5.1 REPRODUCTION
IQ: How does reproduction ensure the continuity of species?

5.1.1- explain the mechanisms of reproduction that ensure the continuity of a species, by analysing sexual and asexual methods of reproduction in variety of organisms:

- ANIMALS: ADV of external and internal fertilisation

ANIMALS:

- External Fertilisation
- Internal Fertilisation

- Fertilisation is the union of male & female gametes and may take place externally/ internally.
- Many marine organisms- external fertilisation as the water enviro. allows the union of gametes to occur without dehydration. Gametes shed direct into water fertilisation occurs fertilised eggs -->adults.
- During internal fertilisation, male gametes are inserted into the female reproductive tract via a penis/ similar structure. (most terrestrial organisms).

- The key to successful fertilisation of ova by sperm is that the gametes each of which is single haploid cell surrounded by membrane, must not meet and dehydrate in process.

External Fertilisation:

- Needs to occur in the presence of water as gametes need to be hydrated
- Suited to aquatic or moist environments
- Example: Frogs, fish and earthworm
- Many gametes are produced & only small number of them are fertilised
- are usually highly successful. In this environment gametes do not dry out/dehydrate

ADV:

- Less time and energy required by parents
- Fertilised eggs carried away by sea settle in area far from parents- reduces competition for resources

DISADV:

- Lower chance of fertilisation
- organisms must produce large numbers of gametes to compensate for the losses from predation and dispersal
- Zygote vulnerable to predation in external environment.

Example:

- Staghorn Coral

Internal fertilisation

- very successful as the mechanism for direct transfer of gametes avoids dehydration and loss by dispersal, so fewer gametes are required.
- success very high as the environment is enclosed in a confined space and protected from predation.
- Cloaca – one single opening for digestive system, reproductive track and urinary track

ADV:

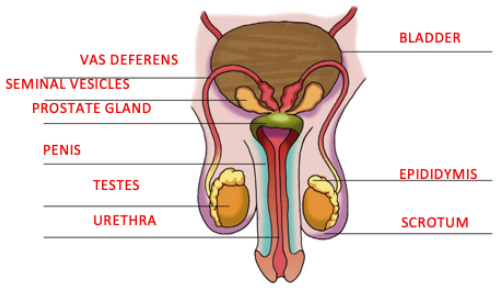
- High chance fertilisation
- Zygote protected by mother body

DISADV:

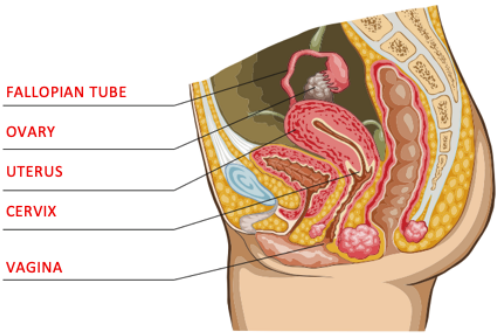
- More time and energy required by parents
- Breeding frequency depends on favourable environment condition

Example:

- humans



Structure	Function
TESTES	Male gonads, responsible for producing male gametes & secreting hormone testosterone.
PENIS	Organ used in intercourse to ejaculate semen containing sperm into female reproductive tract.
EPIDIDYMIS	Coiled tubule where sperm are temporarily stored while they mature.
SCROTUM	A double walled pouch that hangs outside the body to regulate the temperature of the testes for optimal sperm production.
PROSTRATE GLANDS	These glands secrete fluid to nourish the sperm and allow the sperm to swim.
VANS DEFERENS	Also known as the sperm duct, this tubule carries sperm from the epididymis to the urethra
URETHRA	Tube that empties urine from the bladder
BLADDER	Stores urine



Structure	Function
CERVIX	Lower part of the uterus that opens into the vagina Contains a narrow channel that allows sperm to enter the uterus and blood to leave during menstruation.
OVARY	A small oval-shaped glandular organ that contains cells that divide to make female gametes / egg cells (called ova – singular ovum). Female reproductive hormones are made here. Ovulation occurs here - the release of a mature ovum (egg cell) occurs by either of these structures once every 28 days.
UTERUS	A muscular organ where the fertilised egg implants into the wall and the developing foetus is housed. Growth of the baby occurs here during pregnancy (if fertilisation occurs).
FALLOPIAN TUBE	A tube that transports the ovum to the site of fertilisation. Fertilisation (also known as conception) occurs here.

Characteristics	Differences	
	External Fertilisation	Internal Fertilisation
Gametes	Large number of male and female gametes produced	Large number of male gametes and fewer female gametes produced
Union	Occurs in open water environments	Occurs inside the reproductive tract of the female in organisms that live mostly or completely on land
Conception mechanism	Simultaneous release of gametes	Copulation: the male inserts sperm into the female's reproductive tract via the penis or cloaca
Chance of fertilisation	Low because male gametes are released into large open area where there is less chance of successfully uniting with female gametes	High, because male gametes released into confined space where there is more chance of successfully uniting with female gametes
Number of offspring/zygotes	Usually a larger number than internal fertilisation but many zygotes perish so a smaller number of offspring survive	A smaller number of offspring than in external fertilisation because very few perish (higher success rate)
Parental investment	Usually no parental care	Parental care of eggs and/or developing young is more common

PLANTS: asexual & sexual reproduction

PLANTS:
Asexual Reproduction:
- Vegetative Propagation
- Apoximis

Sexual Reproduction

FUNGI:
Asexual Reproduction:
- Budding
- Spores
- Fragmentation

Sexual Reproduction:

BACTERIA:
- Binary Fission

PROTISTS:
Asexual Reproduction:
- Binary Fission
- Budding

Sexual Reproduction:

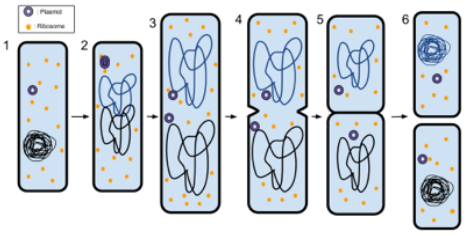
- Reproduction allows the survival of a species from one generation to the next.
- There are two types of reproduction – asexual and sexual.
- The simplest way that organisms can reproduce is asexually. Asexual reproduction is the production of identical offspring from just one parent.
- Asexual reproduction produces new individuals by mitosis, a process of nuclear division in which each daughter cell receives an identical copy of every DNA of the parent cell.
- The offspring are therefore clones individuals that are genetically identical, unless genetic mutation occurs.

Asexual reproduction:
- Occurs in unicellular organisms, fungi, plants and animals
- Results in large numbers of new individuals being produced relatively quick
- Is an advantage in an unchanging environment when individuals are adapted to
- their environment
- Results in a lack of variation in a population (individuals are genetically identical to
- parents). If conditions become unfavourable, then all individuals are vulnerable and
- could die, leading to the extinction of the population.

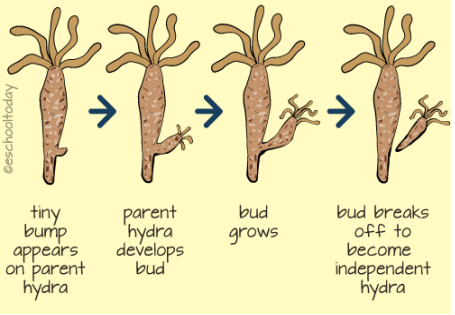
Types of asexual reproduction

- FUNGI- budding, spores
- BACTERIA- binary fission
- PROTISTS- binary fission, budding

Binary Fission:
- Equal division of parent cell into two new cells
- Examples: Bacteria, Protozoan



Budding:
- Outgrowth, asymmetrical cell division
- Small bud grows out of parent cell
- Bud breaks off and grows
- Parent is larger than the offspring
- Examples: Yeast



Fragmentation:

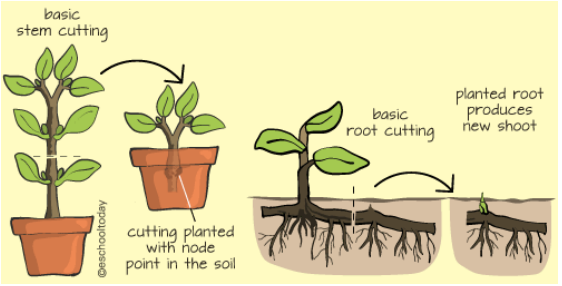
- Part of organism breaks off and regenerate into a new individual
- Examples: Animals including marine worms and echinoderms

Spore Formation:

- Spores released into environment and germinate into new individuals
- E.g. fungi, plants, including mosses and ferns

Vegetative Propagation:

- Plant separates to form new independent plants from leaves stems and underground stems
- Many plants including flowering plants
- potatoes
-

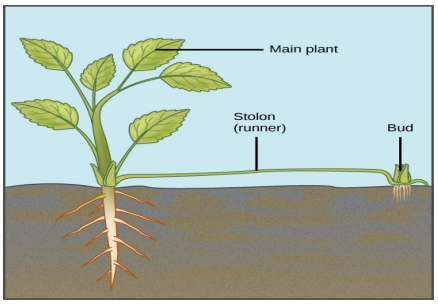


Pathogenesis:

- A type of cloning resulting from the formation of a new individual from an unfertilised egg; all offspring are clones of the female parent (i.e. no males are produced)
- Animals, including insects (e.g. wasps, ants), lizards and birds

Budding from Runner:

- Runner from main plant to new bud
- Occurs above the group
- Bud forms when runner touches the ground

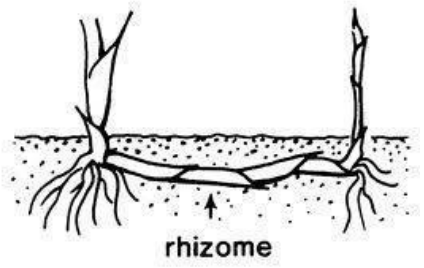


Taking a cutting and planting it

- Section of plant is cut and planted in different location
- Roses

Rhizomes:

- Long, modified stems that grow underneath the soil. New plants start growing from the nodes at the stem
- Ginger, lawn, grass, ferns and irises



Sexual Reproduction

- mixing of genetic information from two parents.
- Usually involves the union of male and female gametes (sperm and egg) to form a unique individual.
- Most multicellular organisms. Formed by the process of cell division called meiosis.

- Feature specialized structures in which haploid gametes are produced.
- Fusion of haploid gametes during fertilisation produces a diploid zygote

Haploid – having one set of chromosomes (*n*) found in gametes
Diploid – having two sets of chromosomes (2*n*) usually found in somatic cells (body or non-reproductive cells)

- *Zygote* – a fertilised egg. Offspring inherit one set of chromosomes from the mother (maternal chromosomes) and one set from the father (paternal chromosomes).
- A zygote arises as a result of the fusion of haploid gametes, when the chromosome number changes from haploid to diploid.

Hermaphrodites

- Having both male and female sex organs
- Advantage when there is a low population density or animals are non-motile such as coral
- Disadvantage – Gametes have fewer genetic combinations resulting in offspring with low genetic variation
- Example: Snails

	Method	Advantages	Disadvantages
Sexual	1. Haploid gametes produced by parent organisms 2. Sperm fertilises the ovum 3. Fusion of gametes produces zygote, containing genetic material from both parents	- Combination of different genes/traits - Variation - Ability to adapt to environment	- Time intensive - Energy intensive - Mating partner - Fewer offspring produced
Asexual	Offspring arise from a single organism, and contain the genetic material of only that organism. E.g. Fission, budding, sporogenesis, vegetative propagation	- Quick - Not energy intensive - No requirement for mates - Exponential colony growth	- Clones of parent = lack of diversity - Reduced adaptive ability – reliant solely upon mutation

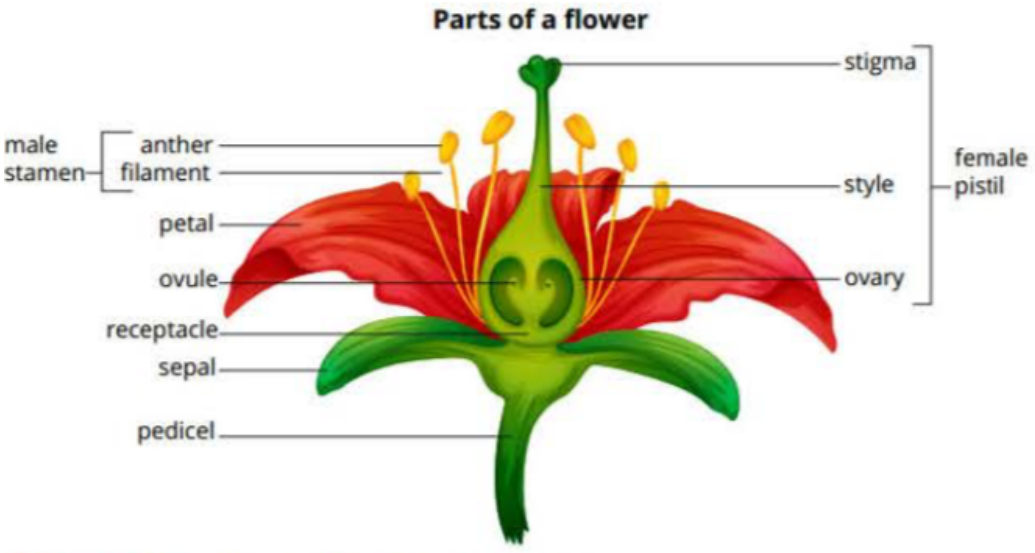
Sexual reproduction in plants

- As plants are sedentary (still), they use a range of strategies to ensure gametes come together.
- In flowering plants, this typically relies on insects such as bees in search of nectar; at the same time, they transfer pollen (containing the male sex cell) from the anther of one plant to the stigma of another of the same species.

Angiosperms: Flowering Plants

- Male sex organs – STAMEN
  Anther – where pollen grains are formed
  Filament – stalk that carries the anther. The length determines whether the anthers are contained inside the petals for insect pollination or hanging outside for wind pollination
- Female sex organs – CARPEL
  Stigma – sticky top surface of the flower to which pollen adheres. May be relatively small and smooth (insect pollinated plants) or large and feathered (wind pollinated plants)
  Style – joins the stigma to the ovary
  Ovary – where ovules are formed

Gymnosperms have 'naked seeds' that are not in ovaries, exposed on modified leaves example in conifers the leaves form cones.



Self-Pollination

- Pollen attaches to the stigma of the same flower
- Ensures the survival if reproductive partners are scarce

Cross – Pollination

Pollen attaches to the stigma of a different flower
Increases genetic variation and ensures survival if sudden change occurs in the environment

SEED DISPERSAL:

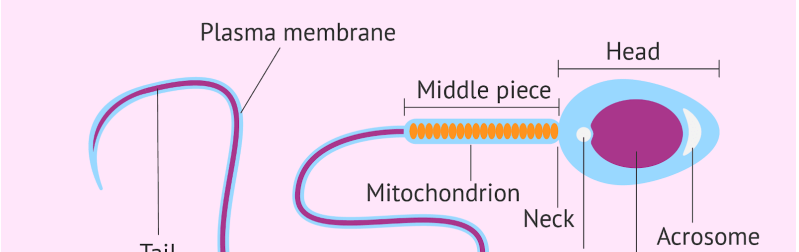
- Wind, animal, self

5.1.2 Analyse the features of fertilisations, implantations and hormone control of pregnancy and birth in mammals

- Mammals reproductive mechanisms to maximise reproductive success:
  - Internal fertilisation
  - Implantation
  - Pregnancy
- Hormones coordinate the reproductive cycle
  - Ensures greater reproductive success

Fertilisation :

- fusion of male and female gametes to form a zygote (fertilised egg)
- Occurs in fallopian tube
- Equal contribution from male and female parents
- Occurs in 4 steps:
  1. Sperm uses enzymes from acrosome to dissolve and penetrate protective layer (zona pellucida) surrounding the egg to reach the cell membrane
  2. Molecules on sperm surface bind to protein receptors on the egg's cell membrane and the nucleus of the sperm enters the cytoplasm in the egg cell
  3. Change at the surface of the egg occur to prevent the entry of other sperm cells
  4. Fusion of the haploid egg and sperm nuclei results in the diploid zygote cell – the fertilised egg

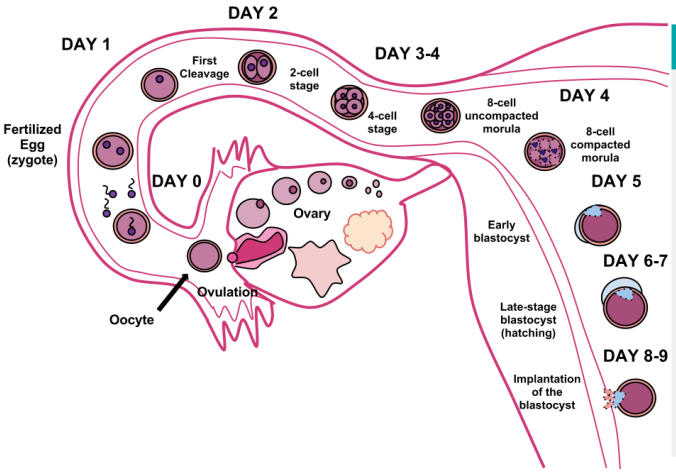


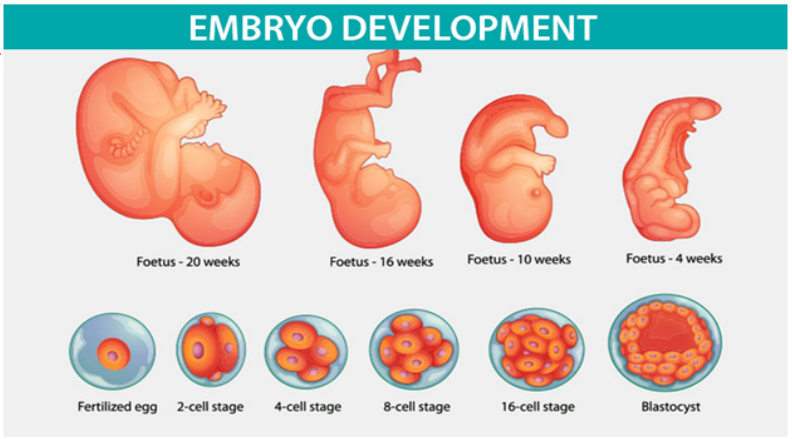
Implantation:

After fertilisation zygote travels down oviduct and is at the **cleavage stage**

- Zygote undergoes mitosis
- When it reaches the uterus, it is a blastocyst stage
  - More cell division
  - Cell differentiation
- Day 8-9 – attaches to the uterine wall
  - Outer layer attached to endometrium through finger like projections
  - Area develops into placenta

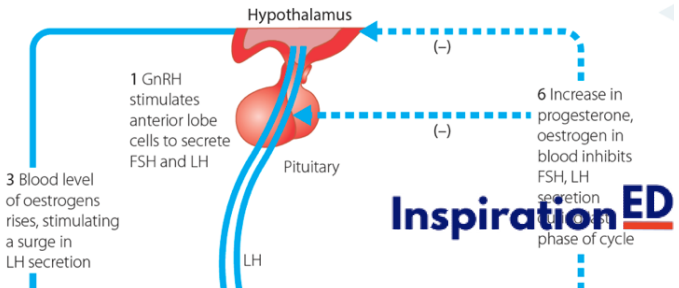
- Embryo Development
- Blastocyst becomes gastrula
- 3 different layers of cells
- Eventually becomes an embryo
- 3-8 weeks
- Becomes foetus
- 9-40 weeks
- Forms basic adult features





Hormones:

- Chemical messengers that can travel in the bloodstream
- Often made of proteins or lipids
- Pituitary gland:



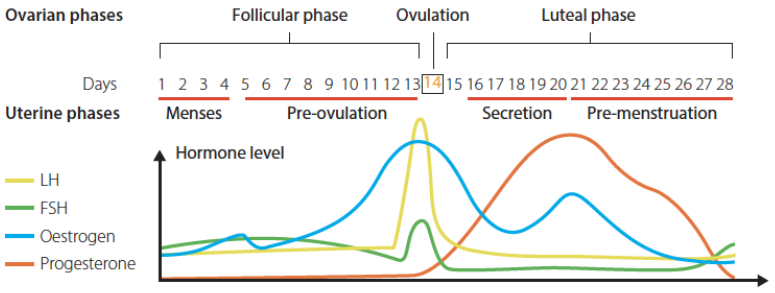
    - Secretes hormones that stimulate other endocrine glands
    - Regulates the release of their hormones for growth, metabolism and reproduction
    - located at the base of the brain, above the roof of the mouth.
- Sex hormones:
    - Specifically affect the growth or functioning of the reproductive organ or development of secondary sex characteristics
    - Produced in specialised ovaries tissue, pituitary gland and adrenal cortex
- Vital role in development and functioning of the male and female reproductive systems

The Ovarian Cycle:

- Females born with all the eggs (ova) they will produce
- Ova are immature and partly developed in the ovary
- Puberty start of ovarian and menstrual cycle
- Ova in ovaries begin to divide as enveloped by single layer of cells forms follicles
- Hormones trigger development and maturation of ova each month until menopause
- Repeats approximately every 28 days
- Start of cycle:
    - few follicles begin to develop
    - one enlarges to reach maturity

The Follicular phase:

- egg is pushed to one side of follicle as cell produces fluid
- dominant follicle moves to surface of ovary and creates a budge
    - mature Graafian follicle
    - development to maturity takes 10-14 days
- Follicle releases oestrogen- causes surge in LH production- results in ovulation
- Oocyte is released from mature follicle – ovulation
- Ovulation occurs in the middle of a cycle in the ovary – day 14



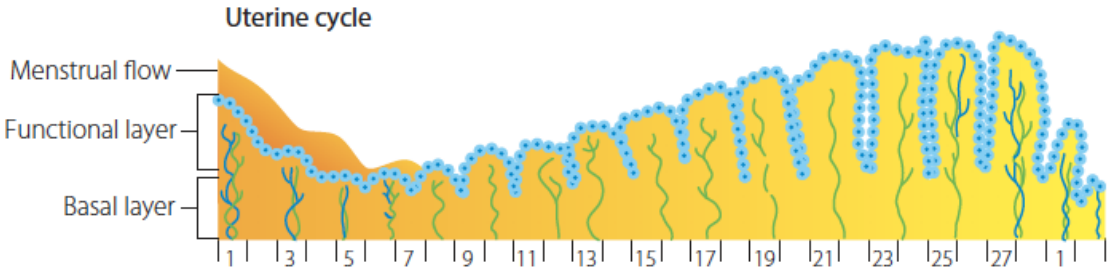
The Luteal Stage:

- Begins after ovulation
- The burst follicle is the corpus luteum
- Secretes progesterone, preparing uterus for pregnancy

If not Pregnant (egg not fertilised within given time): The Menstrual Cycle:

- Repeats every 28 days
- Starts with period
- Lining of uterus is shedding
- Accompanied by bleeding
- Days 5-12 new endometrium forms
- Day 14 ovulation occurs

- After ovulation:
- Corpus luteum secretes progesterone as well as oestrogen
- Progesterone peaks 8-10 days after ovulation around expected implantation time



Hormones produced during menstrual cycle:

Hormones	Produced by	Target organ	Functions
Follicle stimulating hormone (FSH)	Pituitary Gland	Ovaries	Promotes development of follicles and secretion of oestrogen
Luteinising Hormone (LH)	Pituitary gland	Ovaries	Promotes ovulation, development of corpus luteum and secretion of progesterone
Oestrogen	Ovaries	Reproductive Tract	Thickens endometrium and promotes development of secondary sexual female characteristics
Progesterone	Ovaries	Uterus	Prepares uterus for and maintains endometrium during pregnancy
Human Chorionic Gonadotropin (hCG)	Placenta	Ovaries	Maintains corpus luteum for production of progesterone and stops ovulation

Male Hormones

- testosterone
  - produced in testes
  - develops secondary sexual characteristics
  - spermatogenesis – production of sperm

Hormones during pregnancy

Hormone	Source	Effect on body
Human chorionic gonadotropin (hCG)	Embryo	• Maintains corpus luteum maintain the endometrium • Pregnancy tests measure the presence of this hormone
Oestrogen	Ovaries and the placenta	• Promotes the growth of endometrium
Progesterone	Corpus luteum and placenta	• Stimulates secretion of mucus by cells lining the uterus • Growth of blood vessels • Suppresses uterine activity-supporting foetal development and risk of foetus being disturbed or expelled by the uterine contractions

Hormones at Birth:

For a baby to be born:

1. Muscles in the uterus must contract to expel the baby
2. Tissue of cervix must soften so that the cervix can dilate (widen) to allow the passage of the baby

- Hormones involved:
- Prostaglandins
- secreted by the walls of the uterus initiates labour
- makes uterus tissue more sensitive for other hormones to act on

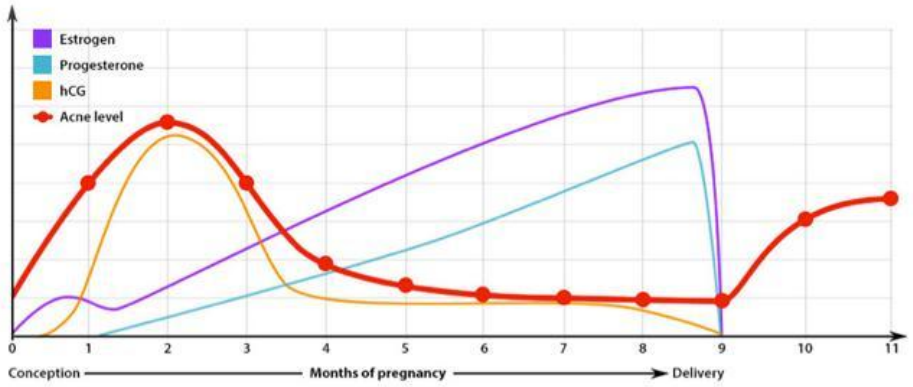
- Oxytocin
- Promotes coordinated contractions of the smooth muscle of the uterus
- Softening of the cervix

- More contraction results in high levels of oxytocin release – positive feedback

- Progesterone and oestrogen levels will decline during labour and contractions become stronger
- Prolactin
- Mainly produced it the pituitary gland (sometimes uterus and breast)
- Stimulated milk production in the breast
- Prolactin and oxytocin release are further stimulated by suckling of the baby
- Important role in maternal behaviour and bonding

Hormonal Control

- Prevents pregnancy- varying the level of sex hormones
- Oral contraceptive pills
- Prevent release of an ovum – inhibits ovulation so there is no egg to be fertilised
- Thickens cervical mucus- inhibit sperm mobility
- Changing lining of the uterus- implantation is difficult



5.1.3 Evaluate the impact of scientific knowledge on the manipulation of plant & animal reproductions in agriculture

- Increase and proliferation of scientific knowledge huge advancements in agriculture
- Enables processes to become more efficient and productive
- Understanding fundamental principles of reproduction
  Able to manipulate desired outcomes

Selective Breeding

- Based on understanding that phenotypic traits are hereditary
- Farmers selectively mate plants with desirable traits
- May influence cross-breeding or pure-breeding practices
- Current breeds used in agriculture produced through selective breeding – Angus cow

Artificial insemination

- Method
  1. Detection of female cows in oestrus (animals that are sexually receptive)
  2. Collection of semen
  3. Insemination usually performed using an insemination gun which shoots semen into the cervix of desired animals

ADVANTAGES:	DISADVANTAGES:
o Reduces danger to animals of injury during transit o Many females can be inseminated	o Reduction in genetic diversity

o Can increase number of endangered species o Can increase reproduction of population with desires attributes o Timing – able to synchronise births	o Costly – requires specialised equipment o Time consuming o Ethical issues

Artificial pollination
- o Method
  1. Pollen (sperm) removed from stamen of one plant
  2. Pollen applied to the stigma of another plant
  3. Pollen fertilises ovum

ADVANTAGES:	DISADVANTAGES:
o Produces hybrid plants o Increase genetic variability within population o Drone pollinators can also work in bee niche o Cross-breeding of favourable traits o Ensuring successful pollination of all plants results in higher crop yield	o Reduced biodiversity

Genetic Engineering
- o Knowledge of DNA structure and improvement of genetic techniques
  - Gene cloning and transgenic
  - Enabled agriculturalist to manipulate organisms at fundamental level
- o Allowed introduction of new desired traits in organisms:
  - Dt cotton – insect resistance
  - Golden rice – increased nutritional value
  - Strawberries – frost resistance

5.2 CELL REPLICATION
IQ: How important is it for genetic material to be replicated exactly?

5.2.1 Model the processes involved in cell replication:
- o Mitosis and meiosis

STRENGTHS	LIMITATIONS
- Convenient and cheap - Modelled	- Didn't thoroughly show all the steps

MITOSIS: Cell division in eukaryotic for growth & repair

2 MAIN STEPS:
- MITOSIS division of nucleus
- CYTOKINESIS division of cytoplasm

The Cell Cycle:
- o Cell division & enlargement occur in a repetitive cycle
- o 1 complete cycle takes 18- 22 hours
- o Mitosis is only 1 part of cell cycle & takes 1- 2 hours

Before cell divides, undergoes preparation for division INTERPHASE. This takes longer & eukaryotes spend 90% of lifetime in this phase.

INTERPHASE:
- o G_1- Gap phase during which cell enlargement takes place before DNA replicates.

- S- Synthesis period where DNA replication occurs
- G_2- Second gap phase after replication, when cell prepares for division.

DNA replicates (identical copy), not yet recognisable as individual chromosomes.

PROPHASE:

- Chromosome become visible
- Each chromosome has 2 copies DNA
- Chromosomes split longitudinally in 2 arms (chromatids), held together by a centromere.
- Each chromatid has 1 copy DNA
- Nuclear membrane breaks down and no longer visible
- SPINDLE FIBRES begin to form

METAPHASE:

- Chromosomes line up along centre of cell, each attached to spindle fibres by a centromere

ANAPHASE:

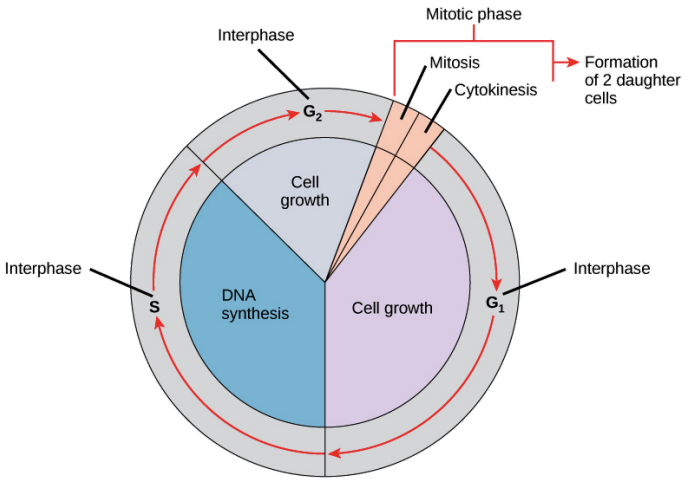
- Spindle fibres contract and as new daughter chromatids begin to separate, now termed daughter chromosomes
- Daughter chromosomes pulled towards opposite poles of cells

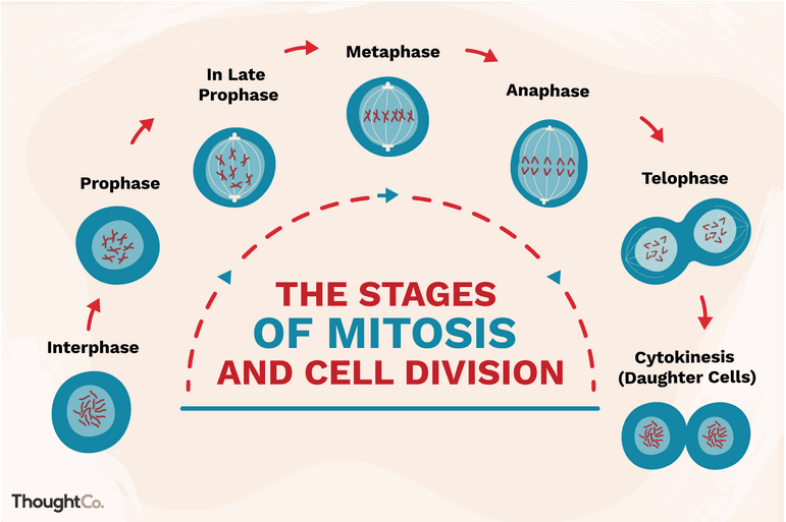
TELOPHASE

- Daughter chromosomes gather at opposite poles
- Nuclear membrane reappears
- MITOSIS now complete 2 nuclei with identical chromosomes to each other & to original nucleus in parent cell.

CYTOKINESIS:

- Division of the cytoplasm, separating 2 daughter nuclei so each in own cell, each cell only has 1 nucleus
- ANIMAL CELL- Cytoplasm constricts (pinch) in centre cell, between 2 daughter nucleus and pinches off
- PLANT CELL- cell plates form while nucleus still in telophase- cellulose is then deposited either sides, forming a cell wall to separate 2. Cellulose is then deposited either sides, forming a cell wall to separate the 2.





MEISOSIS: cell division in eukaryotic that give rise to haploid gametes

During meiosis, genetic variation arises as a result of the behaviour of chromosomes at 2 stages:

- CROSSING OVER
- When chromosomes RANDOMLY SEGREGATE & paternal and maternal ASSORT INDEPENDANTLY of each other.

During Meiosis 1:
PROPHASE 1:

1. Chromosomes line up in homologous pairs (1mat + 1 pat chromosomes in each)

2. DURING CROSSING OVER – introduces genetic variation } the exchange of genetic material between homologous chromosomes causes the mix of genes = increased number of combos in genes offspring

ANAPHASE 1:

3. The chromosomes in each pair separate, so that 1 entire chromosome moves into a daughter cell. Separation- RANDOM SEGREGATION, ensures the chromosomes number in resulting gametes will be half original cell.

o The manner in which these chromosomes separate (independent assortment) – the pat and mat sort independently
o = Future genetic variation.

DURING MEIOSIS 2: (no further variation, just replication to form 4 haploid gametes)

- the 2 daughter cells that result from meiosis 1 undergo meiosis 2
- the behaviour of chromosomes in the second meiotic division does not further affect genetic variation.

o DNA replication using the Watson and Crick DNA model, including nucleotide composition, pairing and bonding.

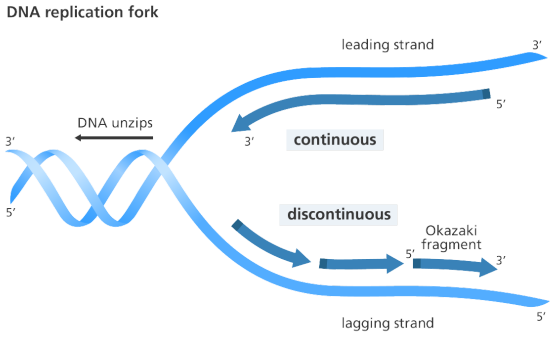
- The used model of DNA is called the Watson and Crick model.
- It is a double helical nucleic acid molecule, which carries genetic information, encoded as sequences of nucleotide baes.
- A single nucleotide is a phosphate, bound to deoxyribose sugar group and a nitrogenous base (adenine (2), thymine (2), guanine (3) or cytosine (3))
- DNA replication is the process by which a DNA molecule makes a copy of itself during cell division.
- DNA is described as being semi-conservative because it is made of one old and one new strand.

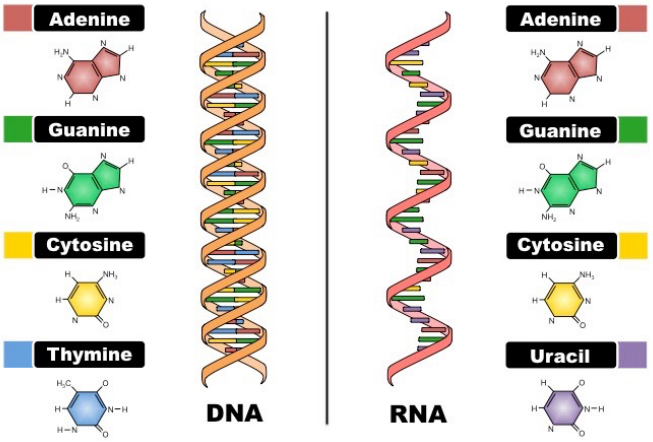
<u>the process</u>

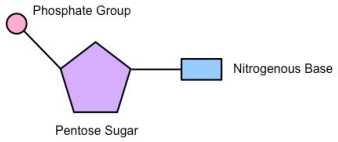
1. The DNA molecule has to 'unzip' its double helix
2. The unzipping is carried out by an enzyme called helicase which breaks the hydrogen bonds that hold the bases together
3. The 'Y' shape that is created by this is called the replication fork, and act as the templates for the new DNA strands
4. One of the strands is in the 3' to 5' direction (towards the fork), which is called the leading strand; the other is in the 5' to 3' direction and is called the lagging strand. They have different replication processes because of their direction
5. Once all of the bases are matched up, after the individual processes, and enzyme called exonuclease strips the primers. The gaps left are filled with more nucleotides
6. The new strands are proofread to make sure there are no mistakes
7. DNA ligase (enzyme) seals the two strands of DNA into double strands

8. The final result of DNA replication is two DNA molecules, made up of one old and one new strand. The DNA automatically winds itself up.

- LEADING STRAND
  - A short piece of RNA, known as a primer (produced by primase), binds to the end of the strand. This acts as a starting point for the replication
  - DNA polymerase binds to the leading strand and moves along it, adding complimentary nucleotide bases in the 5' to 3' direction
  - THIS PROCESS IS CONTINUOUS
- LAGGING STRAND
  - Numerous RNA primers, made by the primase enzyme, bind at various points along the lagging strand
  - Chunks of DNA are added to the strand, which are known as Okazaki fragments, in the 5' t o 3'
  - THIS PROCESS IS DISCONTINUOUS BECAUSE OF THE DIRECTION
  - The Okazaki fragments will be joined up later







5.2.2 assess the effect of the cell replication processes on the continuity of species

DNA REPLICATION:
- A fundamental heredity unit, directs all processes in a cell
- Reproduction of cell dependent on this, as the creation of new cells require more DNA produced
- By copying gen mat, replication ensures that info important for life is transferred
- If DNA not replicated before mitosis & meiosis, cell division would only have half amount of DNA and resulting cells could die due to inadequate
- Is a high fidelity (accurate) process, ensuring that DNA strands carry same gene and encoding all essential proteins in life.

MITOSIS:
- Essential for development and growth. It increases number cells in organism, allowing for development of a multicellular body
- Replaces old, ensures tissue continue to function effectively & efficiently
- Humans, mitosis allows us to develop to maturity when can pass gen mat offspring
- Some reproduce by asexual reproduction, which is facilitated by mitosis

MEIOSIS:
- Gametes are end products- haploid cells
- Combo gametes during sexual reproduction creates new organism
- Introduces variation

- Genetic diversity ensure continuity of species

5.3 DNA & POLYPETIDE SYNTHESIS
IQ: Why is Polypeptide Synthesis important?

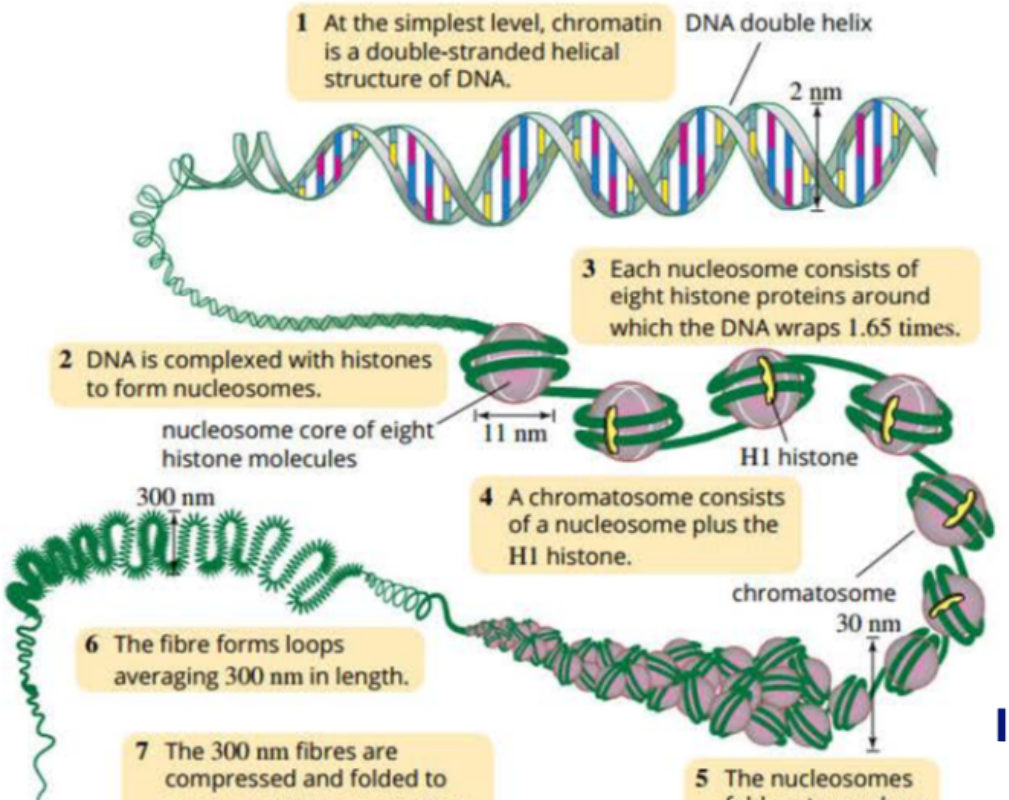
5.3.1 Construct appropriates representations to model & compare the forms in which DNA exists in Eukaryotes & Prokaryotes:

EUKARYOTES:	PROKARYOTES:
o Chromosomal DNA is in the nucleus, which is separated from the cytoplasm by a double layered membrane o DNA is tightly packed- coiled around histones forming nucleosomes, which are condensed into chromatin and packed as chromosomes in nucleus o Coiled in chromosomes o Larger GENOMES o Long non- coding repetitive sequences o Linear DNA	o DNA just chillin in cytoplasm o Contain plasmids- small circular DNA o Chromosomal DNA is in a region of cytoplasm called the nucleoid, lacking a membrane o Not packaged by proteins o Smaller, more compact genome o Extra- chromosomal DNA plasmids

Gene Expression in Prokaryotes & Eukaryotes:

- Gene expression only consists of polypeptide synthesis and occurs completely within the cytoplasm. But as DNA is located in the nucleus of eukaryotes polypeptide synthesis in eukaryotes are much more complex

Packaging of DNA in Eukaryotes:

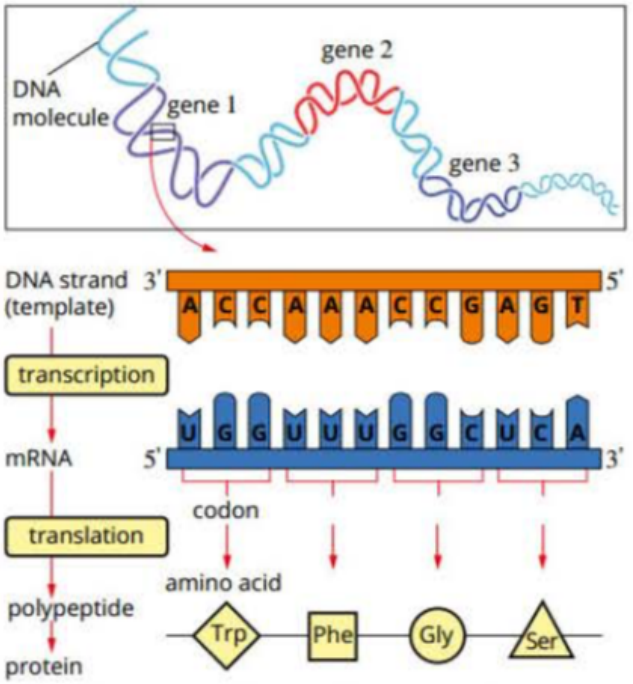


5.3.2 Model the process of polypeptide synthesis

- Transcription & Translation
- Assessing the importance of mRNA & tRNA in transcription & translation
- Analysing the function & importance of polypeptide synthesis
- Analysing how genes & environment affect phenotypic expression

Polypeptide Synthesis:

- A Polypeptide is a chain of amino acids that are held together by peptide bonds - proteins are made up of several of these.
- The genetic code represents the inherited genetic information stored in DNA as triplet bases within sections called genes, This info is transcribed to RNA, then used to synthesise the amino acid sequence that from polypeptides thru process called gene expression.
- Chains of polypeptide combine to from proteins. They are biological molecules that carry out most of the functions
- There are two main steps of protein synthesis;
  - Transcription
    - DNA is copied to make mRNA
  - Translation
    - Message taken to the ribosomes, and proteins are made with tRNA
- The ribosomes hold everything in place and form the bonds between amino acids. They are made up of RNA



Role of DNA in Polypeptide Synthesis:

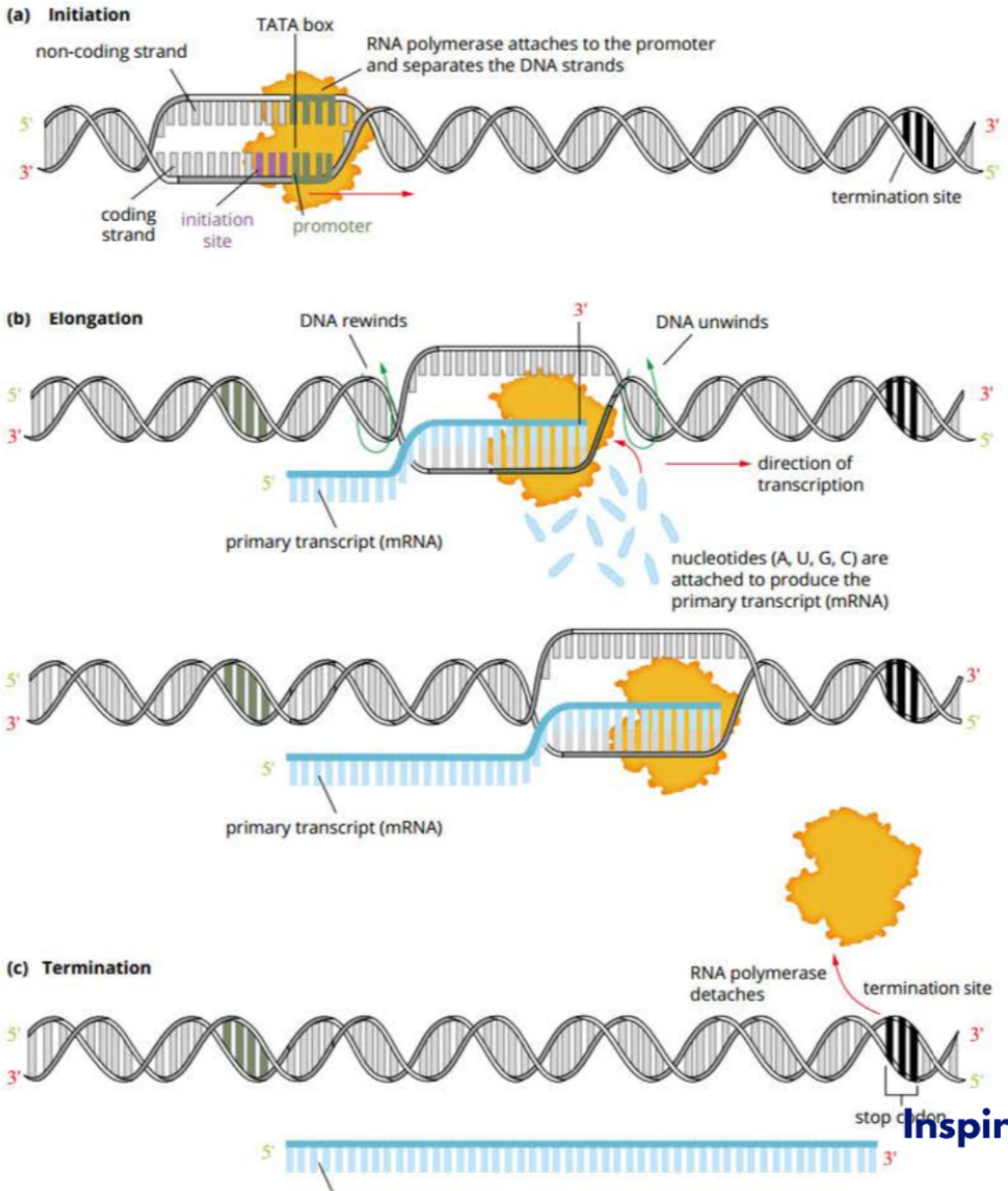
- Provides instruction, which are translated by RNA into polypeptides and proteins.

- Stores and transmits hereditary info as a sequence of nucleotides
- Group of 3 nucleotides are called triplets
- When a DNA triplet is transcribed into mRNA, the triplet is then called a codon

Transcription

- The process of turning genetic information stored in the DNA into a intermediary molecule mRNA.
    1. RNA polymerase binds to a 'promoter', which signals the DNA to unwind, and allows the enzyme to read the bases.
    2. The mRNA molecule is built, using complimentary bases.
    3. mRNA detaches from the DNA strand.

- mRNA is used to transport information out of the nucleus for processing.
- It is a 'messenger' ribonucleic acid.
- Once the mRNA molecule is made, there are modifications that can occur - these modifications are called splicing - and can increases variability.
- In eukaryotes the mRNA molecule will travel out of the nucleus so it can be translated in the endoplasmic reticulum, whereas in prokaryotes, all aspects of this process occur in the cytosol.



Translation

- The process of turning information encoded as mRNA into a polypeptide chain.
    1. mRNA attaches to a ribosome and act as docking stations for tRNAs to deposit their specific amino acids.
    2. The ribosome matches the codon and anti-codons (in tRNA) together.
    3. A polypeptide bond is formed, and continues to grow as amino acids are added
    4. Once a stop sequence is reached, the chain detaches and then folds.
    5. You now have a protein.
- The genetic sequence of the mRNA is read by the ribosomes, and then translated into groups of three nucleotides (codons).
- tRNA molecules transfer amino acids from the cytoplasm to the ribosomes, where they are joined to from a polypeptide chain based on the sequence of nucleotides in the mRNA.

Function & Importance

- Polypeptide synthesis is the method used to produce proteins in cells, which are the building blocks.
- Polypeptide synthesis is important for the increasing complexity of organisms
- the more proteins, the more complex the organism.
- At each stage of polypeptide synthesis there are opportunities for variation, which is crucial for the continual development and adaptation of species.

Phenotypic expression

- Genotype is the genome/ genetic makeup of an organism
- Phenotype is the outward appearance of an organism, such observable traits, biochemistry and physiology
- Whilst an organism's genetic make-up will remain static throughout its life, its observable traits may change over time as a result of their environment.
- By understanding our genotypes we are able to predict our phenotypes to a certain extent.
- External factors, such as the environment, have a say in how our genes are exposed.
- Certain genes can be 'switched on' at different stages of development or are only expressed in response to certain events (extreme temps etc).
- We can consider genotype as containing a range of phenotypic possibilities due to different environmental influences.

5.3.3 Investigate the structure & function of proteins in living things

- Proteins are all composed of the same fundamental building blocks - amino acids.
- Amino acids are organic compounds which have a central carbon, bound to an amine group, a carboxyl group, a hydrogen and a R-group.
- The way amino acids are differentiated is by the R-group, which varies in each type, and gives the molecule different properties.
- Amino acids become a part of polypeptide chains through the formation of polypeptide bonds, which occurs during polypeptide synthesis in the ribosome.
- Polypeptide chains fold to become proteins.
    - This folding process is influenced by interactions between amino acids side groups.

PRIMARY STRUCTURE

- Sequence of amino acids
- Arrangement of amino acids sequences in a polypeptide chain

SECONDARY STRUCTURE

- The formation of alpha-helices and beta sheets (one or the other)

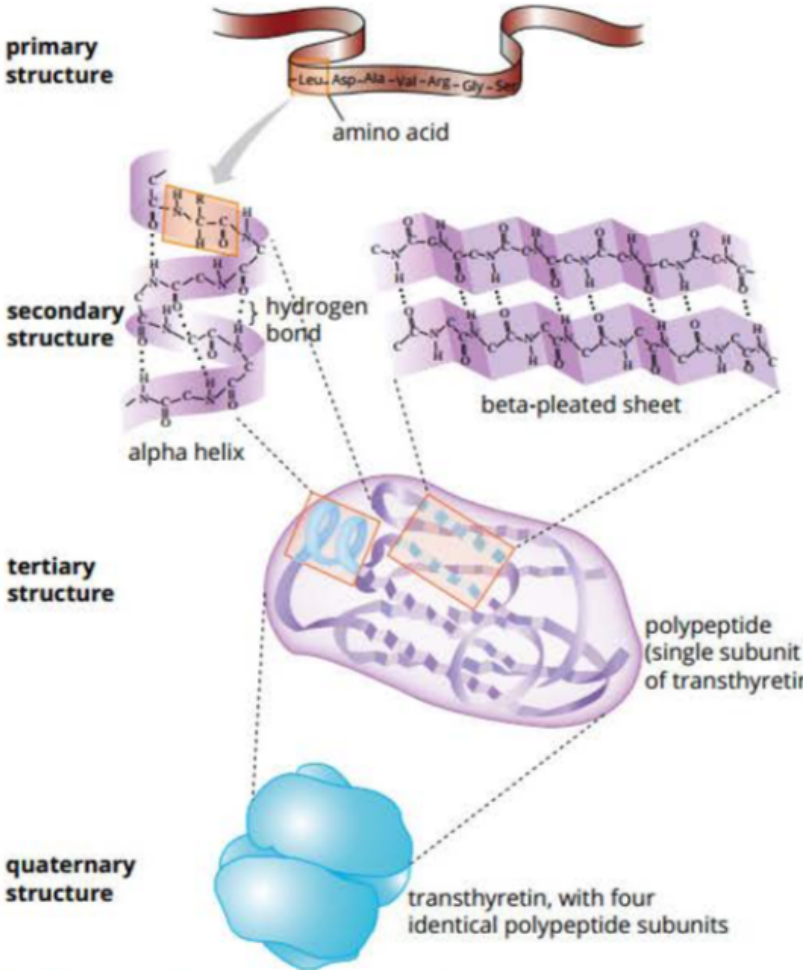
- Strong/ helix shape
- Weak/ layered

TERTIARY STRUCTURE

- Formation of 3D shape

QUATERNARY STRUCTURE

- Interaction of protein subunits



Function:

STRUCTURE AND SUPPORT e.g. collage, keratin	Proteins form the basis of the cellular cytoskeleton, and composes important macro-molecular structures (hair, nails etc)
TRANSPORT AND STORAGE	Proteins in the cellular membrane are responsible for trafficking molecules in and out of the cell. Storage proteins reserve important biological materials for use in the body.
ENZYMES	Proteins may function as biological catalysts, carrying out thousands of chemical reactions inside the cells.
ANTIBODIES	Proteins form an important part of the immune response by recognising and binding to foreign particles.
MESSENGERS	Hormones are proteins which transmit signals around the body, allowing the complex array of biological processes which occur to be coordinated effectively.

5.4 GENETIC VARIATION

IQ: How can the genetic similarities & difference within & between species be compared

5.4.1 Conduct practical investigations to predict variation in the genotype of offspring by modelling meiosis, including crossing over of homologous chromosomes, fertilisation and mutation

- Variation is first introduced during meiosis, specifically the process of crossing over.
- That process involves homologous chromosomes crossing over creates new combinations of chromatids.
- Gamete formation sorts chromosomes independently of one another, meaning that a number of different chromosome combinations nay be formed.
- During fertilisation there is further opportunity for variation to be introduced, as alleles from P1 are combined with P2, which then creates a full set of chromosomes.
- By tracing the inheritance of two characteristics very simply through meiosis and fertilisation, it is possible to see a number of potential offspring variations.
- Some traits are not determined by simple dominance, but are the result of multiple alleles, which again further increases the potential for variation.
- Mutations that occur during meiosis can also introduce new allele variants.

GENOTYPE: The entire set of genes in an organism

GENETIC CONSEQUENCE OF MEIOSIS:

- 1 cell undergoes 2 meiotic divisions to generate 4 haploid cells
- The genes in each are combo
- Crossing over & random segregation, allowing individual alleles of mat and pat to assort independently = GENETIC VARIATION
- arms of homologous chromosomes break & exchange gen mat.
- Genes that occur on same chromosome said to be LINKED
- Crossing over ensures that not all linked genes are inherited together
- The exchange introduces genetic variation.
- Independent assortment also = variation
- Each pair separates (during anaphase 1) and 1 entire chromosome of each pair moves into daughter cells (each chromosome still has 2 sister chromatids attached)
  This separation of maternal and paternal chromosomes not only halves the chromosome number in gametes, but also leads to genetic variation, depending on which chromosome (paternal or maternal) of each pair ends up in which daughter cell.
- This is termed independent assortment of chromosomes and produces different combinations of genes in different gametes.

GENETIC CONSEQUENCE OF FERTILISATION:

- 2 haploid gametes fuse to form diploid zygote
- The genes in zygote are a combo of genes contributed to parents: 50% from both (not taking into account material that was exchanged during crossing over).

VARIABILITY:

- Variation in gene content of gametes give rise to variations, increasing variability
- Mutation may further contribute to genetic variation in an individual & genetic variability within a population.

MODELS OF MEIOSIS are simplifications of actual process, designed to demonstrate specific aspects, such as introduction of genetic variation. These models have limitations, for example not demonstrating all aspects of the process.

5.4.2 Model the formation of new combinations of genotype produced during meiosis

- Interpreting examples of autosomal, sex- linkage, co- dominance, incomplete dominance & multiple alleles
- Constructing and interpreting information & data from pedigrees & Punnett squares

- The interaction of alleles from each parent determines the genotype and phenotype of the offspring.
- Where alleles are located will determine whether an offspring inherits a trait

Definitions:

- GENE - a section of DNA encoding particular characteristics
- ALLELE - alternative forms of a gene
- HOMOZYGOUS - identical alleles in a gene pair
- HERTEROZYGOUS - different alleles in a gene pair

Allele location:

autosomal inheritance

- when traits are passed on the autosomes (not sex-chromosomes)
- In autosomal inheritance, an offspring will inherit one set of chromosomes from each parent equally.
- Autosomal characteristics are passed on to both sexes with equal frequency.

sex-linkage inheritance

- When traits are passed on the sex chromosomes and can be either X-linked or Y-linked.
- Sex-linked traits are passed on the sex chromosomes of an organism.
- In humans this means the X and Y chromosomes.
- Female offspring inherit one maternal and one paternal X chromosome, while male offspring inherit on maternal X and one paternal Y.
- If different genes are present on either X or Y, one sex will be more affected than the other.

Allele interactions:

- Mendelian genetics describes patterns of inheritance where traits are influenced by the interaction of a single pair of alleles.
- Multiple alleles, is when inheritance of a trait is dictated by 3 or more alleles.

Autosomal dominant

- A trait is determined by the expression of a dominant allele.
- In order to express the dominant phenotype, only one copy of the allele is needed
- Black or White

Autosomal recessive

- Describes the pattern of inheritance where two recessive alleles are required to be inherited in order for a trait to be phenotypically expressed.
- Recessive traits may skip generations, and are generally less prevalent in the population that auto-dominant.
- Sex-linked genes may also exhibit simple dominance or recessive patterns of inheritance.

Co-dominance

- When both alleles in a gene pair are fully expressed
- Some alleles in a gene pair may be co-dominant, which results in a third possible phenotype
- Speckled

MODULE 5 Heredity

In-complete dominance

- When an allele for a certain trait is not completely expressed over its paired allele.
- Mixed colour- both alleles partially shown

Pedigrees and Punnett squares:

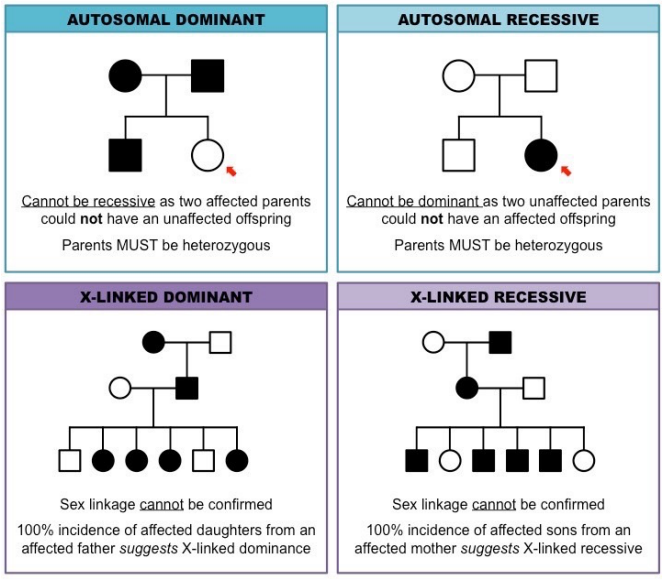
pedigrees

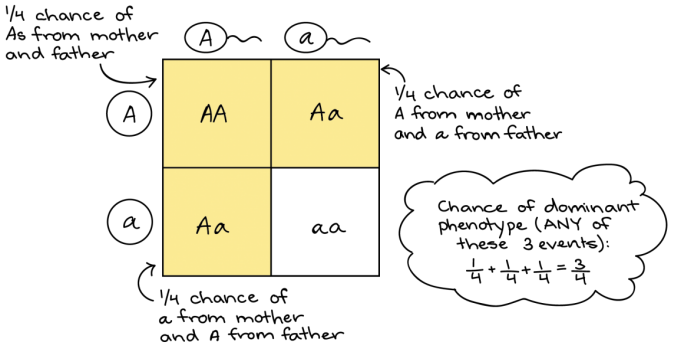
- Pedigrees are charts displaying the phenotypic characteristics of organisms across generations.
- Pedigrees are used to show how traits are passed within families, using standard symbols.
- When interpreting pedigrees we are trying to trace certain alleles passing from parents to offspring.
- It is important to think about whether the parents are homozygous or heterozygous for a trait.
- Dominant traits cannot skip generations, but recessive traits can.
- Sex-linked traits generally affect one gender at a higher frequency.

punnet squares

- diagrams used to predict the genetic outcome of sexual reproduction.

1. Identify the genotypes of the parents.
2. Draw a box with four squares
3. Split up each parent's alleles to align one with each box
4. Fill it in and interpret the information
   a. What are the probabilities of offspring genotypes?
   b. What are the probabilities of offspring phenotypes?
   c. What are the ratios of different genotypes and phenotypes?





INCLUDE KEY FOR BOTH PEDIGREES AND PUNNET SQUARES!!!

5.4.3 collect, record and present data to represent frequencies of characteristics in a population, in order to identify trends, patterns, relationships and limitations in data, for example:

- examining frequency data
- analysing single nucleotide polymorphism (SNP)

SINGLE NUCLEOTIDE POLYMORPHISM:

- a change of a single nucleotide at a specific position on the genome. This may be a substitution (e.g. change A for G), insertion (adding a new nucleotide), or deletion (removing a nucleotide)
- SNPs = 90% of al variation within human population
- Occur at different frequencies in different populations, regions, and cultural groups
- Analysing SNPs allows us to generate population genetics
- It can be a substitution, insertion or deletion.
- SNPs account for more than 90% of all differences across the human population, occurring once every 300 nucleotides on average, but most commonly within non-protein coding DNA.

- Genome studies have identified around 85 million SNP variants across individuals, which shows the potential for SNPs to cause human variation and therefore evolution.

5.5INHERITANCE PATTERNS IN POPULATION
IQ: Can population genetic patterns be predicted with any accuracy?

5.5.1 investigate the use of technologies to determine inheritance patterns in a population using, for example:

DNA Sequencing:

- DNA profiling (fingerprinting) is a technique that allows scientists to determine an individual's unique DNA characteristics
  - Widely used in forensics
  - Used to identify patterns of inheritance

METHOD:

1. Isolate DNA from cells
2. Identification of sequential order of nucleotides
3. Computational processing
   comparison of whole genome
   transcription and translation of genes in silico

- Single nucleotide information
- Each individual has a different genome
- Identification of differences and similarities
- Modern computation allows patterns to be identified
- Trace inheritance of genes, alleles and SNPs between people

DNA PROFILING:

- Technique allowing for identification of an individual's DNA characteristics, this is unique to each individual, like a fingerprint

METHOD:

1. Collect DNA samples from cells (common practice: blood, hair follicles, skin, mouth swabs)
2. Digest DNA- cut the DNA into small pieces using restriction enzyme
3. DNA fragments separated by gel electrophoresis
4. Gel visualised to show band pattern, and 'fingerprints' can be compared

- Visualisation of fingerprints allows for comparison of patterns
- Degree of similarity = degree of relatedness
- May observe conserved regions of DNA
- Effectiveness in humans due to large stretches of 'junk DNA'

5.5.2 investigate the use of data analysis from a large-scale collaborative project to identify trends, patterns and relationships, for example:

- the use of population genetics data in conservation management
- population genetics studies used to determine the inheritance of a disease or disorder
- population genetics relating to human evolution

Conservation management:

- Conservation genetics is a field which combines the knowledge and approaches of population and molecular genetics with ecology and biodiversity
- Identifies and proposes strategies to protect species or variants at risk of extinction
  - preserving genetic diversity
- The State of the World's Animal Genetic Resources for Food and Agriculture

Inheritance:

- Haplotype is a group of alleles inherited together from a single parents. They are tightly linked in a cluster on a certain chromosomes
- Haplogroup is a group of similar haplotypes which share a common ancestral SNP
- By sequencing large populations, scientists have been able to gather information on how specific DNA sequences are passed through generations
- International HapMap
  - Collaborative project
  - Helps determine how diseases/disorders are inherited across populations

Human Evolution:

- Different cultural groups are often linked by the prevalence of certain haplotypes
- By mapping haplotypes globally, we can trace the movement and evolution of the human species from its ancestors
- We are able to trace human mitochondrial DNA haplogroups
  - Each group is defined by differences in mitochondrial DNA (mtDNA), which is only inherited through the mother
  - We are able to trace maternal lines
- By analysing mtDNA haplogroups, the evolution of the human race has been traced back to 'Mitochondrial Eve', the most recent common ancestor
  - 200,000 years ago in Africa

GENETIC DATA

CONSERVATION MANAGEMENT

- Uses population genetics to identify alleles at risk / at a low frequency in populations
- Proposes ways to manage and preserve biodiversity
- Preservation of genetic diversity

INHERITANCE OF DISEASE

- Only a 0.8% nucleotide variance among humans
- All genetic disease must be contained within this
- Use of computational genetics allows us to identify variants leading to disease + disorder

HUMAN EVOLUTION

- Different cultural groups are often linked by prevalence of certain genetic patterns (haplotypes)
- Mapping haplotypes allows us to trace the movement and evolution of human species (e.g. mtDNA)

HAPLOTYPE: a group or 'cluster' of alleles inherited together from a single parent

Look up:

- Human Genome Project
- International HapMap Project