Heredity Study Notes

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TOPIC 1 - Reproduction:

Inquiry Question: How does reproduction ensure the continuity of a species

1.1 Sexual and Asexual Reproduction

- *Explain the mechanism of reproduction that ensures the continuity of a species, by analysing sexual and asexual methods of reproduction in a variety of organisms, including:*
  - *animals: advantages of external and internal fertilisation*
  - *plants: asexual and sexual reproduction*
  - *fungi: budding, spores*
  - *bacteria: binary fission*
  - *protists: binary fission, budding*

- Sexual reproduction involves the **fusion of genetic material from two parents**.It produces offspring that are **genetically unique**.
- Asexual reproduction involves only a single parent, which **produces a new individual from part of itself**. It produces offspring that are **genetically identical** to the parent.

Sexual Reproduction:

Advantages:
- Variation in the population
- Species are better able to adapt to their environments
- Disease events less likely to affect entire population

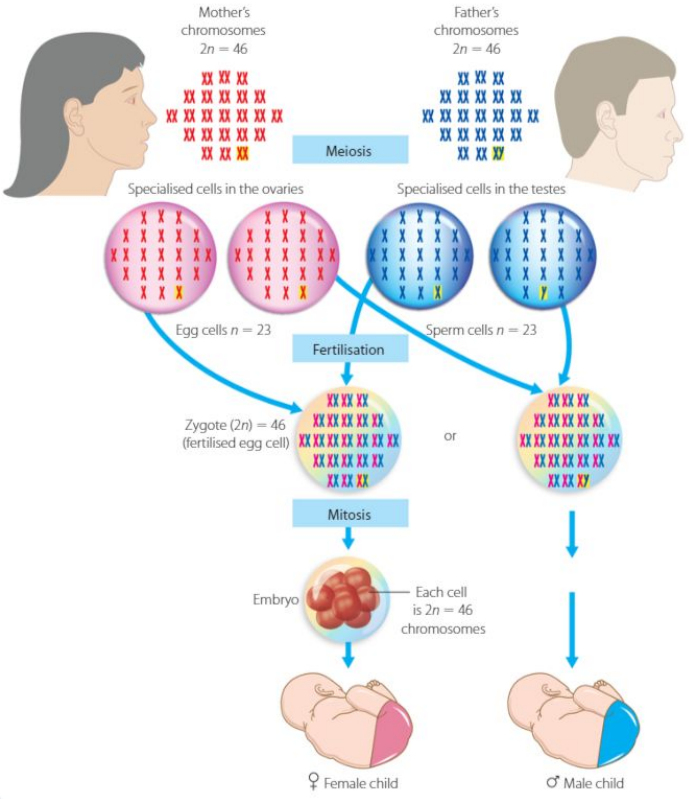
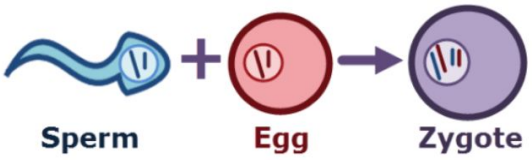
Disadvantages:
- Large time and energy invested
- Requires a mating partner
- Fewer offspring produced

Asexual Reproduction:

Advantages:
- Rapid population of an environment
- No requirement for mates
- Able to be enacted under external pressures
- No requirement for investment in care of offspring

Disadvantages:
- Lack of diversity
- May result in large-scale extinction events
- Reduced ability to adapt to external pressures

Animal Reproduction:

Sexual Reproduction	• Gametes are produced by parent organisms through meiosis • Each gamete contains half the necessary number of chromosomes • The male gamete (sperm) fertilises the female gamete (egg/ovum) by either internal fertilisation or external fertilisation • Fusion of gametes results in the production of a zygote which contains a genetic combination of genetic material from both parental organism • Deemed as advantageous since the combination of chromosomes from two organisms increase variation which assists with survival • Deemed as disadvantageous since it requires mating of two organisms which is dependent on syncing fertility cycles and the production of offspring is slower and less prolific than asexual reproduction • Example → Humans and most other mammals use sexual reproduction in the reproduction of offspring. In humans this occurs through sexual intercourse whereby the male deposits sperm into the vagina of the female.  
Asexual Reproduction	• Asexual reproduction includes fission, budding, fragmentation, and parthenogenesis • Binary Fission

	- The parent organism divides equally in two, so as to produce two genetically identical daughter organisms - This method of cloning occurs in Planaria (flatworms) but is also common to bacteria and protists (e.g. euglena, amoeba) ● Budding - Cells split off the parent organism, generating a smaller daughter organism which eventually separates from the parent - This method of cloning occurs in Hydra but is also common to many species of yeast ● Fragmentation - New organisms grow from a separated fragment of the parent organism - Fragmentation is the breaking of the body into two parts with subsequent regeneration. If the animal is capable of fragmentation, and the part is big enough, a separate individual will regrow - This method of cloning is common to starfish and certain species of annelid worms - Note that in fragmentation, there is generally a noticeable difference in the size of the individuals, whereas in fission, two individuals of approximately the same size are formed. ● Parthenogenesis - Embryos are formed from unfertilised ova (via the production of a diploid egg cells by the female) - This method of cloning occurs in certain species of insect, fish, amphibians and reptiles - Bees use parthenogenesis to produce haploid males (drones) and diploid females (workers). If an egg is fertilized, a queen is produced. The queen bee controls the reproduction of the hive bees to regulate the type of bee produced.

<u>Internal vs External Fertilisation (within animals):</u>

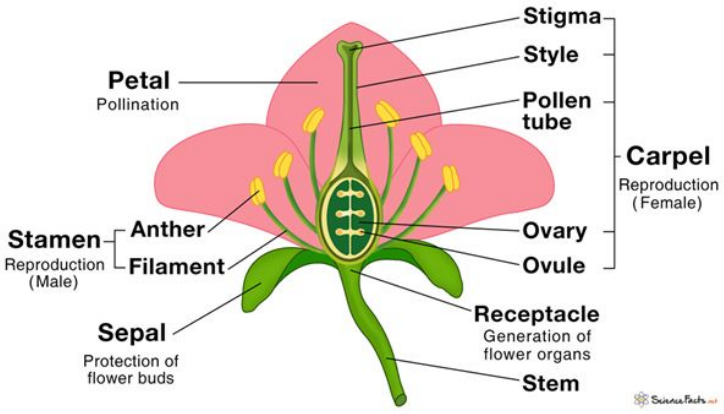
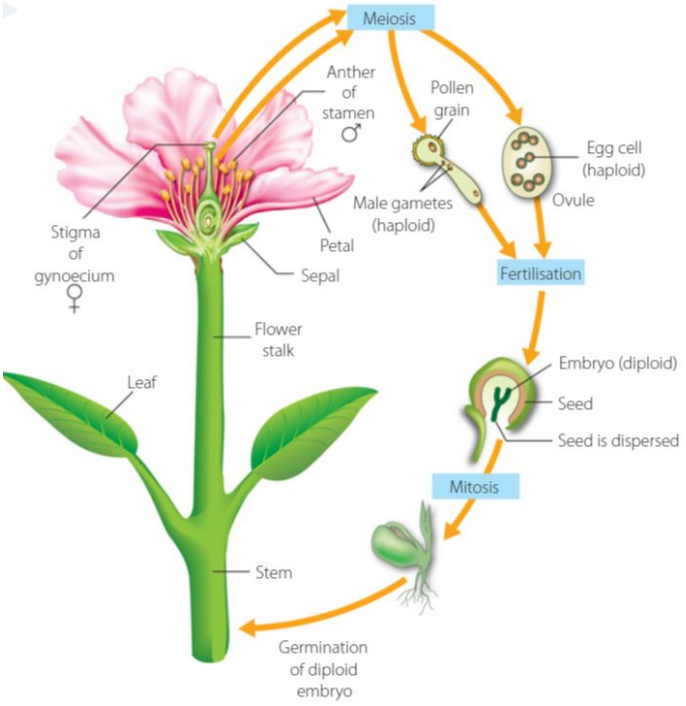
	Internal fertilisation	External fertilisation
Definition	Internal fertilization is the union of an egg cell with a sperm during sexual reproduction inside the female body. For this to happen there needs to be a method for the male to introduce the sperm into the female's reproductive tract.	External fertilization is a mode of reproduction in which a male organism's sperm fertilizes a female organism's egg outside of the female's body. It is contrasted with internal fertilization, in which sperm are introduced via insemination and then combine with an egg inside the body of a female organism.
No. of gametes produced	Species that use internal reproduction produce relatively few gametes. Because the male directly deposits sperm into the female's body, fewer gametes are needed. External fertilization requires the male and female animals to produce larger numbers of gametes.	A large number of gametes are produced in external fertilisation, this is because otherwise the union of gametes would be much less likely to occur. This is due to the open environment not being overly hospitable.
Hazards in	Disadvantages of internal fertilization are	Millions of eggs must be produced by

process	that there are fewer offspring produced at a given time because it is sometimes difficult for the male and female to come into intimate contact. Additionally, the risk of sexually transmitted diseases also increases.	individuals, and the offspring produced through this method must mature rapidly. The survival rate of eggs produced through broadcast spawning is low.
Benefits	Increased likelihood of fertilisation as egg and sperm are in close proximity with increased protection from the environment which means higher survival rates of offspring.	Large number of gametes produced generally means more offspring. It is also a simpler behavioural process which does not require mating rituals
Strategies in the process	Internal fertilization has the advantage of protecting the fertilized egg from dehydration on land. The embryo is isolated within the female, which limits predation on the young. Internal fertilization also enhances the fertilization of eggs by a specific male.	External fertilization in an aquatic environment protects the eggs from drying out. Broadcast spawning can result in a greater mixture of the genes within a group, leading to higher genetic diversity and a greater chance of species survival in a hostile environment.
Example	Humans are fertilized internally. The male reproductive organ enters the female reproductive organ and sperm is then released. The sperm travels towards the female egg and fertilization occurs in the fallopian tubes of the female.	Salmon, cod, trout, and char are all examples of the fish that externally fertilize. The female and male both release their gametes into the water, where they diffuse together and fertilize. If sperm is released too late, there is a higher chance that a different fish's sperm has already reached the eggs.

<u>Plant Reproduction:</u>

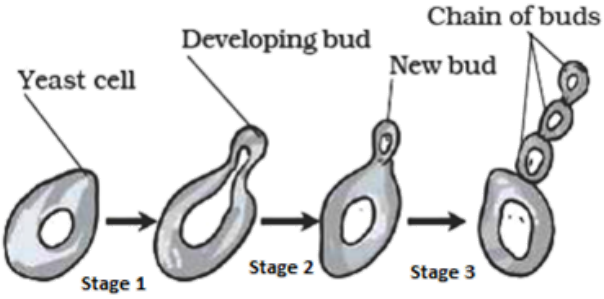
Asexual Reproduction	● Structural modifications to the stem or roots of the plant result in the production of new individuals, without the need for production of seeds or spores ● Can occur through vegetative propagation (asexual reproduction from a plant cutting) and spore formations (e.g. moulds, ferns) ● Vegetative propagation: - Vegetative reproduction results in new plant individuals without the production of seeds or spores. - Many different types of roots exhibit vegetative reproduction. The corm is used by gladiolus and garlic. Bulbs, such as a scaly bulb in lilies and a tunicate bulb in daffodils, are other common examples of this type of reproduction. - A potato is a stem tuber, while parsnip propagates from a taproot. ● This method is deemed advantageous as offspring are clones of parent plants meaning favourable traits are effectively passed through generations. This is economically advantageous for farmers to ensure consistency in their crops. It is also less energy-intensive than sexual reproduction meaning the population can increase rapidly and exploit suitable habitats quickly. ● This method is deemed disadvantageous as pathogens may spread easily from parent to offspring. This reduction in genetic diversity increases the susceptibility of species

	to new diseases and evolution is reduced due to the lack of genetic variation.

Sexual Reproduction	• Flowering plants (angiosperms) sexually reproduction • Flowers are the reproductive organs of sexually reproducing plants • As with animals, offspring are produced by the fusion of two haploid gametes • Male gametes (pollen) are produced and transferred to the female ovules. This process is pollination. (assisted by wind/insects) • After pollination, fertilisation occurs and the ovules grow into seed in a fruit which disperse once ripe. The seed is then freed from the fruit. • This method is considered advantageous as it creates genetic diversity within a species, leading to higher levels of disease resistance and a great ability to adapt to changing conditions • This method is considered to be disadvantageous as it can prevent favourable genes from being passed to offspring which is not possible for an isolated organism  

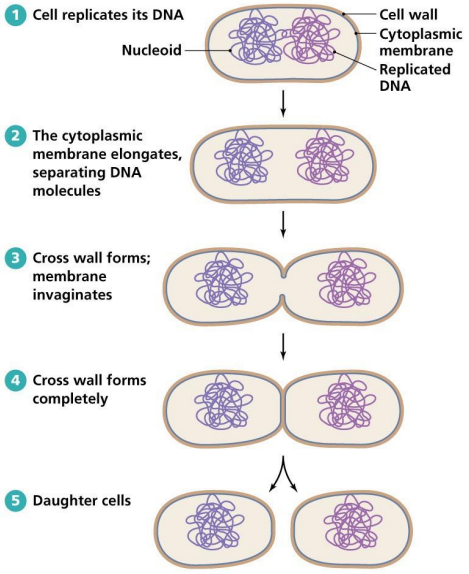
Fungi Reproduction:

The key difference between budding and spore formation is that budding is a type of asexual reproduction in which a new organism originates from a small bud-like structure developed on the parent organism, while the spore formation is a form of asexual reproduction in which new individuals originate directly from the spores of the parent.

Asexual Reproduction	● Production of spores allows for offspring to be widely distributed in the environment, increasing colonisation. ● They can also be produced easily in large numbers
Sexual Reproduction	● Sexual reproduction in the fungi consists of three sequential stages: plasmogamy, karyogamy, and meiosis. The diploid chromosomes are pulled apart into two daughter cells, each containing a single set of chromosomes (a haploid state) ● Plasmogamy: - The fusion of two protoplasts (the contents of the two cells), brings together two compatible haploid nuclei. - At this point, two nuclear types are present in the same cell, but the nuclei have not yet fused. ● Karyogamy: - Results in the fusion of these haploid nuclei and the formation of a diploid nucleus (i.e., a nucleus containing two sets of chromosomes, one from each parent). - The cell formed by karyogamy is called the zygote. - In most fungi the zygote is the only cell in the entire life cycle that is diploid. - Once karyogamy has occurred, meiosis (cell division that reduces the chromosome number to one set per cell) generally follows and restores the haploid phase. The haploid nuclei that result from meiosis are generally incorporated in spores called meiospores.
Budding	● Budding: the nucleus divides and a bulge form in the side of the cell, which is then split off by cytokinesis and the bud detaches itself from the mother cell ● Buds are produced by one parent only (through mitosis) and are genetically identical to that parent. Spores allow fungi to expand their distribution and colonize new environments. They may be released from the parent thallus, either outside or within a special reproductive sac called a sporangium. ● This occurs in yeast 
Spores	● Spores: mitosis produces genetically identical cells to the parent which are distributed into the environment by wind or vectors. ● Fungi produce spores that disperse from the parent organism by either floating on the

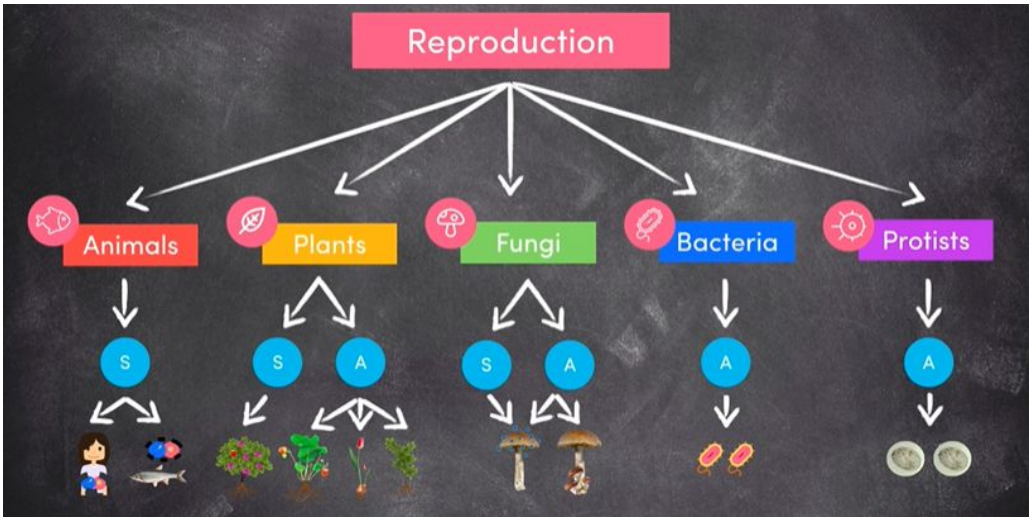
	wind or hitching a ride on an animal. Fungal spores are smaller and lighter than plant seeds. The giant puffball mushroom bursts open and releases trillions of spores

Bacteria Reproduction:

Binary Fission	• A method of asexual reproduction • A single cell divides into two identical daughter cells • Begins with DNA replication where the genetic information of the bacteria is copied and divided into two • The cell elongates and splits into two (cytokinesis), producing daughter cells with identical genetic information • This method is very rapid and only requires a single organism in order to produce offspring • Disadvantageous as it results in a lack of genetic diversity. This means the population has a lower chance of organism survival. This can be overcome by high rates of mutation during DNA replication and HGT by plasmids which can be passed from bacteria to bacteria 

Protists Reproduction:

Binary Fission	• The predominant method of asexual reproduction for protists • Binary fission occurs when a single protist divides its nucleus and then divides itself into two separate organisms. • The organism will then divide into separate organisms for each nuclei that has been created.
Budding	• Budding occurs when a new organism grows from the body of the parent organism to form a new colony.



1.2 Fertilisation and Implantation

- *Analyse the features of fertilisation, implantation and hormonal control of pregnancy and birth in mammals*

Sexual reproduction begins with the development of gametes. In females this occurs in the ovaries where eggs are produced and released into the fallopian tubes. There is a 12-24 hour window after release called ovulation during which fertilisation has the chance to occur. If fertilisation occurs the zygote begins to divide and migrate from the fallopian tubes into the uterus. The blastocyst then embeds itself into the wall of the uterus, this establishes pregnancy.

- **Fertilisation** → the fusion of two haploid gametes to form a single diploid zygote and in turn, initiate the development of a new organism. Also known as generative fertilisation, insemination, pollination, fecundation, syngamy and impregnation, is the fusion of gametes to give rise to a new individual organism or offspring and initiate its development.
- **Implantation** → when a fertilised egg adheres to the wall of the uterus. Implantation is the stage of pregnancy at which the embryo adheres to the wall of the uterus. At this stage of prenatal development, the conceptus is called a blastocyst. It is by this adhesion that the embryo receives oxygen and nutrients from the mother to be able to grow.
- Sexual reproduction begins with development of gametes
- In females this occurs in the ovaries, where eggs (ovum) are produced and released into the fallopian tubes
- **Fertilisation** occurs between 12-24 hours after release called ovulation; where the fusion of an egg and a sperm cell can be successful
- After **fertilisation** has occurred the zygote begins to divide and migrate from the fallopian tubes into the uterus
- The blastocyst (the ball of divided cells) implants itself into the wall of the uterus called the endometrium.
- The endometrium provides nutrients and oxygen to the embryo.
- This happens about a week after fertilisation and established pregnancy.

<u>Hormones:</u>

- Definition → hormones are chemical messengers produced by the body which travel in the blood to other cells where they have an effect
- Pituitary gland → an endocrine gland situated just below the hypothalamus that secretes a number of hormones that regulate other endocrine glands including ovaries and testes
- Hormones play a vital role in all aspects of the development and function of the male and female reproductive systems

<u>Menstrual Cycle:</u>

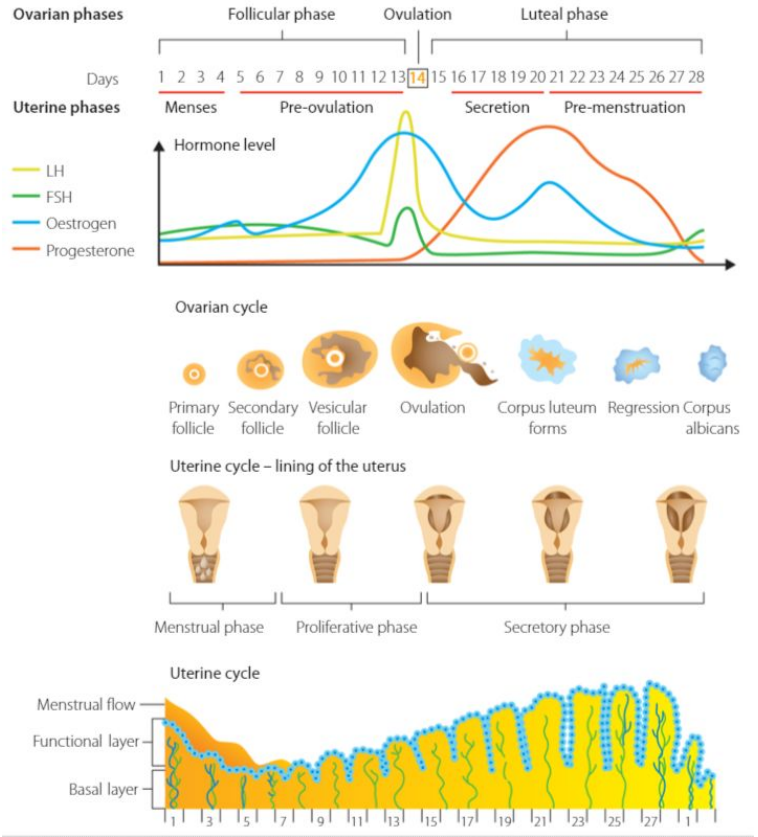
- The menstrual cycle is the regular natural change that occurs in the female reproductive system that makes pregnancy possible. The cycle is required for the

production of oocytes, and for the preparation of the uterus for pregnancy. The menstrual cycle occurs due to the rise and fall of estrogen.

- The changes in the ovaries are simultaneous with changes in the uterus which is the menstrual cycle
- During menstruation, the endometrium breaks down and tears away and this is accompanied by bleeding
- The first day of bleeding is the beginning of the follicular phase which ends on the day of ovulation
- After menstruation, a new endometrium is formed
- After ovulation, the corpus luteum, which is enlarging within the ovary, secretes the hormone progesterone, as well as some oestrogen into the bloodstream
- If an egg is fertilised, it becomes implanted on the uterus and pregnancy results
- There is a secretion of progesterone and oestrogen in the uterine wall, these are produced by the corpus luteum and later by the placenta
- During pregnancy the placenta forms and this also creates HCG
- **HCG** → a hormone produced by the placenta after implantation. The presence of HCG is detected in some pregnancy tests (HCG pregnancy strip tests). HCG supports the normal development of an egg in a woman's ovary and stimulates the release of the egg during ovulation. HCG is secreted from the placenta during pregnancy. The hormone stimulates the corpus luteum to produce progesterone to maintain the pregnancy. Smaller amounts of hCG are also produced in the pituitary gland, the liver, and the colon
- **Corpus luteum** → a mass of cells that form in an ovary and is responsible for the production of the hormone progesterone during early pregnancy. Corpus luteum depends on whether or not fertilisation occurs.

<u>The Ovarian Cycle</u>

- Females are born with all the eggs they will have in their lifetime
- Hormones that are secreted during puberty trigger the development and maturation of ova each month (except during pregnancy) until menopause.
- At the start of a cycle, a few follicles begin to develop but usually only one enlarges more than others to reach maturity
- Ova in the ovaries become surrounded by a single layer of cells that envelop them and begin to divide. This results in the formation of primary follicles in the ovary
- Follicular Phase: follicle cells secrete fluid pushes the egg to one side of the follicle



    - During this phase uterus sheds lining
    - Egg moves to uterus
    - Maturity of follicle takes 10-14 days
    - Follicle cells secrete fluid which pushes the egg to one side of the follicle
    - There is a surge of LH in this time which results in ovulation
    - LH surge also stimulates the next phase of the ovarian cycle where the corpus luteum forms and progesterone is synthesised
    - An egg is released (ovulation) and the egg moves towards the uterus
    - If there is sperm present then fertilisation can occur
    - Ovulation usually occurs in the middle of the cycle
- Luteinising Phase
    - Occurs for 14 days
    - Beings after ovulation
    - Large masses of these cells are called corpus luteum
    - Secrets progesterone
    - This is the phase after ovulation when the burst follicle in the ovary enlarges and changes colour, building up a yellow protein called lutein
    - The corpus luteum secretes the hormone progesterone which acts on the uterus, preparing for pregnancy

<u>Mammalian Pregnancy:</u>

- The hormones that have the most significant role during pregnancy include oestrogen and progesterone
- Oestrogen:
    - Made by the placenta during pregnancy
    - Stimulates the female's body to release an egg
    - Aids blood flow to the developing mammal and helps to stimulate and aid the development of different organs
    - Secreted in the later months of pregnancy to produce progesterone
- Progesterone:
    - Initially released by the ovaries and then later by the placenta
    - In early stages of pregnancy, it stimulates the thickening of the uterine lining
    - As pregnancy continues, progesterone levels rise allowing the placenta to work properly and the uterus to relax
    - Help's mothers immune system to tolerate the growing infant (usually recognised by the mother's body as foreign
- Oxytocin:
    - Stimulates the production of milk
    - Causes the uterine muscles to contract → causing labour and birth
    - Excreted from the pituitary gland
    - Stretches the cervix for birth

<u>Hormones in the female reproductive cycle:</u>

- Endocrine glands regulate and control the ovarian and menstrual cycles in a coordinated manner and these cycles are synchronised to ensure fertility → this ensures continuity of a species

- The pituitary gland secretes a number of hormones that regulate other endocrine glands, including ovaries in females
- Oestrogen and progesterone, produced by the ovaries and controlled by hormones of the pituitary regulate the:
  - Ovarian cycle
  - Menstrual cycle
  - Maintenance of pregnancy
  - Preparation and maintenance of lactation
- The pituitary secretes two gonadotropic hormones:
  - Follicle stimulating hormone (FSH) which causes follicle development in women, causing an egg to mature in an ovary and it also stimulates ovaries to secrete oestrogen. Also causes sperm production in men.
  - Luteinising hormone (LH) which causes the development of the corpus luteum after ovulation as well as lactation. Also causes production of testosterone in men.

1.3 Manipulating Reproduction in Agriculture

- *Evaluate the impact of scientific knowledge on the manipulation of plant and animal reproduction in agriculture*

<u>Scientific Knowledge:</u>

- The increase in proliferation of scientific knowledge means that there have been large advancements in agriculture, enabling processes to become more efficient and productive.
- By understanding reproduction it is possible to manipulate this to receive desired outcomes.
- Scientific knowledge including understanding of seasonal breeding, hormonal regulation, pregnancy and embryonic development is necessary for the manipulation of reproduction in agriculture

<u>Selective Breeding:</u>

- This is based on the understanding that phenotypic traits are heredity so farmers selectively mate plants with desirable traits
- In agriculture it is mostly done to produce higher quality food or more yield
- This may influence cross-breeding or pure breeding practices
- Many current breeds used in agriculture have been produced through selective breeding practices (e.g. Angus cows)
- Steps:
  - Determine the desirable traits
  - Interbreed parents who show desirable traits
  - Select the offspring with the best form of the trait and inbreed the offspring
  - Continue this process until the population fully exhibits the desired trait

Issue	Impact
Health of the animal or plant	Crops usually appear unaffected by a transgene but animals may experience adverse effects that affect growth rates
Uncontrollable pest plant species	Only a concern if the transgene promote rapid growth and there was a selective advantage to having the genes in a wild environment
Loss of biodiversity	Reduction of genetic variation may be harmful with environmental changes. However, GM technology may accelerate the trend of used to re-introduce characteristics into the population specifically with seed banks
Reduced genetic variation	Reduces genetic variation, increasing the likelihood of detrimental homozygous recessive traits in offspring

<u>Artificial Insemination:</u>

- Benefits:
  - Timing meaning able to synchronise births
  - Passing of favourable traits (e.g. increased milk production or quality of meat)
  - Ensuring successful pregnancy increases yields
  - Semen can be transported from one place to another (without moving entire animal)
- Method:
  1. Detection of female cows in oestrus (sexually receptive)
  2. Collection of semen - may be performed manually using an artificial vagina or by stimulation
  3. Insemination usually performed using an insemination gun which shoots semen into the cervix of the desired animal

<u>Artificial Pollination:</u>

- Benefits:
  - Cross-breeding of favourable traits
  - Self-pollination (i.e. creation of genetically similar offspring)
  - Ensuring successful pollination of all plants, resulting in high crop yields
- Method:
  1. Pollen (sperm) removed from stamen of one plant
  2. Pollen applied to the stigma of another plant
  3. Pollen fertilises ovum

<u>Genetic Engineering:</u>

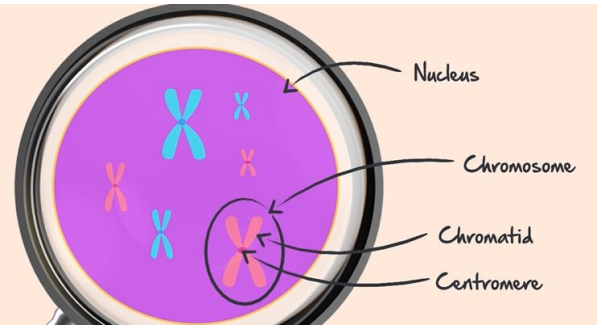
- Knowledge of DNA structure and improvement of genetic techniques has allowed agriculturalists to manipulate on a fundamental level.
- This allows for new desired traits to be put into an organism such as:
  - Bt cotton: insect resistance
  - Golden rice: increased nutritional value
  - Strawberries: frost resistant
- Roughly 170.3 million hectares of GM crops were grown globally in 2012

TOPIC 2 - Cell Replication:

Inquiry Question: How important is it for genetic material to be replicated exactly

- *Model the processes involved in cell replication including but not limited to:*
  - *Mitosis and meiosis*
  - *DNA replication using the Watson and Crick DNA model, including nucleotide composition, pairing and bonding*

Cell division is essential to the survival of all organisms - whether it is to replicate itself (bacteria/fungi/unicellular organisms) or for growth/repair and maintenance of cells/tissues within a multicellular organism.

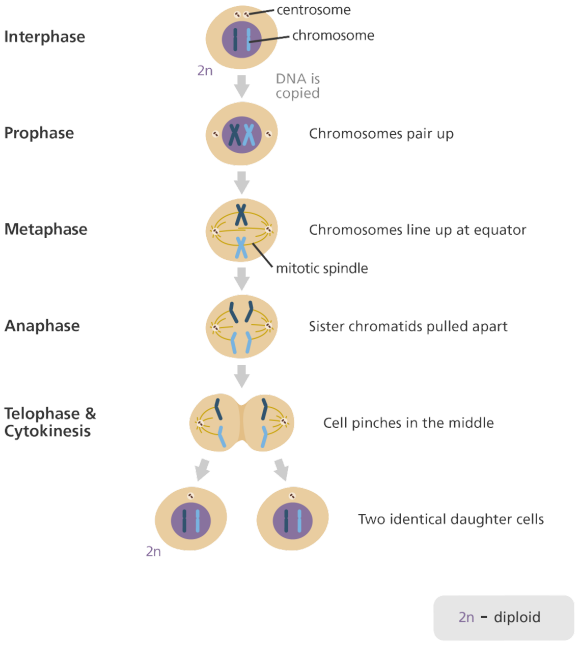


Cell replication → the process by which cells replicate their genetic material and divide to form new cells

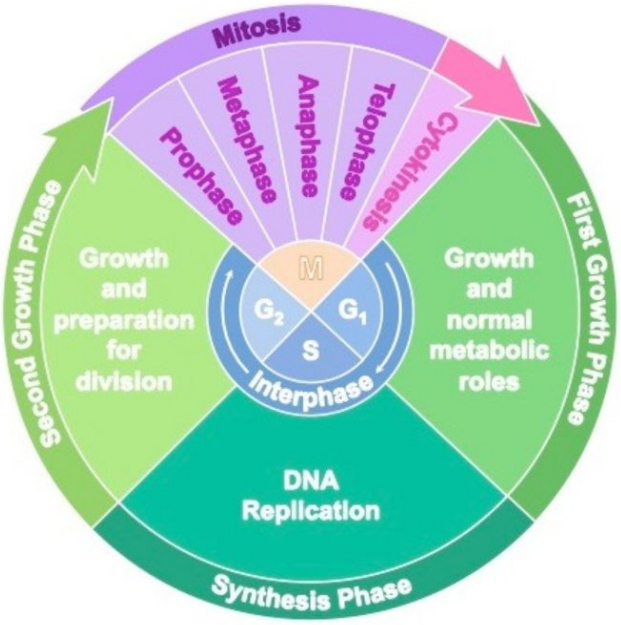
2.1 Processes of Cell Replication

2.1.1 Mitosis

- A type of cell division done by most of the cells in your body
- It's important for cells to divide so that growth can occur as well as repairing damage in your body
- Mitosis DOES NOT make sperm or egg cells
- Done to produce identical body cells
- Mitosis only takes up 10% of the whole cell cycle but it is still critical
- The nucleus holds DNA and is vital in making sure mitosis occurs
- Mitosis can only occur when there are condensed units of chromosomes
- Chromosomes → made of DNA and protein (humans have 46 chromosomes, 46 chromosomes are found in most human body cell nuclei)



Cell Cycle:



Interphase:

- Growing, carrying out DNA replication and normal cell functions
- Cell prepares itself for division
- DNA replication occurs to produce two copies of each chromosome
- Cells spend most of their time in interphase

Prophase:

- Prophase → meaning beginning step and the nucleus is still there
- Duplicated chromosomes condense (this means they become thicker and more visible)
- The mitotic spindle forms at either end of the dividing cell. These spindles are composed of stands or microtubules which length and shorten to pull chromatids apart

Metaphase:

- Pairs of condensed chromosomes (sister chromosomes) line up along the equator of the cell
- The nucleus has been disassembled and is no longer there meaning

Anaphase:

- Sister chromatids are drawn to opposite poles of the dividing cell by the mitotic spindle (spindles are fibers that help the chromosomes move the the other ends of the cell)
- Microtubules bind to chromatids at the kinetochore and begin to shorten, separating pairs from each other
- There is now only one copy of each chromosome at either end of the cell

Telophase:

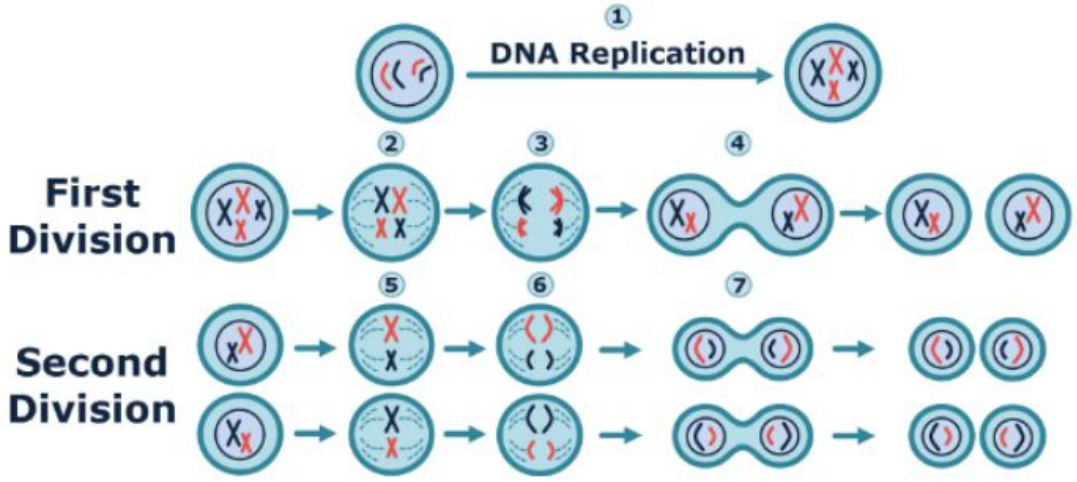
- Chromosomes are actually at the complete opposite ends
- New nuclei are forming on each side to make these two new cells
- The nuclei are surrounding the chromosomes on both sides

Cytokinesis:

- Responsible for the final separation into two cells
- They do this by splitting the cytoplasm which completes after the PMAT mitosis stages

2.1.2 Meiosis

- A process that contributes to genetic variety
- Has two 'rounds'
- Doesn't make body cells instead makes sperm and egg cells → gametes
- Gametes have 23 chromosomes so that when gametes unite they make 46 chromosomes together
- Meiosis is called a reduction division because you have a starting cell that has 46 chromosomes and the ending cell have only 23 chromosomes
- Cells produced from meiosis are non-identical meaning that they are different to each other (random segregation and crossing over) and different to the parent cell



Meiosis I:

- Divide to create 2 daughter cells

Interphase I:

- DNA replication occurs to produce two copies of each chromosome

Prophase I:

- Chromosomes condense and the nuclear envelope breaks down
- Homologous chromosomes pair up, aligning next to each other along their full length.
- Crossing over occurs between homologous chromosomes. This is when segments of DNA at the same locus swap to create new gene combinations

Metaphase I:

- Homologous pairs (not the individual chromosomes) line up along the equator of the separating cell

Anaphase I:

- Homologous pairs are separated → being pulled by the meiotic spindle)
- Sister chromatids remain attached

Telophase I:

- Chromosomes arrive at opposite ends of the cell
- Two diploid daughter cells are formed by cytokinesis

Meiosis II:

- Cells divide again and create 4 daughter cells from already divided cells
- The final four daughter cells are haploid, meaning they have only one copy of each chromosome.

Prophase II:

- Chromosomes condense and the nuclear envelope breaks down

Metaphase II:

- Chromosomes line up along the equator of the dividing cells

Anaphase II:

- Sister chromatids are separated by the spindle microtubules and pulled towards opposite poles of the cell

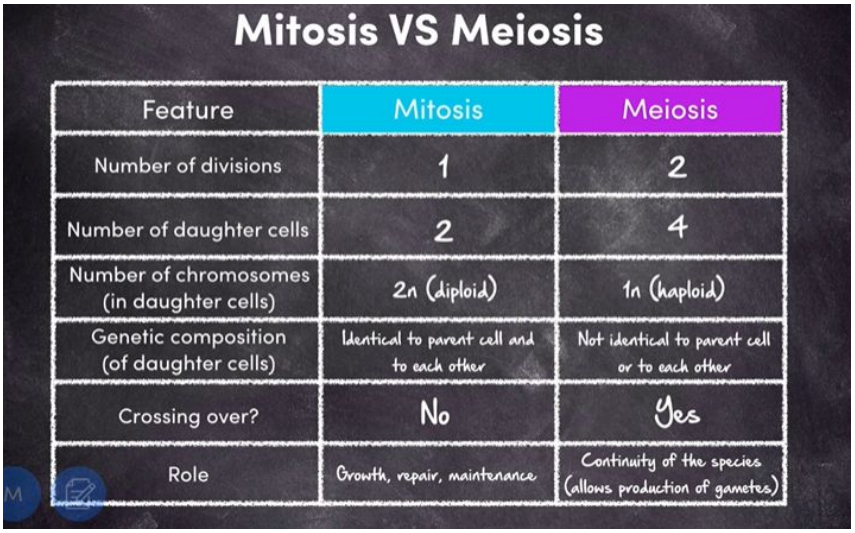
Telophase II:

- Cytokinesis splits the dividing cell into two new cells
- Nuclear membranes form around each set of chromosomes
- Four haploid daughter cells are formed, each containing half the number of chromosomes of the parent cell

Mitosis and Meiosis Comparison:

	Mitosis: Phase of the cell cycle where the cell separates its DNA into two sets and divides, forming two new (identical) cells	**Meiosis:** A two-step process of cell division that is used to make gametes (sex cells)
Function	It is the process of cell renewal and growth in a plant, animal or fungus.	Ensures that all organisms produced via sexual reproduction contain the correct number of chromosomes. Meiosis also produces genetic variation by way of the process of

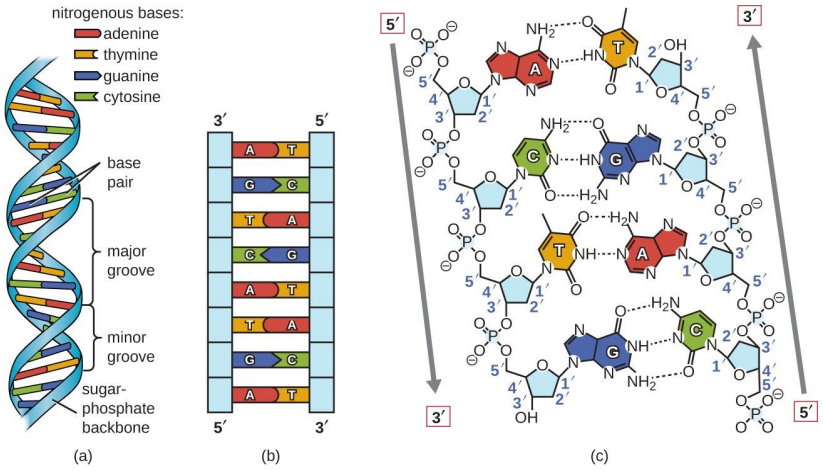
		recombination.
Type of cells involved	Body cells (somatic)	Sex cells (germline)
Location in body	All in body (except reproductive organs)	Sex organs
No. of daughter cells produced	Two daughter cells are produced	Four daughter cells are produced
Change in chromosome number	After mitosis two identical cells are created with the same original number of chromosomes, 46. Haploid cells that are generated through meiosis, such as egg and sperm, only have 23 chromosomes, because, remember, meiosis is a "reduction division."	The number of chromosomes is reduced from 46 (23 pairs) to 23 during the process of meiosis. Because they have only half the total chromosomes in a somatic cell, they are termed haploid (n). In a human egg or sperm, there are 23 chromosomes, one of which is an X or Y.
No. of divisions in the nucleus	One cell divides twice. Mitosis produces 2 diploid cells.	Two successive nuclear divisions occur, Meiosis I (Reduction) and Meiosis II (Division). Meiosis produces 4 haploid cells.
Importance for continuity	Mitosis is important because it creates new body cells that are needed for growth, repair and maintenance. Mitosis allows for you to damage yourself (e.g. hurting your knee) and creates new cells to repair the damage. Without mitosis the body would never be able to heal properly. Because mitosis allows growth it also increases an organism's chances of reaching an age where they can reproduce meaning new organisms are made which is essential for the continuity of species.	Meiosis is important because it allows for the continuity of a species as organisms can produce offspring with new gene combinations. This then creates variation within a population. A population that has variation is more likely to survive a sudden environmental change.



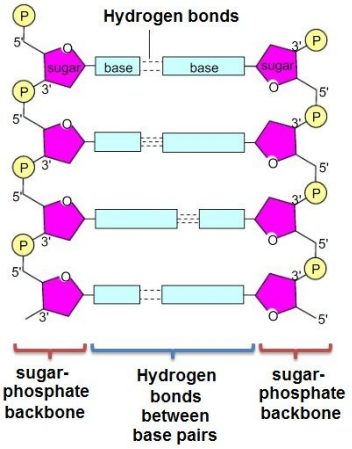
2.1.3 DNA Replication Models

Model of DNA structure (Watson and Crick):

- DNA is a double helical acid molecule which carries genetic information which has been coded into nucleotide bases
- DNA is double stranded, composed of stacked and complementarity bonded nucleotides
- A single nucleotide is a phosphate bound to a sugar group which is then bound to a nitrogenous base (either A - Adenine, T - Thymine, G - Guanine, C - Cytosine)
- Nucleotides are phosphates bonded to sugar, forming a sugar-phosphate backbone. Inwardly facing nitrogenous bases are bonded either C-G or A-T (by hydrogen bonding)

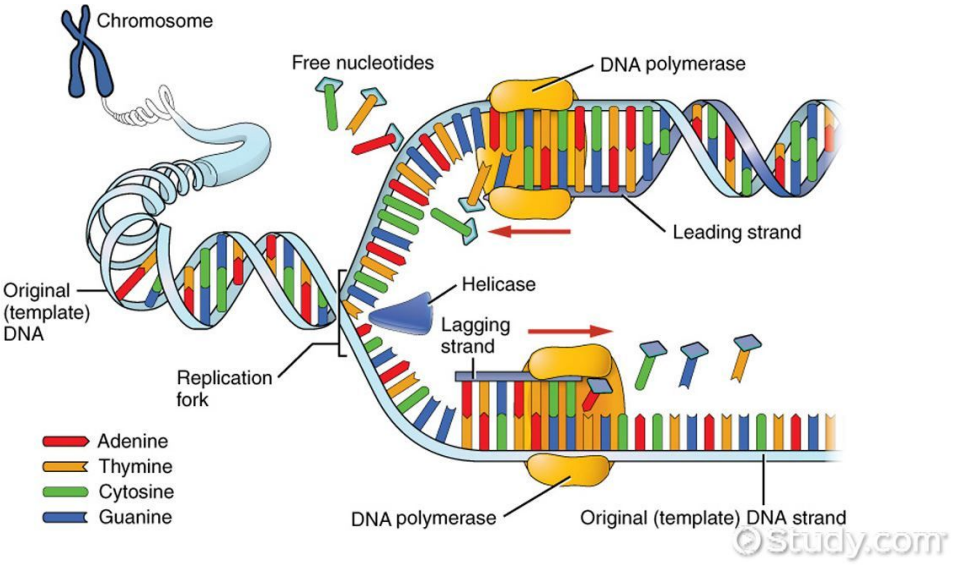


- Watson and Crick:
  - In Watson and Crick's model, the two strands of the DNA double helix are held together by hydrogen bonds between nitrogenous bases on opposite strands.
  - A and T are found opposite to each other on the two strands of the helix, and their functional groups form two hydrogen bonds that hold the strands together
  - The model of DNA replication proposed by Watson and Crick is based on the hydrogen-bonded specificity of the base pairs.
  - Dispersive replication results in daughter duplexes that consist of strands containing only segments of parental DNA and newly synthesized DNA.



- According to the semiconservative replication model, the two original DNA strands (i.e., the two complementary halves of the double helix) separate during replication; each strand then serves as a template for a new DNA strand, which means that each newly synthesized double helix is a combination of one old (or original) and one new DNA strand. Conceptually, semiconservative replication made sense in light of the double helix structural model of DNA, in particular its complementary nature and the fact that adenine always pairs with thymine and cytosine always pairs with guanine.

- The process of DNA replication is as follows:
  1. Initiation (unzipping): the enzyme helicase unwinds and separates complementary DNA strands by breaking the hydrogen bonds between nitrogenous bases → done by DNA polymerase
  2. Elongation: small pieces of RNA called primers bind to the ends of the strands, signalling the starting point of replication. DNA polymerase binds to separated DNA strands at primer sites and begins to add new base pairs which are complementary to the strand. For example, where the polymerase recognises A it will bind a T
  3. Termination: DNA polymerase reaches the end of the DNA molecule and two identical daughter strands have now been produced. Strands recoil into the double helix shape creating two new DNA molecules. Nuclease enzymes essentially 'proofread' the double helix structures.



2.2 Continuity of a Species

- *Asses the effect of the cell replication processes on the continuity of a species*

Genetic variation is needed in a species to ensure that it can live on the continue to thrive

2.2.1 DNA replication

- DNA replication is important to create genetic variation in a species
- DNA is the fundamental hereditary unit which directs all processes in a cell
- Reproduction of cells is dependent upon DNA replication as new cells require more DNA to be made
- By copying by copying the genetic material of a cell, replication ensures that information is transferred through generations
- If DNA wasn't replicated before mitosis and meiosis, cell division would halve the amount of DNA and cells would die due to inadequate amounts of genetic information

2.2.2 Mitosis

- Mitosis is essential for development and growth of organisms
- Mitosis increases the number of cells in an organism, allowing for development of a multicellular body
- Mitosis also allows for old cells to be replaced, ensuring that tissues continue to function effectively and efficiently
- For organisms like humans, mitosis allows us to develop to maturity when we can pass our genetic information onto offspring through sexual reproduction
- Some organisms reproduce by asexual reproduction, which is facilitated by mitosis. In these cases, mitosis creates the next generation of organisms

2.2.3 Meiosis

- Gametes are the end product of meiosis - haploid cells with half the number of requisite chromosomes to make a full cell, capable of all the things cells can do
- The combination of gametes during sexual reproduction creates new organisms, which have inherited traits from both parents
- Unlike mitosis, meiosis purposefully introduces variation. Processes of crossing over independent assortment and random segregation allow for combinations of different alleles, increasing variation in offspring and the wider population
- Genetic diversity is very important for the continuity of species as mutation and variation are essential factors for survival and evolution

TOPIC 3 - DNA and Polypeptide Synthesis:

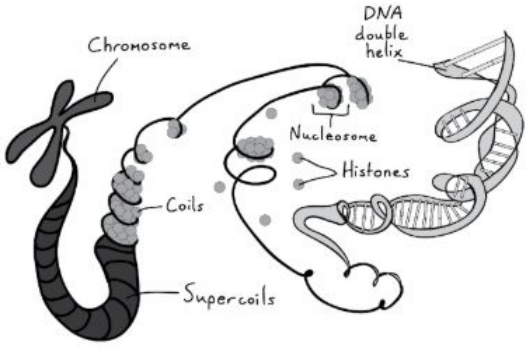
Inquiry Question: Why is polypeptide synthesis important?

3.1 Genetic Material Storage

- *Construct appropriate representations to model and compare the forms in which DNA exists in eukaryotes and prokaryotes*

3.1.1 Eukaryotes

- The defining feature of eukaryotic cells is that they have a nucleus - this is where their DNA is stored.
- Eukaryotic DNA is found wound tightly around small proteins called histones.
- This helps the DNA to condense into a relatively small amount of space.
- Coiled DNA forms supercoils, which are packed together to form chromosomes
- On average, eukaryotes have larger genomes than prokaryotes with long non-coding and repetitive sequences
- Eukaryotic DNA is also linear - it does not link up like prokaryotic DNA



3.1.2 Prokaryotes

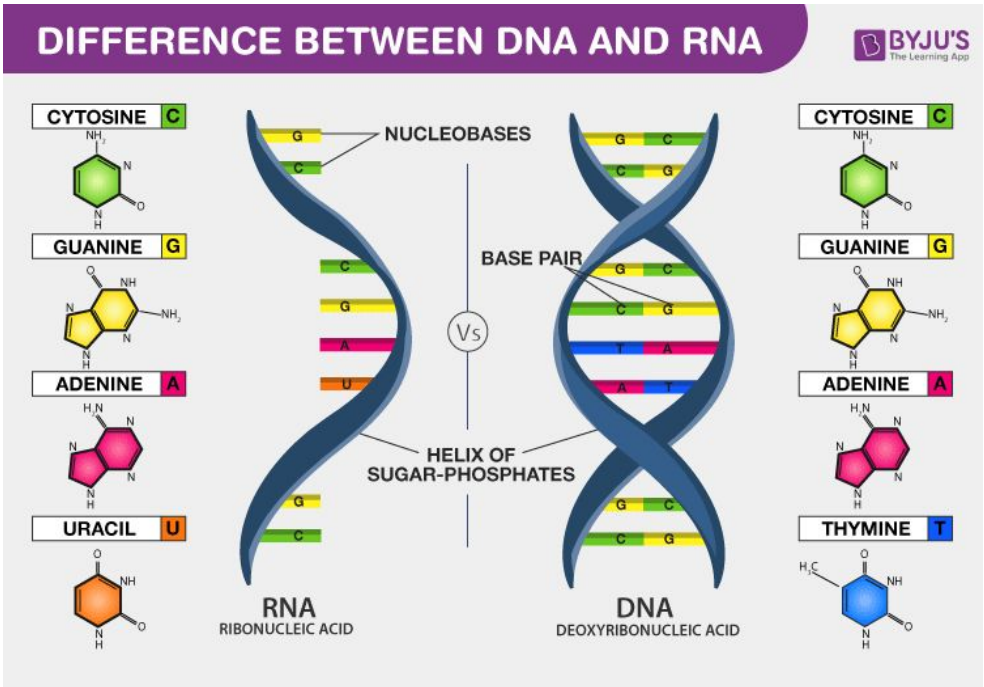
- Prokaryotic cells have free-floating, circular chromosomes, found in the cytoplasm
- The DNA is not bound or packaged by proteins (unlike eukaryotic DNA which is wound by histones)
- Prokaryotes have smaller, more compact genomes with very little repetitive DNA
- Prokaryotes also have small, extra-chromosomal segments of DNA called plasmids
- Plasmids are able to be transferred between organisms to pass genetic material horizontally within generations
- Prokaryotes do not contain introns and therefore polypeptide synthesis cannot occur

3.2 Polypeptide Synthesis Processes

- *Model the process of polypeptide synthesis including:*
    - *Transcription and translation*
    - *Assessing the importance of mRNA and tRNA in transcription and translation*
    - *Analysing the function and importance of polypeptide synthesis*
    - *Assessing how genes and environment affect phenotypic expression*

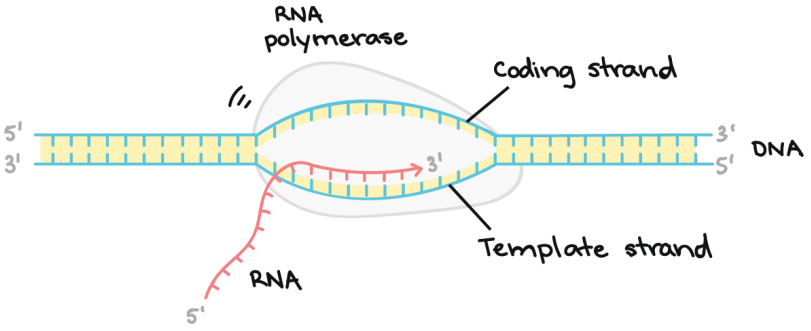
- Definition → refers to how polypeptides are made and it has two stages - transcription and translation
- Transcription and translation are the processes used to turn genetic information into structural and functional molecules used in cells
- Our cells contain a large amount of information stored as DNA
- This information needs to be expressed somehow → expressed through proteins
- The processes used to express genetic information are called transcription and translation
- A step by step guide to producing proteins from DNA
- DNA contains a code for our genes
- Each gene is responsible for a particular group of amino acids (polypeptide) in our body
- The DNA (instruction) needs to be converted into a protein
- Proteins are complex and are made up of one or more polypeptide chain
- DNA → polypeptide chain → protein
- Two steps involved in getting protein from DNA
    1. Transcription - in the nucleus
    2. Translation - in the cytoplasm (ribosome)
- Basic steps include:
    1. DNA in the nucleus
    2. DNA unzipped
    3. Messenger RNA (mRNA) transcribes (transcription) → this is when T is replaced by U
    4. mRNA transported to ribosome
    5. Transfer RNA (tRNA) translates the mRNA (codon of mRNA matches with the anticodon of tRNA is also carrying an amino acid)
    6. An amino acid is joined by a peptide bond to produce a polypeptide
    7. Polypeptide folds to create protein

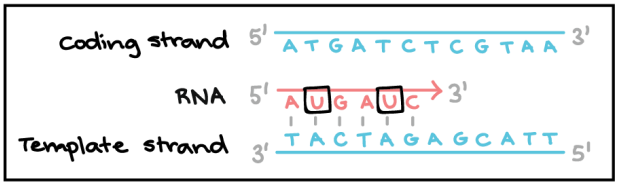
DNA and RNA Comparison:



3.2.1 Transcription

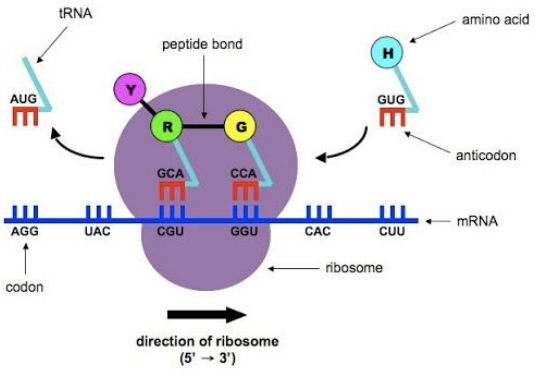
- Transcription → the process of turning genetic information stored in the DNA into an intermediary molecule, mRNA
- There is lots of DNA in human cells which contains thousands of genes and even more
- DNA is important as it contains all our genetic information meaning it must be kept safe hence mRNA
- mRNA transmits information out of the nucleus for processing, is single stranded, contains a ribose sugar (instead of deoxyribose) and has U (Uracil) instead of T (Thymine)
- mRNA = messenger ribonucleic acid
- Throughout transcription, DNA is read by a polymerising enzyme which then adds complementary nucleotides to create a new molecule
- The steps are as follows:
  1. Nucleotides with the help of enzymes, move along one strand of the exposed gene and form a molecule of mRNA
  2. The mRNA copies then move out of the nucleus into the cytoplasm
  3. Moves out of the pores in the nucleus to find a ribosome to attach itself to

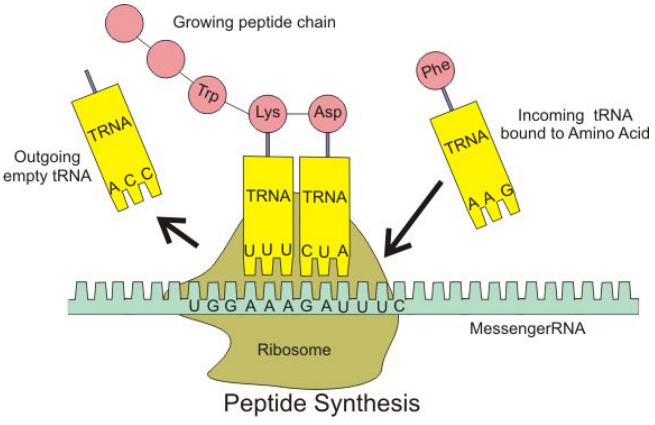




3.2.2 Translation

- Translation → the process of turning information encoded as mRNA into a polypeptide chain
- The genetic sequence of the mRNA molecule is 'read' by ribosomes (which is why transcription takes the mRNA to the ribosome)
- The code is translated in groups of 3 nucleotides called a codon
- Codon → a triplet of bases on the mRNA that pair up with a triplet of bases on the tRNA (anticodon)
- Since there are more codons than amino acids there is some overlap and this means mutations are allowed to the DNA during replication or mRNA during transcription
- The steps are as follows:
    1. The mRNA attaches to a ribosome and attaches to it at the start codon (always AUG)
    2. Two tRNA molecules also come and bind to the ribosome/chain complex bringing with them the amino acid linked with a peptide bond
    3. The appropriate molecule of tRNA attaches to and carries that activated amino acid to the ribosome. Anticodon bases pair with codon in order to bring the specific amino acid to the correct place
    4. A second transfer RNA molecule picks up another activated amino acid and brings it to the ribosome, matching anticodon to codon
    5. Then the first tRNA is released and the amino that was attached to it is not transferred to the second tRNA. This is how the amino acid chain starts to build-up
    6. The process is repeated as the mRNA moves along until it reaches the stop codon
    7. The first tRNA releases its amino acid to the second transfer RNA and leaves the site. The two amino acids form a peptide bond using ATP as its energy source.
    8. The ribosomes read the next codon. The corresponding tRNA brings the activated amino acid to the ribosome
    9. The second tRNA releases its 'load' of amino acids to the third tRNA and leaves the site. Amino acids link up, forming peptide bonds using ATP.
    10. The process repeats itself until the entire message is read and all amino acids are brought in sequence, forming a polypeptide chain
    11. The polypeptide chain folds into its final conformation and is completed and released → this is now a 'mature' protein and is ready for use in the cell.





3.2.3 Function and importance of polypeptide synthesis

- Used to produce proteins in cells
- Proteins are very important for the cell and organism to function
- Effective polypeptide synthesis is vital for multicellular organisms as they require a high degree of coordination which is controlled by proteins
- Often when protein synthesis goes wrong, cancer can occur
- Also important as it adds to the complexity of organisms and this is due to the fact that in both transcription and translation there are opportunities for variation.

3.2.4 How genes and the environment affect phenotype

- Genotype → the genome or genetic make-up of an organism (can include traits that don't change and are ingrained in our genome i.e. eye colour)
- Phenotype → the characteristic that is outwardly displayed on an organism (includes observable traits, biochemistry and physiology)
- Genetic make-up of an organism remains the same throughout its life, the observable traits can change over time due to the environment.
- Genotypes can predict phenotypes
- Genetic information is inherited from parents and contains a set of genes which then tell the cell what to express to create phenotypes
- Certain genes can be 'switched on' at different stages of development or only expressed in response to certain events
- Example → identical twins who have the exact same genetic code but who often develop different characteristics as they age
- Genotype can be considered as containing a range of phenotypic possibilities due to different environmental influences
- Genotype + Environmental Factors = Phenotype

<u>Environment:</u>

- Environmental factors can affect an organism's gene expression and therefore it's phenotype
- Can include temperature, soil pH, nutrition, sunlight exposure
- Example → pH of soil affects colour of Hydrangeas:
  - The flowers exhibit pink and blue colours depending on the pH of the soil
  - If the soil pH is less than 6, the petals will be blue
  - If the soil pH is greater than 7, they are pink

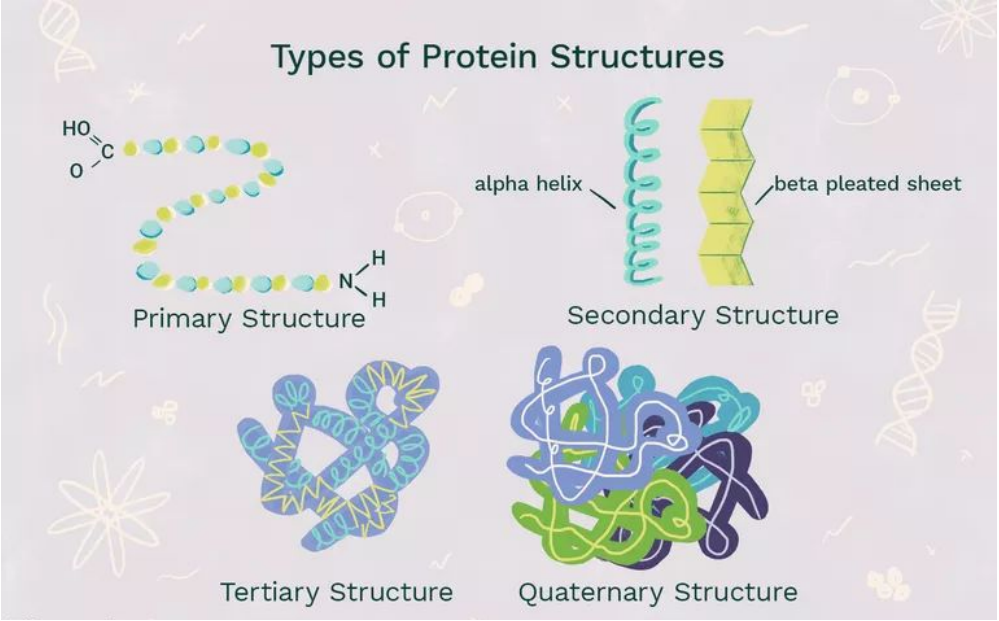
3.3 Protein structure and function

- *Investigate the structure and function of proteins in living things*

3.3.1 Structure

- 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
- Amino acids become part of polypeptide chains through the formation of polypeptide bonds
- There are 20 different amino acids

- Amino acids make up polypeptide chains which then form a 3D shape and become a protein
- Function of a protein depends entirely on its structure
- Protein structure can be categorised into these 4 elements:
  1. Primary → linear sequence, provides information on folding
  2. Secondary → folding or coiling due to hydrogen bonds (can be alpha helix, beta sheet or random coil)
  3. Tertiary → fold further to be more compact due to different bond types
  4. Quaternary → two or more polypeptide chains join to create a single functional protein



3.3.2 Function and Importance

- Proteins are a major component of every cell
- Two main groups of proteins are structural proteins and enzymes
- Structural proteins:
  - Proteins which maintain cell shape and make our connective tissues
  - This means that consuming proteins is super important for the growth, repair and maintenance of tissue
  - Example → Collagen, found in skin and bone as it increases flexibility
- Enzymes:
  - Known as biological catalysts
  - Function is to control the rate of all the chemical reactions that are occuring within a living organism
  - They act on a specific molecule (substrate) to either break it down into a more simple substance or synthesise it into something more complex
  - Example → DNA polymerase which catalyses the formation of the replicate DNA strands by bonding the nucleotides together
- Messenger proteins:

    - Hormones are chemicals which are secreted into the blood and travel to target tissues where they cause a change in activity
    - Hormones cause a change in the activity of the target tissue
    - The role of hormones is to help regulate body processes
- Storage and transport:
    - Storage proteins bind to certain substances and hold them in one place
    - Transport proteins bind to certain substances and carry them around the body
- Immunity:
    - Antibodies are proteins involved in the immune response
    - Antibodies react with antigens to help remove them from the body
    - When an antigen enters the body are released and latch onto them

TOPIC 4 - Genetic Variation:

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

Genes and Alleles:

- Genes are parts of chromosomes and are the units of inheritance passed on from parents to offspring
- Alleles are alternative forms of the same gene
- There are dominant and recessive alleles and dominant traits are stronger and more likely to show up
- Dominant = upper case (e.g. B, R, G)
- Recessive = lower case (e.g. b, r, g)
- Genotype = genetic make-up of the organism
- Phenotype = appearance characteristics or function of the organism
- An organism can be homozygous or heterozygous for a trait
- Homozygous = two alleles the same (both dominant RR or both recessive rr)
- Heterozygous = one of each allele (Rr but the dominant characteristic shows up)

4.1 Predicting Variation

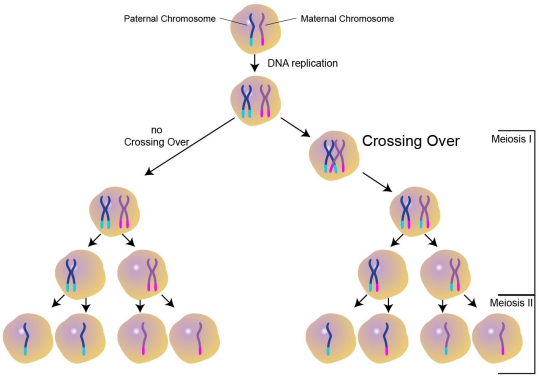
- *Conduct practical investigations to predict variations in the genotype of offspring by modelling meiosis, including the crossing over of homologous chromosome, fertilisation and mutations*

Variation:

- Definition → a term used to describe the differences between the genomes (an organism's complete set of DNA) of individuals of the same species
- Variation is introduced into a population through a variety of different factors
- Variation is first introduced in meiosis.
- Crossing over of homologous chromosomes creates new combinations of chromatids
- Random segregation/crossing over:
    - Occurs in meiosis
    - Random segregation: when chromosomes line up with the matching pairs, they line up in a random order.

- Refers to when 2 homologous chromosomes swap sections of to produce new gene combinations. Critical as it changes the genetic composition of the chromosomes.

- Gene formation sorts chromosomes independently of one another meaning that a number of different chromosome combinations may be formed. For resulting gametes there is a different combination of alleles in each. This introduces variation.
- During fertilisation there is further opportunity for variation to be introduced
- Fertilisation:
  - During fertilisation, a paternal gamete and a maternal gamete fuse to form the first cell (zygote) of the new organism
  - Important source of genetic variation because it allows for different gametes, which contain different alleles, to combine
  - There is a very large number of possible gametes that could combine
  - There are lots of unique organisms in a species, which all have heaps of unique gametes (due to meiosis). Any of these gametes can combine during fertilistation to form different zygotes.
- Alleles from one parent are combined with those from another to restore a full set of chromosomes. This can result in the generation of different offspring genotypes and phenotypes



- By tracing inheritance of two characteristics very simply through meiosis and fertilisation we can already see a number of potential offspring variations.
- This variation is therefore amplified across the thousands of genes present in the human genome which all combine and recombine in different ways.
- Some traits are not determined by simple dominance but are the result of multiple alleles, further increasing the possibility of variation
- Mutation during meiosis (due to an error in DNA replication) may introduce new allele variants which may be passed onto the offspring.
- Mutation:
  - A mutation refers to a change in the base sequence of an organism's DNA
  - There are different kinds of mutations which can happen for lots of different reasons
  - A change in the DNA = different genes = different traits = different phenotype
- Offspring is able to be predicted from parental genotypes to a certain extent

4.2 Combinations of Genotypes

- *Model the formation of new combinations of genotypes produced during meiosis, including:*
    - *Interpreting examples of autosomal, sex-linkage, co-dominance, incomplete dominance and multiple alleles*
    - *Constructing and interpreting information and data from pedigrees and Punnett squares*

Definitions:

- Gene → a section of DNA encoding a particular characteristic
- Allele → alternative forms of a gene
- Homozygous → identical alleles in a gene pair
- Heterozygous → different alleles in a gene pair
- Genotype → alleles present in an organism's chromosomes
- Phenotype → outward appearance of an organism, determined by alleles expressed

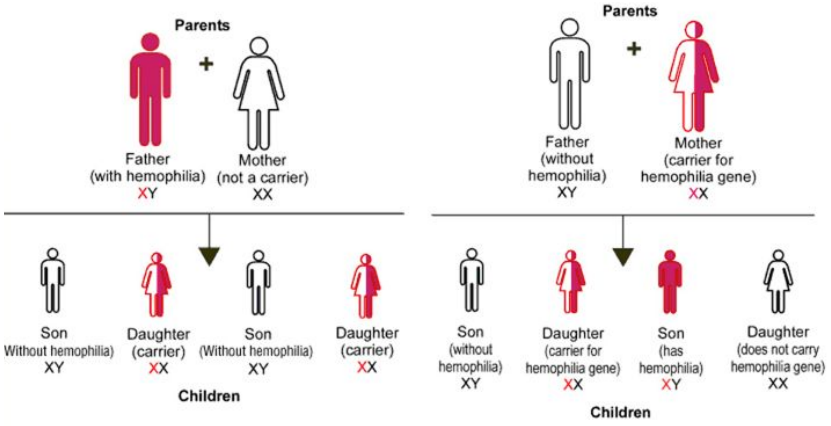
4.2.1 How can the genetic similarities and differences within and between species be compared?

- During meiosis and sexual reproduction, half of the paternal and half of the maternal set of chromosomes are combined
- The combination of alleles from each parent determines the genotype and phenotype of the offspring
- The recombination of alleles allows for new traits to emerge in the population

4.2.2 Where are alleles located?

- There are two seperate categories of inheritance: autosomal and sex-linked
- **Autosomal inheritance** → when alleles are passed on the autosomes (i.e. all chromosomes except for X and Y chromosomes)
    - In autosomal inheritance an offspring will inherit one set of chromosomes from each parent equally
    - For humans, autosomal traits will have their genes located on the first 22 chromosomes - the non-sex chromosomes
    - Autosomal characteristics are passed on to both sexes with equal frequency
- **Sex-linkage inheritance** → when alleles are passed on the sex chromosomes (X or Y). Traits may either be X-linked (i.e. only present on the X chromosome) or Y-linked (i.e. only present on the Y chromosome)
    - Traits are passed on the sex chromosomes of an organism
    - In humans this means the X and Y chromosomes
    - During sexual reproduction, female offspring inherit one maternal X chromosome and one paternal X chromosome (XX). Male offspring inherit one maternal X chromosome and one paternal Y chromosome (XY)
    - If different genes are present on either the X or Y chromosome one sex will be more affect than the other due to this pattern of inheritance
    - Example → Haemophilia is an inherited X-linked disease. The gene for haemophilia is found on the X chromosome. The dominant, undiseased gene is also on the X chromosome. Because females have two X's they may be carriers of

the recessive gene which has the disease but will be unaffected if they also have the dominant allele. If a male inherits the X chromosome with the defect they will always have the disease because the Y chromosome doesn't override it.



4.2.3 How do the alleles interact?

- The interaction of alleles whether they are dominant or recessive will influence whether what version of the gene is expressed

<u>Autosomal dominant inheritance:</u>

- When a trait is determined by the expression of a dominant allele. This means that the phenotype will always be expressed over the other allele inherited.
- In order to express the dominant phenotype, only one copy of an allele is necessary
- Examples → muscular dystrophy and Huntington's disease

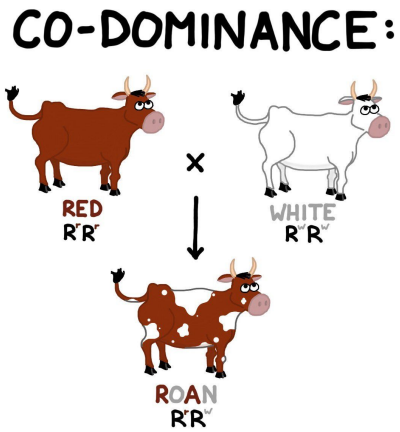
<u>Autosomal recessive inheritance:</u>

- Describes the pattern of inheritance where two recessive alleles are required to be inherited in order for a trait to be phenotypically expressed
- Individuals may be carriers of recessive traits meaning they don't exhibit them but are still able to pass them onto their offspring
- Recessive traits may skip generations and are usually less prevalent in the population than autosomal dominant characteristics
- Examples → sickle cell anemia and cystic fibrosis

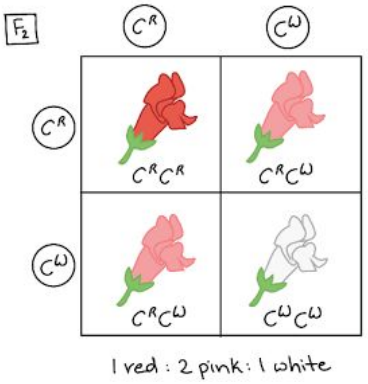
<u>Codominance, multiple alleles and incomplete dominance:</u>

- Sex-linked genes may also exhibit simple dominance or recessive patterns of inheritance
- Genetic is more complex than two alleles interacting and some phenotypic traits are as a result of different inheritance patterns such as codominance, multiple alleles and incomplete dominance.
- Codominance → When both alleles in a gene pair are fully expressed (to varying degrees)
    - Some alleles can be codominant
    - This means that both are fully expressed, resulting in a third possible phenotype.
    - Both alleles are the dominant one
    - The heterozygous individual shows both alleles expressed equally.

- A capital letter represents each of the codominant alleles.
- Example → ABO blood group system can involve alleles for blood type A and type B both being dominant. When an offspring inherits an A allele from one parent and a B allele from another they are both phenotypically expressed in the AB blood type.
- Examples → In roan cows, both brown and white hair is dominant. Both colours are expressed separately



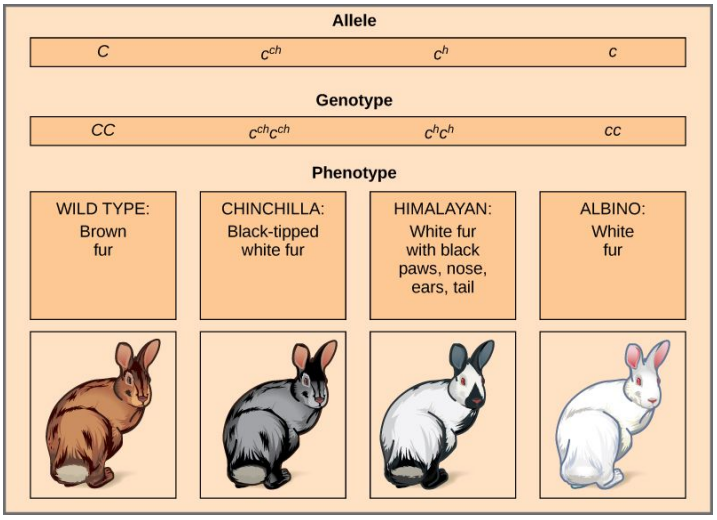
- **Incomplete dominance** → When an allele for a certain trait is not completely expressed over its paired allele. This results in the creation of a third phenotype which is a blended version of the phenotype of both alleles.
  - Incomplete dominance is different to codominance because it is a result of alleles not being fully expressed (i.e. mixed/blended)
  - Examples → Snapdragons mixing to produce pink flowers (cross between red and white flowers) or rabbits with brown fur (cross between red fur and white fur)



- **Multiple** alleles → Inheritance where three or more alleles exist for a single trait
  - Although individually organisms may only inherit two alleles for any given gene, there may be multiple alleles within the population
  - Multiple variations of these genes may display different patterns of dominance
  - Example → Rabbit fur inheritance. The gene for fur colour C has for different alleles; black, chinchilla, himalayan and albino. There is an order of dominance for these alleles: black is dominant to all other alleles, chinchilla is dominant to

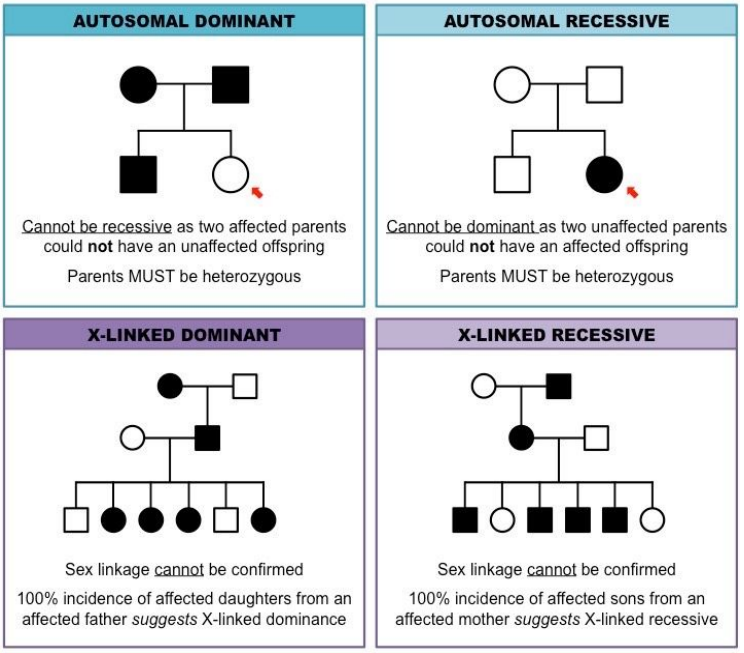
himalayan but recessive to black, himalayan is dominant to albino but recessive to black and chinchilla, albino is recessive to all other alleles.

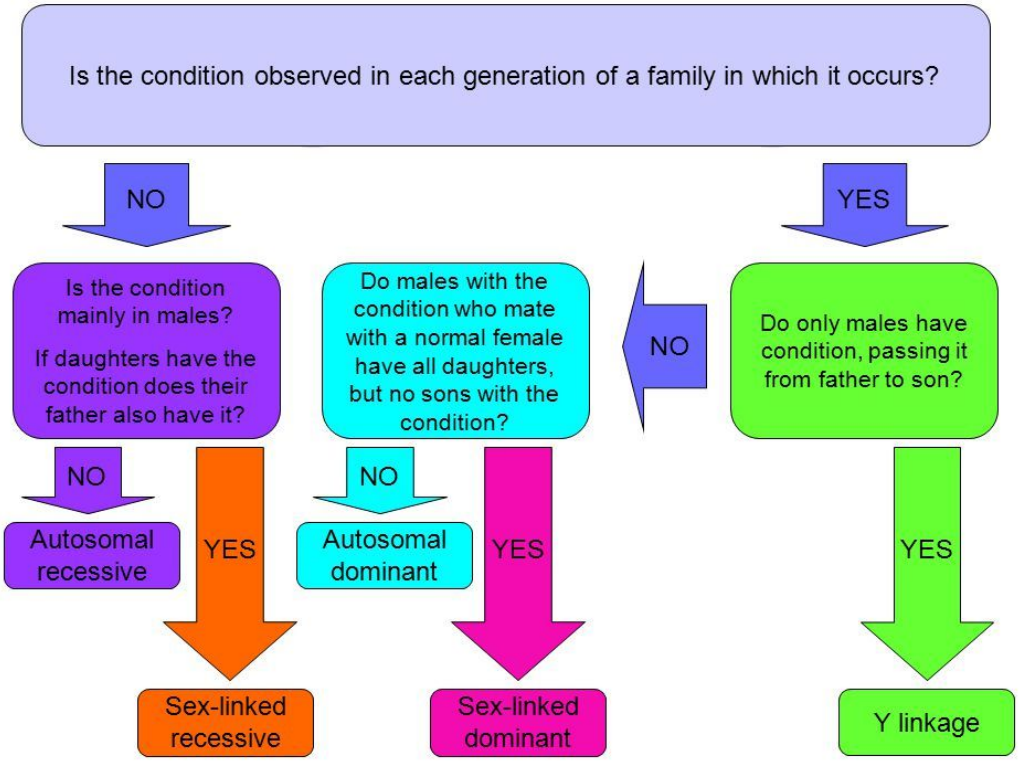
- Inheritance patterns will be determined by the interactions of these different alleles



4.2.4 Constructing and interpreting pedigrees

- Pedigrees → charts displaying the phenotypic characteristics of organisms across generations
- Also known as a family tree
- Shows all the phenotypes of an organism and its ancestors
- Can be used to track or trace characteristics and diseases through many generations
- Can be used to show how traits are passed within families using symbols
- Reading pedigrees enables us to understand how traits are passed from parent to offspring
- By analyzing a pedigree, genotype and phenotype can be determined and also makes it possible to determine whether they are dominant, recessive, autosomal or sex-linked
- Lines between symbols means they are married
- Lines down shows offspring of two above parents
- Females = circles
- Males = squares
- Affected = shaded
- Unaffected = unshaded
- Dominant traits can't skip generations, recessive traits can
- Sex-linked traits generally affect one gender at a higher frequency





4.3 Genetic Data

- *Collect, record and present data to represent frequencies of characteristics in a population in order to identify trends, patterns, relationships and limitations in data:*
    - *Examining frequency data*
    - *Analysing single nucleotide polymorphism (SNPs)*

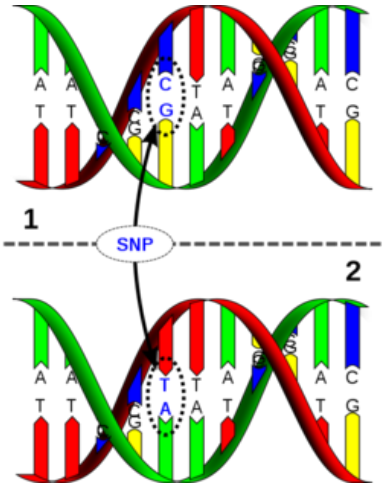
- Genetic differences and similarities within and between species is either determined by at phenotype, genotype, allele or molecular level
- Population genetics → the study of how the gene pool of a population changes over time, leading to a species evolving
- Gene pool → all the alleles of all the genes in a breeding population
- Genetic diversity → the total of all the genetic characteristics in the genetic make-up of a species
- Species that have a greater degree of genetic diversity have a greater potential to adapt and to survive
- Population geneticists study factors that cause changes in allele frequency within a population
- Genetic variation is necessary for a species to adapt to change
- Variation can be affected by population size, mutation, natural selection, genetic drift and migration patterns
- Different species don't exchange genes by interbreeding and they produce sterile offspring
- Macroevolution → over a long period of time, effects whole species

- Microevolution → over a relatively short time period (e.g. bacteria evolving because of antibiotics)
- Genetic variability in a population can be determined by analysing the relative proportion (ratio or %) of a given phenotype, genotype or allele in a population
- Frequency of allele G = $\frac{\text{Number of copies of allele G in the population}}{\text{Total number of copies of the gene (G+g) in the population}}$

4.3.1 Genetic Variation and Frequency of Characteristics

- Genetic variability within a population is essential for evolution by natural selection
- Microevolution can be studied by examining a change in the frequency of alleles in a population over several generations
- Example → skin colour in the red-eyed tree frog is determined by a gene that has two alleles: A (normal) and a (albino)
- Genetic variability in a population can be determined by analysing the relative proportions (ratio or percentage) of a given phenotype, genotype or allele within that population

4.3.2 Single Nucleotide Polymorphism (SNPs)



- Polymorphism → individuals with different phenotypes
- Polymorphism usually arise as a result of a mutation - an error in DNA replication
- SNP → a substitution of a single nucleotide at a specific position in the genome
- A SNP is like a typing error in DNA, where one nucleotide is replaced by another
- SNPs usually arise during DNA replication, where a single nucleotide is incorrectly inserted, creating an error in the DNA sequence at a particular location on a chromosome
- To be termed a SNP (rather than just a mutation), this altered DNA sequence must occur in at least 1% of the population (much more common than mutations)
- SNPs are point mutations
- Usually found within non-coding regions of DNA
- Haplotype is a group of genes (SNPs) that are inherited together from a single parent

<u>Why SNPs:</u>

- Variation in organisms may be associated with phenotypic change but most SNPs occur in non-coding regions of DNA and don't have observable differences
- SNPs are important genetic markers that are currently use to distinguish individuals and to identify things such as disease susceptibility in individuals
- A genetic marker → an identified sequence of DNA at a known site on a chromosome (SNP)
- Individuals within a population show great variation in the genetic markers they have on their DNA - allows scientists to easily tell individuals apart (roughly 10 million SNPs in the human genome)

- Some genetic markers are associated with specific traits or disorders but don't necessarily cause them
- In studies of genetic markers, called genome-wide association studies, computer technology is used to rapidly scan genetic markers across the genomes of many people to find genetic variations associated with a particular disease
- Genome-wide studies are based on the presence of a group of SNP markers (haplotype) associated with a trait rather than trying to link an individual SNP with a trait
- Some applications of identifying haplotypes area:
  - As indicators of disease
  - To establish family lineage and determine the genetic relatedness of individuals
  - To study evolutionary relatedness
- The frequency of SNPs is approximately 1/300 nucleotides in the human genome, giving a total of 10 million SNPs (mostly in non-coding)
- There are many variants meaning it can potentially to drive variation within the human population and then link to evolution
- Advances in technology allow thousands of SNPs to be analysed at the same time (faster and cheaper than sequencing whole genomes)

Limitations of SNPs:

- Scientists have found that many biological questions can be answered using smaller regions of the genome that show polymorphisms
- This data is reliable as long as the regions selected are fairly evenly distributed throughout the genome
- Selection of markers is also important - genetic markers that are closer together give more accurate data → this is because for a haplotype study, SNPs that have been inherited from one parent have to be studied
- If there is crossing over during meiosis, the SNPs on a chromosome might not all be inherited together
- SNP data is being used more frequently

TOPIC 5 - Inheritance Patterns in a Population:

Inquiry Question: Can a population's genetic pattern be predicted with any accuracy?

5.1 DNA Sequencing and Profiling

- *Investigate the use of technologies to determine inheritance patterns in a population using DNA sequencing and profiling*

DNA Sequencing:

- DNA sequencing is the process of determining the nucleic acid sequence – the order of nucleotides in DNA. It includes any method or technology that is used to determine the order of the four bases: adenine, guanine, cytosine, and thymine
- DNA sequencing allows us to find single nucleotide information for entire human genomes

- Each individual will have a different genetic code (except identical twins) and these can be compared using modern computation techniques to determine patterns of inheritance through generations
- Compiling large amounts of sequencing data, we are able to model the changes in the frequencies of genes and alleles in populations over space and time
- Gel allows molecules to be separated by size
- Steps:
    1. Isolate DNA
    2. Run PCR to **amplify** the amount of DNA
    3. Double stranded DNA is separated into single-strands by heating
    4. Run on a gel-electrophoresis
    5. Select the STR's you're going to compare and look for similarities

<u>DNA Profiling:</u>

- DNA profiling is the process of determining an individual's DNA characteristics
- Also called DNA fingerprinting
- A technique that allows scientists to determine an individual's unique DNA characteristics
- Widely used in forensics because it allows for comparison of DNA samples found at crime scenes to help identify individuals
- Can also be useful to identify patterns of inheritance between individuals
- The technique is effective because human DNA contains large stretches of junk DNA, which vary in length and contain different number of repeats
- These are called variable number tandem repeat sequences (VNTRs)
- The individual variability within these sequences allows us to generate individual 'fingerprints' from our DNA
    1. Collection → DNA samples are collected from cells (blood, hair, saliva)
    2. Digestion → DNA cut into small pieces using a restriction enzyme
    3. This creates a mixture of DNA fragments of differing sizes. The composition of this mixture will vary from individual to individual, depending on their DNA sequence
    4. DNA fragments are separated using gel electrophoresis
    5. The gel is visualised to generate an image of the fragments separated into bands. Each band represents a segment of DNA of a certain size. Each individual sample will have a different band pattern due to their individual gene sequence
    6. Patterns of bands are compared

<u>PCR</u>:

- A technique used to amplify DNA in vitro
- PCR is like a scientist doing DNA replication:
    - Involves copying DNA
    - Occurs in a test tube
    - Only a specific region is copied
- Materials needed:
    - DNA sample (e.g. crime scene)

- Free nucleotides
- DNA polymerase → heat sensitive
- Primers → short, single stranded pieces of chemically synthesised DNA
- Buffer → liquid which all of the other ingredients are added; prevents sudden pH changes

- To do a PCR, the mixture needs to be put in a thermal cycler so that the mixture can be heated and cooled in a controlled way → using variations in temperature to control the replication process = PCR

Genetic Testing

- In medicine, DNA sequencing can be used to determine if a patient is at risk of (or affected by) a genetic disease.
- A genetic disease refers to a health condition which is caused by one or more abnormalities in the genome.
- These abnormalities tend to be associated with the presence of particular genes. So, by sequencing a patient's DNA (or, theoretically, their whole genome) and analysing the genes associated with genetic diseases, we can determine whether they have a genetic disease or are likely to develop one.
- For example, sickle cell anaemia is a genetic disorder where people have misshapen red blood cells. It's caused by the substitution of a single nucleotide in the gene that creates haemoglobin. Specifically, the normal DNA code is GAG, and the mutated code is GTG. This changes the amino acid from glutamic acid to valine, so that haemoglobin folds into an abnormal shape. This causes the haemoglobin molecules to clump together, resulting in a crescent-shaped red blood cell which can't carry oxygen as efficiently and can interrupt blood flow. This mutation can be identified using DNA sequencing, so that the person can be diagnosed.
- Determining whether a person has a genetic disease is the first, most important step in ensuring that they receive proper care, treatment and/or management for their condition.

Molecular Biological Research:

- As we've already established, DNA sequencing is a particularly useful tool in scientific research because it can be used to study genomes and the proteins they encode, at a molecular level. This is important in genome mapping, which involves determining the locations of genes and the distances between them.
- This kind of analysis is particularly important in cases where we don't know or understand the gene responsible for a genetic disorder and want to learn more about it in order to improve diagnosis and treatment methods.
- DNA sequencing is also used in research to identify potential drug targets. Drugs are chemicals which produce an effect when administered to the body. To put it in a nutshell, drugs produce their effects in the body by binding to a "target", which is usually a specific enzyme or receptor.
- For example, ibuprofen (the active ingredient in Nurofen) works by binding to and inhibiting cyclooxygenase (an enzyme which promotes inflammation) to reduce pain. Differences in the structure of drug targets (i.e. our receptors and enzymes) lead to

differences in the ability of a drug to bind, so that drugs have different effects on different people.
- DNA sequencing can be used to identify variations in drug binding sites. In this way, DNA sequencing is a step towards personalised medicine - that is, medical treatments tailored to a person's needs, based on the unique variations in their genome.

Evolution:
- We've said it a million times: DNA is like a cell's instruction manual. The order of nucleotides in a DNA molecule stores genetic information, since it can be translated into proteins which then ultimately dictate the functioning of a whole organism.
- On top of coding for the features of a living organism, another, equally amazing feature of DNA is its ability to replicate itself and be passed on from one generation to the next.
- So, DNA doesn't just tell us about the functioning of an organism: it also tells us about where an organism came from, in evolutionary terms!
- This means that DNA sequencing can also be used in evolutionary biology to determine inheritance patterns. That is, it can be used to study how different organisms are related and how they evolved.

Identification:
- Again: everyone has a totally unique DNA sequence. Because of this, DNA sequencing can be used to identify and compare people.
- For example, DNA sequencing can be used to determine someone's biological parents based on their combinations of alleles. It can also be used in forensic investigations to determine the identity of a person at a crime scene.

5.2 Data Analysis

- *Investigate the use of data analysis from a large-scale collaborative project to identify trends, patterns and relationships:*
  - *The use of population genetics data in conservation management*
  - *Population genetics studies used to determine the inheritance of disease or disorder*
  - *Population genetics relating to human evolution*

5.2.1 Conservation Management

- Conservation genetics combines knowledge and approaches from population and molecular genetics with ecology and biodiversity sciences in order to identify and propose strategies to protect species or variants at risk of extinction

5.2.2 Inheritance of diseases and disorders

- There is roughly a 0.8% nucleotide base pair variance among human individuals
- Improvements in computer technology allow scientists to study these variations
- Haplotype → a group of alleles inherited together from a single parent. These alleles are tightly linked in a cluster on certain chromosomes (very likely to be inherited together or 'conserved'

- Haplogroup → a group of similar haplotypes which share a common ancestral single nucleotide polymorphism (SNP)
- By sequencing large sets of populations, scientists have gathered information on how specific DNA sequences are passed down through generations and have determined different haplotypes and haplogroups that exist globally

5.2.3 Human Evolution

- Different cultural groups are often linked by the prevalence of certain haplotypes
- Mapping haplotypes globally, the movement and evolution of human species can be traced
- Example → tracing mitochondrial DNA