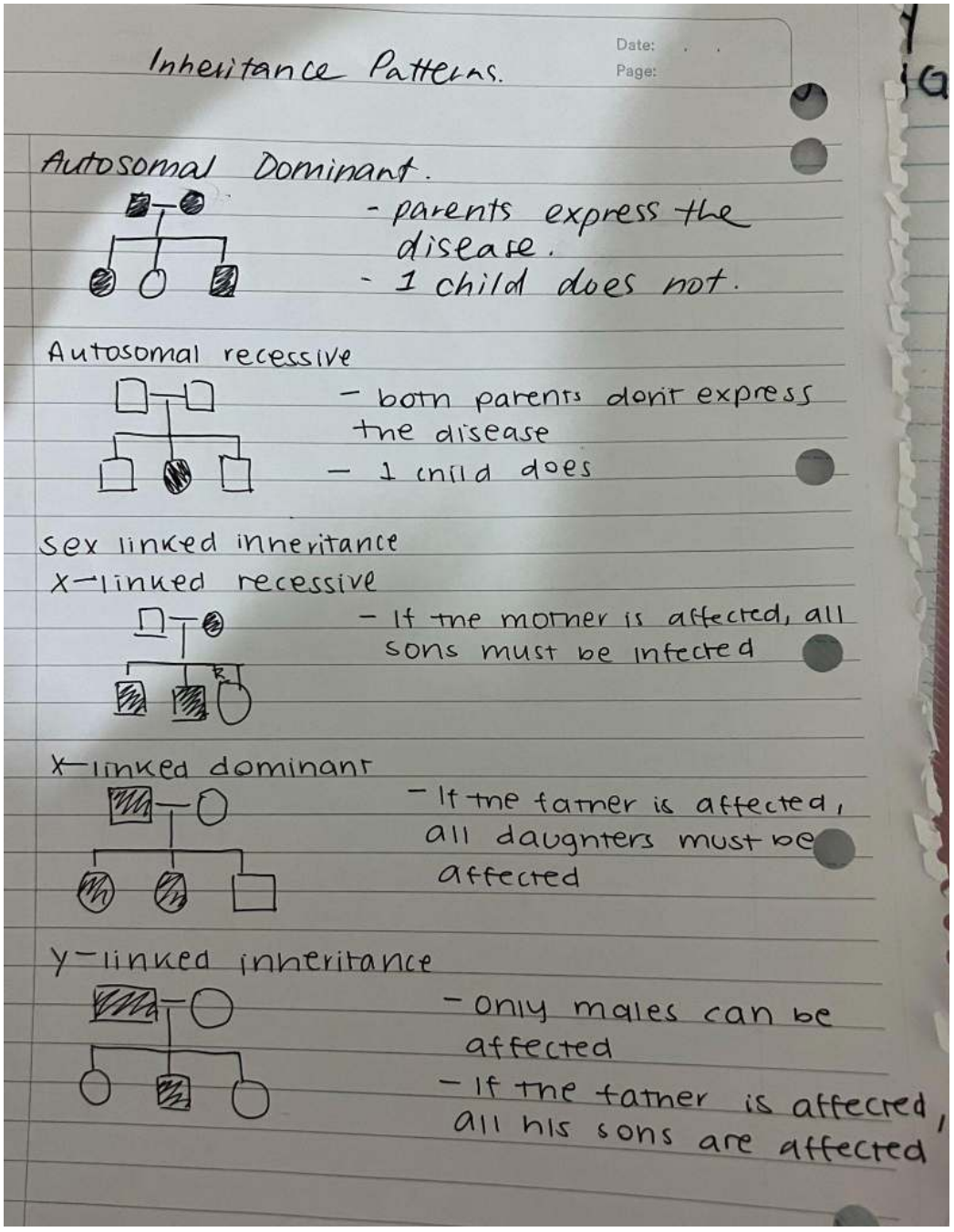
★ Model the formation of new combinations of genotypes produced during meiosis

- Autosomal inheritance

- Autosomal inheritance = traits are passed on non-sex chromosomes (all except X and Y)
- Offspring will inherit one set of chromosomes from each parent equally
- Males and females are equally likely to inherit the gene

Autosomal dominant	• Affected individual carries at least one affected PARENT • Two affected parents can have an unaffected child
Autosomal recessive	• Affected individuals must be HOMOZYGOUS recessive • Unaffected parents (heterozygous; carriers) can produce affected offspring • Can skip a generation
X-linked dominant	• Does not skip generations • Affected males transmit the trait to all their daughters and none

	of their sons ● Unaffected mothers (homozygous recessive) must have unaffected sons
X-linked recessive	● More common in males ● Affected females pass all the traits to their son ● Affected females must have an affected father - may or may not have an affected mother (could be a carrier) ● Affected sons may be produced by unaffected parents (if mum is a carrier)



- Sex linkage

- Traits are passed on the sex chromosomes (X and Y)
- May be X-linked (only the X chromosome) or Y linked (only the Y chromosome)
- Female - homolgous pair of X chromosomes (XX)
- Male - one X chromsome and one Y chromosome (XY)
- Females inherit an X chromosome from each parent
- Males inherit their Y chromosome from father and X chromosome from mother
- Y chromosome is shorter than X chromosome - some characteristics are only coded for by the X chromosome
- Males only have one allele for each sex-linked gene - only have one X chromosome while females have pairs of alleles

- Co-dominance

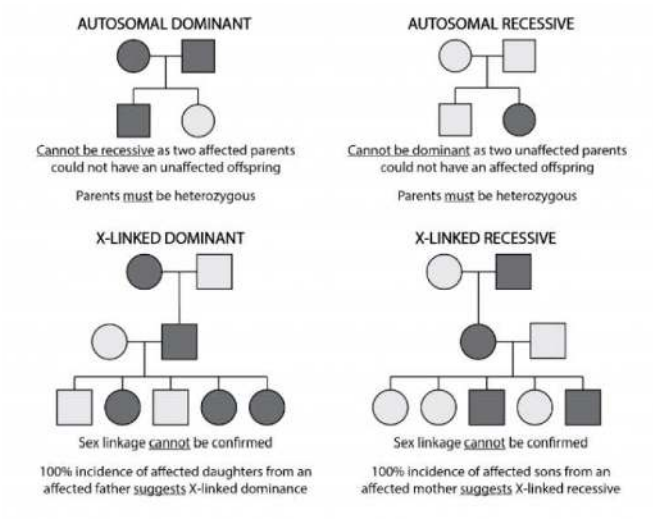
- Both alleles are fully expressed
- Both are dominant
- Written in two different letters, capitalised

- Incomplete dominance

- Blending of alleles - traits are not fully expressed
- Special notation - powers

- Multiple alleles

- Inheritance where three or more alleles exist for a single trait



★ Analysing single nucleotide polymorphism (SNP)

- **SNP** = change in one nucleotide base in the DNA sequence at a specific site - substitution, insertion or deletion
- Some SNP change phenotypes while others don't
- Each combination of SNPS is called a haplotype - set of genetic markers located on a chromosome
- Important genetic markers used to distinguish individuals and identify disease suspectibility in individuals

- Limitations - genetic markers that are closer together give more accurate data - if there is crossing over during meiosis, the SNPS on a chromosome might not all be inherited together

INHERITANCE PATTERNS

IQ5: Can population genetic patterns be predicted with any accuracy?

★ Use of technologies: DNA sequencing and profiling

- DNA sequencing allows to find single nucleotide information for entire human genomes
- DNA profiling allows scientists to determine an individual's unique DNA characteristics

- Polymerase Chain Reaction (PCR)

- Many copies of a DNA can be created in a short amount of time
- DNA primers and other chemicals are added to the DNA sample being studied
- Primers - short lengths of DNA attached to exposed DNA strands at a specific region, from which DNA synthesis can be initiated
- Amplified DNA can then be sequenced or processed further to create a DNA profile

- Gel Electrophoresis

- Technique used to separate DNA fragments according to their size
- DNA samples after sufficient amplification using PCR are loaded into small holes at one end of an agarose gel and an electric current is applied
- Since DNA is negatively charged, the fragments are attached to the positive electorde and are pulled throuhg the gel
- Smaller fragments move through the gel faster than large ones
- When the gel is stained with a DNA-binding dye, DNA fragments can be visualised as bands, each representing a group of same-sized DNA fragments

★ Use of population genetics data in conservative management

- Population genetics = study of genetic variation within a population, including changes in the frequency of genes and alleles within a population

- Inheritance of a disease or disorder

- Conservative management

- Conservation management = how genes are inherited in a population in order to avoid extinction of a species and the maintenance of biodiversity

- Process of determining the exact order of nucleotides of a gene on a chromosome or for the entire organism's genome
- Very expensive, time consuming

Sanger method

1) DNA is extracted from cells

2) DNA is amplified (replicated) via polymerase chain reaction (PCR) (we now have lots of copies of the DNA)
3) The double-strand of DNA is separated into single strands by heating (unwind and unzip)
4) There are four reaction mixtures which include: template DNA, free nucleotides, DNA polymerase, primers, buffers, chain terminating nucleotide
5) DNA polymerase reads template strand to create new complementary strand - this continues until a chain terminating nucleotide is randomly added
6) Process is repeated until every position on the template strand has been identified with chain-terminating nucleotides - now have DNA fragments of varying lengths
7) DNA fragments are separated by gel electrophoresis - the smaller, lighter lengths of DNA migrate further to the bottom, while the longer, heavier lengths of DNA migrate shorter distances - the migration of the DNA fragments creates bands in the gel
8) Sequence read from bottom to top

Gel electrophoresis

- Technique to separate fragments of DNA
- DNA fragments are invisible - they are mixed with fluorescent dye before loading the samples in the wells
- An electric current is applied which causes the DNA to move through the jelly like substance (agarose gel)
- Gel has tiny pores where smaller fragments can fit through - travelling further
- Larger fragments cannot fit through pores - travelling not as far

- Resulting sequence: read straight from diagram, bottom top top
- original/template strand: the complementary bases of the bars

Uses of DNA sequencing

- Genetic testing: to determine if a patient is at risk of a disease that tends to be associated with the prescence of particular gene
- Molecular biological research: study human genome and proteins they encode - important determining locus and distance between genes
- Evolution: DNA tells us where an organism is from - determine inheritance patterns, how organisms are related and how they evolved
- Identification: identify and compare people eg. finding parents, find person at crime scene

DNA profiling process

1) DNA is extracted from cells
2) STR fragments are amplified (replicated) via PCR. Primers are added on both strands of the DNA - tells DNA polymerase where to start and where to stop - 2 alleles for each STR (1 from each parent), both are amplified
3) Fragments separated by gel electrophoresis based on size

- Small STR (less repeats) at bottom
- Longer STR (more repeats) migrate shorter distances
- Patterns of bands (number and location) creates DNA profile

Electropherogram

- DNA profile can be represented by peaks
- STR's appear as single or pairs of peak
- Single peak: same number of repeats on each homologous chromosome

- Pairs: different number of repeats for that STR

Use of DNA profiling

- Determining maternity/paternity of child: child's DNA will consist of combination of parent's DNA
- Identify unknown person: identify suspect of crime or unrecognisable victim