

MODULE 5 – HEREDITY

REPRODUCTION

Inquiry question 1: How does reproduction ensure the continuity of a species?

- explain the mechanisms of reproduction that ensure the continuity of a species, by analysing sexual and asexual methods of reproduction in a variety of organisms, including but not limited to:
 - animals: advantages of external and internal fertilisation
 - plants: asexual and sexual reproduction
 - fungi: budding, spores
 - bacteria: binary fission
 - protists: binary fission, budding.

Define reproduction.	The act or process of reproducing specifically: the process by which plants and animals give rise to offspring and which fundamentally consists of the segregation of a portion of the parental body by a sexual or an asexual process and its subsequent growth and differentiation into a new individual.
ANIMALS	
Define internal and external fertilisation.	<p><u>Internal:</u> Internal fertilisation involves the fusion of male and female gametes <i>within a parent's body</i>. Internal fertilisation tends to occur between terrestrial animals. <i>E.g., reptiles, mammals, and birds.</i></p> <p>VS.</p> <p><u>External:</u> External fertilisation involves the fusion of male and female gametes <i>outside a parent's body</i>. External fertilisation tends to occur between aquatic animals. <i>E.g., algae, fish, and amphibians.</i></p>
Compare internal and external fertilisation.	<p>Internal: Inside parent's body, sperm fuses with the egg to form a zygote & mostly mammals and plants.</p> <p>External: Outside parent's body, sperm fuses with the egg to form a zygote & mostly aquatic species</p>
Define continuity of species.	Continuity and change are a core theme in biology that refers to how genetic information is carried forward. The continuity of a species refers to how a species can reproduce offspring's that are favourable to the ambient environment and avoid extinction in general.
How is continuity of species achieved?	Meiosis. The process of meiosis preserves genetic continuity for future offspring by ensuring that two sexually reproducing organisms produce offspring that have the same number of chromosomes as the parents. This is important for several reasons.
Identify the types	<u>Sexual:</u> Requires 2 parents, where the male and female produce gametes (sperm

of reproduction.	<p>and egg cells), in order to reproduce. The offspring's genetic material is derived from their parents, but is <i>not identical, its mixed</i>. This process in mammals is called meiosis.</p> <p style="text-align: center;">VS.</p> <p><u>Asexual</u>: Requires <i>1 parent</i>, where an offspring is produced through cell division, mitosis, which is the process in humans and many other mammals. The offspring has <i>identical genetic materials</i> to the parent, making it a clone.</p>
Describe the main types of asexual reproduction in <u>animals</u>.	Binary fission, budding, vegetative propagation, spore formation, fragmentation, parthenogenesis and apomixis.
Describe sexual reproduction in animals.	<p>In some species, such as fish, the male releases sperm over the eggs after the female has laid them.</p> <p>In other species, such as birds and most mammals, including human beings, the male releases sperm into the female reproductive tract.</p> <p>The female reproductive system is kept moist so that the sperm can travel to the eggs.</p> <p>In internal fertilisation, less eggs are produced compared to external fertilisation.</p>
PLANTS	
Describe the main types of asexual reproduction in <u>plants</u>.	The different types of asexual reproduction are binary fission, budding, vegetative propagation, spore formation (sporogenesis), fragmentation, parthenogenesis, and apomixis.
Describe sexual reproduction in plants.	<p>Angiosperms: Vascular flowering plants where seeds are enclosed in an ovary.</p> <p>Gymnosperms: Vascular plants with enclosed seeds often configured as cones.</p>

<p>Structure of Flowers.</p>	
<p>Male and female parts of a flower.</p>	
<p>Pollination</p>	<p>Pollination is the transfer of pollen from an anther of a plant to the stigma of a plant, later enabling fertilisation and the production of seeds, most often by external pollinating abiotic (wind, water) and biotic (insects, birds) agents to carry out fertilisation and transfer male gametes from the anther to the stigma.</p> <p>There are 2 forms of pollination: 1) Self-pollination and 2) Cross pollination. Pollination requires external agents, such as wind, water, insects (bees) and animals (birds).</p>
<p>How are seeds dispersed once they develop in the ovary?</p>	<p>Seeds are dispersed by animals and birds (eaten and deposited in droppings); by wind, by water and by attaching to animal fur.</p>

FUNGI

<p>What are fungi?</p>	<p>A diverse group of eukaryotic, unicellular, or multicellular, heterotrophic organisms.</p> <p>Above ground = fruiting body</p> <p>Below ground = mycelium</p>
<p>Budding in fungi.</p>	<p>Occurs in yeasts, which the process includes a bud developing on the surface of the yeast cell, with the cytoplasm of the bud being continuous with that of the parent cell. This ensures the continuity of a species, as the genetic information is the same.</p>
<p>Spores in fungi.</p>	<p>Fungal spores are microscopic biological particles that allow fungi to be reproduced. They serve a similar purpose of seeds in the plant world. Fungi decompose organic waste and are essential for recycling of carbon and minerals in our ecosystem. A fungal spore is a haploid cell produced by</p>

	mitosis, from a haploid parent cell.
BACTERIA	
What are bacteria?	<p>Bacteria are small single-celled organisms.</p> <p>Bacteria are found almost everywhere on Earth and are vital to the planet's ecosystems.</p> <p>Some species can live under extreme conditions of temperature and pressure.</p> <p>The human body is full of bacteria, and in fact is estimated to contain more bacterial cells than human cells.</p>
Binary fission in bacteria.	<p>Bacteria reproduce through a process called binary fission.</p> <p>During binary fission, the chromosome copies itself, forming two genetically identical copies.</p> <p>Then, the cell enlarges and divides into two new daughter cells.</p> <p>The two daughter cells are identical to the parent cell.</p>
PROTISTS	
What are protists?	<p>Protists are a group of loosely connected mostly unicellular eukaryotic organisms that are not plants, animals, or fungi.</p> <p>There is no single feature such as evolutionary history or morphology common to all these organisms and they are unofficially placed under a separate kingdom called Protista.</p>
Binary fission in protists.	<p>Protists have the ability to reproduce using binary fission.</p> <p>In binary fission, an organism duplicates its genetic material, or deoxyribonucleic acid, and then divides into two parts known as cytokinesis.</p>
Budding in protists.	<p>Budding occurs when a new organism grows from the body of its parent.</p> <p>They can, however, also reproduce by sexual reproduction.</p> <p>Protists reproduce sexually through the processes of conjugation and alternation of generations.</p> <p>Conjugation is a temporary union of protists in which they exchange genetic material.</p>

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

Features of fertilisation.	Haploid egg and sperm travel in opposite directions and eventually unite in the fallopian tubes whereby fertilisation takes place to produce diploid zygote.
Definitions.	<p>Implantation: When a fertilised egg cell attaches to the wall of the uterus in early pregnancy before it starts developing.</p> <p>Ovulation: Process in which a mature egg is released from the ovary.</p> <p>Fertilisation: Fusion of gametes to give rise to a new individual organism or offspring.</p> <p>Pregnancy: Condition between fertilisation and birth, where egg develops into foetus.</p> <p>Birth: Emergence of a baby or other young from a body of its mother.</p>
What is hormonal control?	<p>Key hormones that regulate pregnancy:</p> <ul style="list-style-type: none"> - hCG - Progesterone - Estrogen

	<ul style="list-style-type: none">- Oxytocin- Prolactin									
What is hormonal control in pregnancy and birth in mammals?	<p>Hormonal control in pregnancy and birth in mammals includes:</p> <ul style="list-style-type: none">- hCG: maintains the corpus luteum (so it can produce progesterone and stop ovulation. Also increases blood supply to the pelvic area.- Progesterone: Prevents lactation and uterine contractions before birth and rises gradually throughout the pregnancy.- Estrogen: Assist in organ development of the foetus and increases gradually throughout the pregnancy.- Oxytocin: Causes the release of milk and stretches the cervix for birth.- Prolactin: Stimulates lactation (milk secretion)									
Stages of development from sex cell to implantation.	<p>Zygote is the first stage after fertilisation, which results in a diploid cell from a haploid egg and sperm.</p> <ul style="list-style-type: none">- Morula: Next stage after zygote undergoes mitosis to become 2 cells. Continue to divide until 16 cells, then enters the uterus.- Blastocyst: Next stage which morula has divided by mitosis into many smaller cells organised into 2 layers. Cells ready to become specialised, stage where implantation occurs into the uterine lining.- Gastrula: Develops from the blastocyst when 3 layers of cells have formed.- Embryo: Further specialisation occurs with tissues and organs developing and functioning. In humans 3-8 weeks after fertilisation, or 5-10 after last period.- Foetus: Final stage of development when all major organs have developed. Last stage until birth.									
What are hormones and where are they produced?	<p>Hormones are chemical messengers released from endocrine glands and circulate in blood. Bind to receptors on target organs and initiate response. Sex hormones regulate reproduction and development of secondary sexual characteristics.</p> <p>Endocrine glands include testes in males, Ovaries in females, pituitary gland or master gland, and hypothalamus which is the control centre to regulate reproductive cycle.</p>									
Hormonal control of reproduction in humans.	<table><tr><th>Hormone</th><th>Where produced</th><th>Function in reproduction</th></tr><tr><td>Testosterone</td><td></td><td></td></tr><tr><td>Oestrogen</td><td></td><td></td></tr></table>	Hormone	Where produced	Function in reproduction	Testosterone			Oestrogen		
Hormone	Where produced	Function in reproduction								
Testosterone										
Oestrogen										

	Progesterone		
	Luteinising hormone (LH)		
	Follicle stimulating hormone (FSH)		
	Oxytocin		
	Prolactin		
	Gonadotropin releasing hormone (GnRH)		
Impact of scientific knowledge on manipulation of plant and animal reproduction.			

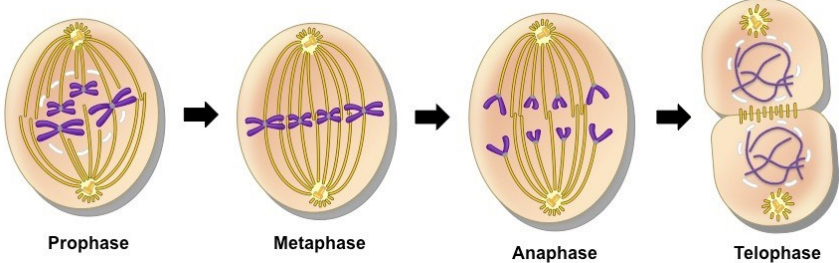
- evaluate the impact of scientific knowledge on the manipulation of plant and animal reproduction in agriculture

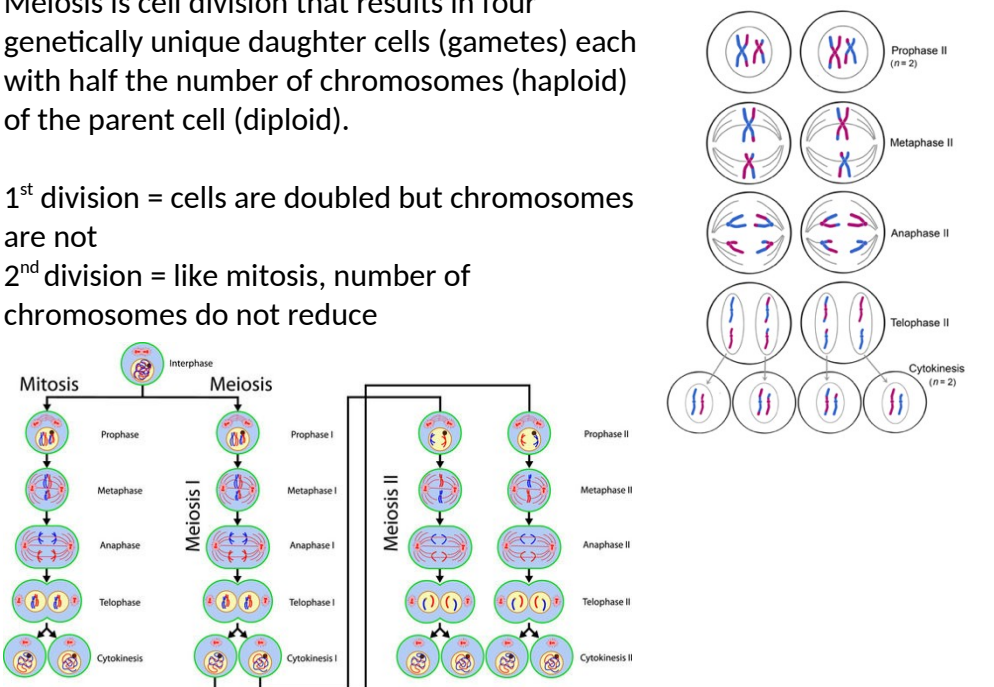
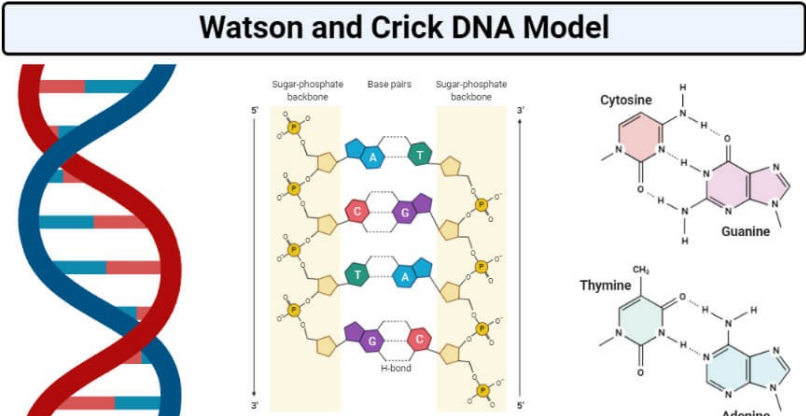
What is agriculture?	Despite differences in the fertilization process, the development of plant and animal embryos is similar. A plant embryo is contained within a seed, which provides the nutrients it needs to grow, while an animal embryo develops within an egg, outside the organism, or within a uterus, inside the female parent organism.
What is selective breeding?	A form of artificial selection imposed by humans, conducting deliberate crosses of living organisms to obtain a combination of desirable offspring.
What is the manipulation of plant reproduction?	Maize: Has been selectively bred from the wild grass teosinte. Modern maize (corn) produces larger cobs with more kernels.
What is the manipulation of animal reproduction?	Dogs – domestic dogs were selectively bred from wolf species. Labradors were bred as soft-mouthed and strong swimmers for duck hunting.
Cloning and transgenic organisms.	Cloning is the process of producing plants or animals that are genetically identical to the parent. Whilst transgenic organisms have a section of DNA from a different individual or species inserted into their genome. Example: Dolly the Sheep.

CELL REPLICATION

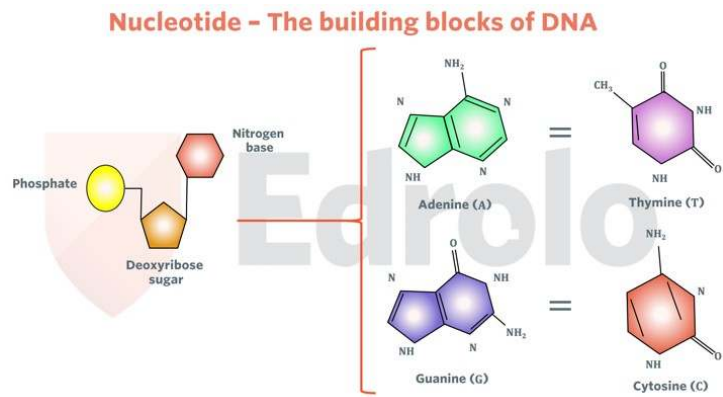
Inquiry question 2: How important is it for genetic material to be replicated exactly?

- 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.

What is mitosis?	<p>Mitosis is cell division that results in two genetically identical daughter cells, each having the same number and kind of chromosomes as the parent nucleus (diploid). Growth, repair, and maintenance.</p> <p>During <u>interphase</u>, the cell grows, and the nuclear DNA is duplicated.</p>  <p>The diagram illustrates the four stages of mitosis in a sequence of four circular cells. 1. Prophase: Chromosomes are condensing and spindle fibers are beginning to form. 2. Metaphase: Chromosomes are aligned in the center of the cell. 3. Anaphase: Sister chromatids are separating and moving to opposite poles. 4. Telophase: Two new nuclei are forming, and the cell is beginning to divide. Arrows indicate the progression from Prophase to Metaphase, then to Anaphase, and finally to Telophase.</p> <p><u>Prophase</u>: Prophase is the first phase of mitosis, the process that separates the duplicated genetic material carried in the nucleus of a parent cell into two identical daughter cells. During prophase, the complex of DNA and proteins contained in the nucleus, known as chromatin, condenses.</p> <p><u>Metaphase</u>: Metaphase is a stage during the process of cell division (mitosis or meiosis). Normally, individual chromosomes are spread out in the cell nucleus. During metaphase, the nucleus dissolves and the cell's chromosomes condense and move together, aligning in the centre of the dividing cell.</p> <p><u>Anaphase</u>: Anaphase is the fourth phase of mitosis, the process that separates the duplicated genetic material carried in the nucleus of a parent cell into two identical daughter cells.</p> <p><u>Telophase</u>: Telophase is the fifth and final phase of mitosis, the process that separates the duplicated genetic material carried in the nucleus of a parent cell into two identical daughter cells. Telophase begins once the replicated, paired chromosomes have been separated and pulled to opposite sides, or poles, of the cell.</p>
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<p>What is meiosis?</p>	<p>Meiosis is cell division that results in four genetically unique daughter cells (gametes) each with half the number of chromosomes (haploid) of the parent cell (diploid).</p> <p>1st division = cells are doubled but chromosomes are not 2nd division = like mitosis, number of chromosomes do not reduce</p> 
<p>What is DNA replication?</p>	<p>DNA replication is the process by which the double stranded DNA molecule, is 'unzipped' by enzymes, and free nucleotides bind to each of the exposed backbones to create two identical DNA molecules</p>
<p>How does DNA replication occur?</p>	<p>Replication occurs in three major steps: the opening of the double helix and separation of the DNA strands, the priming of the template strand, and the assembly of the new DNA segment. During separation, the two strands of the DNA double helix uncoil at a specific location called the origin.</p>
<p>Watson and Crick DNA model.</p>	<div data-bbox="432 1198 1241 1612"> <p style="text-align: center;">Watson and Crick DNA Model</p>  </div>

Nucleotide composition.



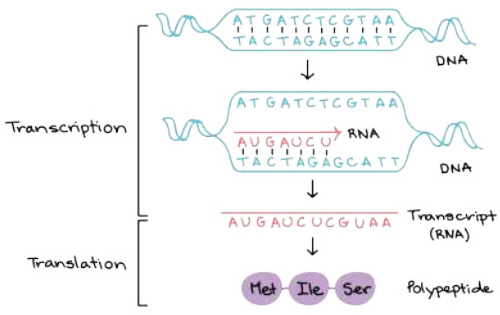
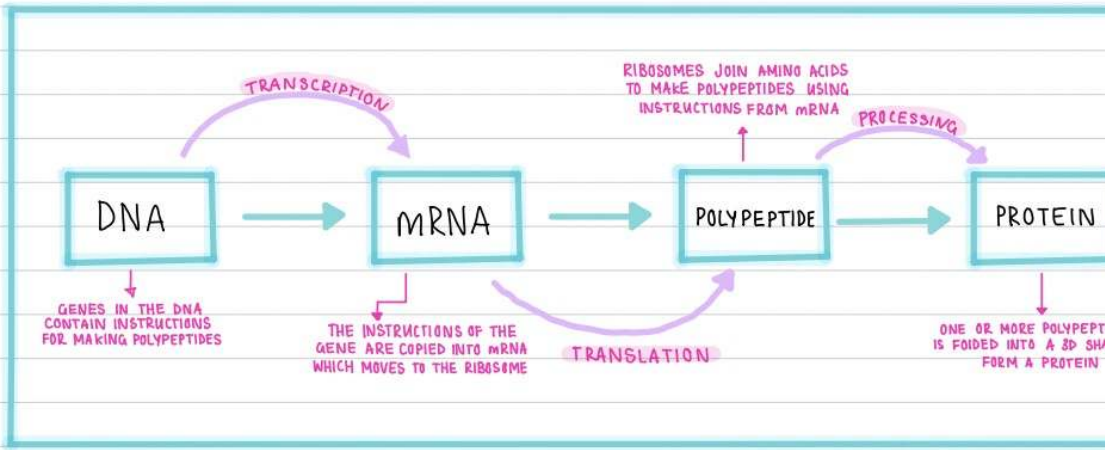
- assess the effect of the cell replication processes on the continuity of species.
- Helps in formation of haploid gametes for carrying out sexual reproduction. Plays a vital role in maintaining ideal number of chromosomes in organisms. Introduces new characteristics in organisms as a result of recombination due to crossing over.

DNA AND POLYPEPTIDE SYNTHESIS

Inquiry question 3: Why is polypeptide synthesis important?

- construct appropriate representations to model and compare the forms in which DNA exists in eukaryotes and prokaryotes.
- 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

DNA Diagram	<p>DNA is double stranded. Genetic material found in nucleus. Has code to make amino acids, cannot leave nucleus. Code has to go to cytoplasm. Ribosomes are protein making factory. RNA transports DNA.</p> <p>Antiparallel DNA Strands DNA Ladder Double Helix</p>
RNA	RNA has Adenine, Guanine, Cytosine, Uracil and NOT Thymine. Uracil is one of four nucleotide bases in RNA. In RNA, uracil pairs with adenine.
mRNA	A type of RNA found in cells. mRNA molecules carry the genetic information needed to make proteins. They carry the information from the DNA in the nucleus of the cell to the cytoplasm where the proteins are made. Also called messenger RNA.
tRNA	Transfer RNA (abbreviated tRNA) is a small RNA molecule that plays a key role in protein synthesis. Transfer RNA serves as a link (or adaptor) between the messenger RNA (mRNA) molecule and the growing chain of amino acids that make up a protein.
Protein synthesis	Protein synthesis is the process in which cells make proteins. It occurs in two stages: transcription and translation.
Transcription	Transcription, as related to genomics, is the process of making an RNA copy of a gene's DNA sequence. This copy, called messenger RNA (mRNA), carries the gene's protein information encoded in DNA.
Translation	DNA translation is the term used to describe the process of protein synthesis by ribosomes in the cytoplasm or endoplasmic reticulum. The genetic information in DNA is used as a basis to create messenger RNA (mRNA) by transcription.

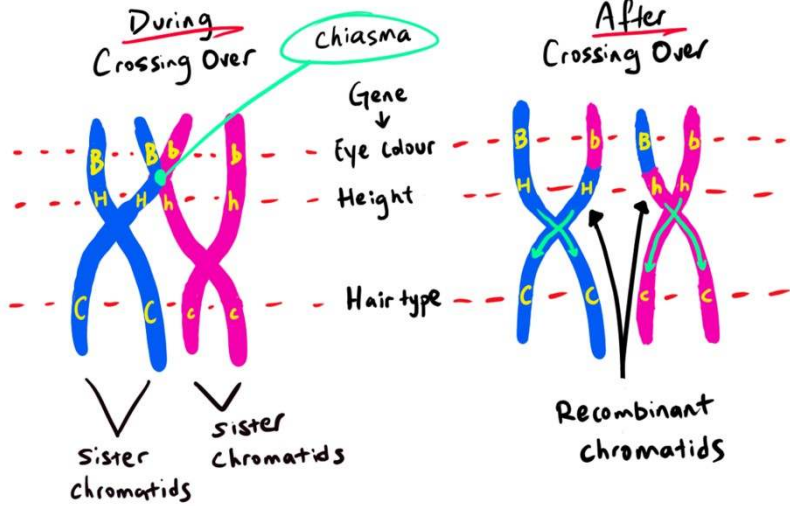
	<p>Single stranded mRNA then acts as a template during translation.</p> 
What is a peptide?	<p>Peptides are short chains of amino acids that act as building blocks of proteins such as collagen, elastin, and keratin.</p> <p>These proteins are the foundations of your skin and are responsible for its texture, strength, and resilience.</p>
Polypeptide synthesis.	<p>Polypeptide synthesis is the biological production of peptides, which are organic compounds in which multiple amino acids are linked via peptide bonds.</p> <p>They are the building blocks of proteins, which are essential to cell function.</p> 
Phenotypic expression	<p>Phenotypic expression is the result of a complex interplay between an organism's genes and its environment.</p> <p>During growth and development, organisms undergo a programmed series of phenotypic changes.</p> <p>Phenotypic expression thus varies throughout growth and development, even when the environment is homogenous and static.</p>
How genes and environment affect phenotypic expression.	<p>The term "phenotype" refers to the observable physical properties of an organism; these include the organism's appearance, development, and behaviour.</p> <p>An organism's phenotype is determined by its genotype, which is the set of genes the organism carries, as well as by environmental influences upon these genes.</p>



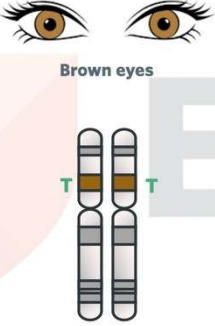
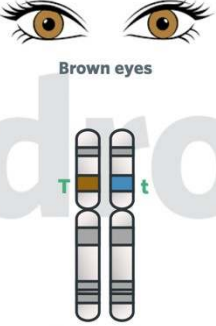
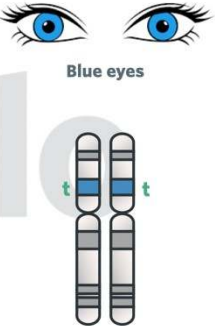
- investigate the structure and function of proteins in living things.

GENETIC VARIATION

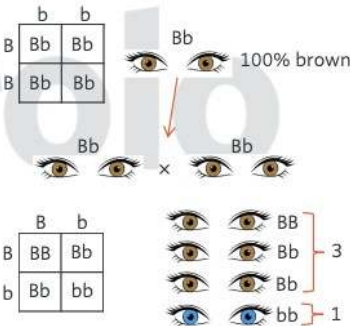


Inquiry question 4: How can the genetic similarities and differences within and between species be compared?

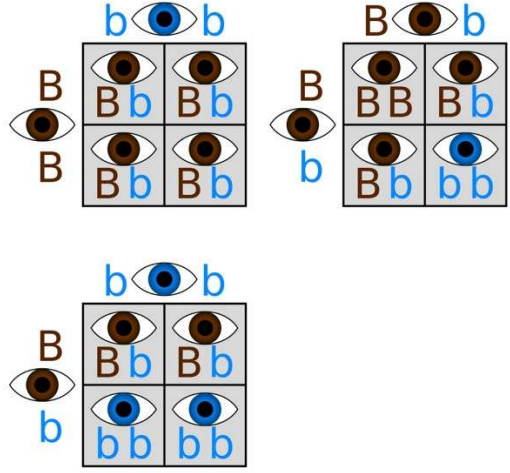
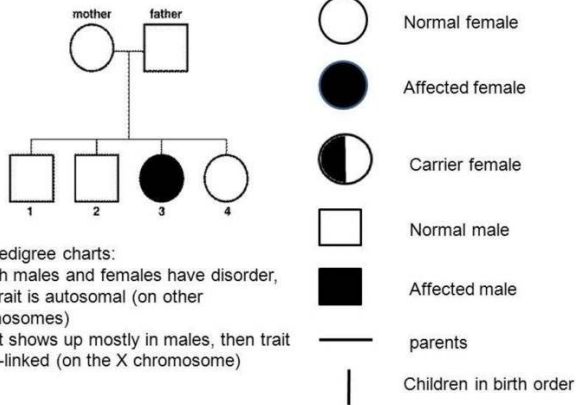
- conduct practical investigations to predict variations in the genotype of offspring by modelling meiosis, including the crossing over of homologous chromosomes, fertilisation, and mutations.

What is a genotype?	The entire set of genes in the cells of an organism. In a narrower sense, however, it can refer to different alleles, or variant forms of a gene, for particular traits, or characteristics.
Crossing over	<p>Crossing over is the process involving the exchange of corresponding gene segments of non-sister chromatids between homologous chromosome pairs (double-stranded chromosome pairs for the most organisms). This effectively creates new allele combinations, known as recombination.</p> 
Homologous Chromosomes	Two chromosomes in a pair – normally one inherited from the mother and one from the father.
Heterozygous	Individual organisms bearing different alleles (Rr) are known as heterozygous.
Homozygous	Individuals carrying two identical alleles (RR or rr) are known as homozygous.
Variation	Genetic variation is the difference in DNA sequences between individuals within a population. Variation occurs in germ cells i.e., sperm and egg, and also in somatic (all other) cells.
Gene	A section of DNA that codes for a particular characteristic. E.g., Flower colour

Allele	<p>An alternative form of a gene. E.g., Purple, or white</p> <p>Brown and blue eyes</p> <p>Key:  Allele for dominant allele  Allele for recessive allele</p> <p>Phenotype</p> <p>Genotype</p> <p>Homozygous dominant </p> <p>Heterozygous </p> <p>Homozygous recessive </p>
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- model the formation of new combinations of genotypes produced during meiosis, including but not limited to:
 - interpreting examples of autosomal, sex-linkage, co-dominance, incomplete dominance, and multiple alleles.
 - constructing and interpreting information and data from pedigrees and Punnett squares.

Autosomal VS Dominant	<p>Autosomal means that the gene in question is located on one of the numbered, or non-sex, chromosomes. Dominant means that a single copy of the mutated gene (from one parent) is enough to cause the disorder.</p> <p>Eye colour in humans.</p> <p>BB x bb</p> <p></p>
Sex-linkage	<p>The human species has a diploid number of 46 chromosomes. Of these 46 (23 pairs) chromosomes, 44 (22 pairs) of these are called autosomes and 2 (1 pair) are called the sex chromosomes (aka allosomes). There are two possible sex chromosomes in humans: X and Y. The Y chromosome is a smaller version of the X chromosome - it has some genes specific for male development, but not much more. The X chromosome has many more genes for various other biological functions (nervous system development, platelets and blood clotting, colour</p> <p>Affected father (XY) x Unaffected mother (XX)</p> <p>X-linked, dominant allele (red dot on X) vs Recessive allele (blue dot on X)</p> <p>Legend:  Affected  Unaffected</p> <p>Offspring: Unaffected son (XY), Affected daughter (XX), Unaffected son (XY), Affected daughter (XX)</p>

	vision) as well as some sex-related genes.
Co-dominance	Codominance essentially means that no allele can block or mask the expression of the other allele.
Incomplete dominance	Incomplete dominance is a condition in which a dominant allele does not completely mask the effects of a recessive allele.
Genotype vs. Phenotype	A person's genotype is their unique sequence of DNA. More specifically, this term is used to refer to the two alleles a person has inherited for a particular gene. Phenotype is the detectable expression of this genotype – a patient's clinical presentation.
Punnett squares	
Pedigrees	<p style="text-align: center;">Pedigree Chart</p>  <p>Reading pedigree charts:</p> <ul style="list-style-type: none"> • If both males and females have disorder, then trait is autosomal (on other chromosomes) • If trait shows up mostly in males, then trait is sex-linked (on the X chromosome)

- 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)

Population genetics	Population genetics is a sub-field of evolutionary biology that deals with the study of allele frequencies in populations and how these change overtime. It aims to explain the phenomena of adaptation, evolution, and speciation.
Polymorphism	Polymorphism refers to a discontinuous genetic variation resulting in the occurrence of several different forms or types of individuals among the members of a single species.

Gene pool	Consists of all genes, including all the different alleles, that are present in a population.
Phenotype frequency	The proportion of occurrences of a phenotype per generation. E.g., 40% of Australian have O+ blood, about 2% of the world's population have green eyes.
Genotype frequency	The proportion of occurrences of a genotype per generation. E.g., FF, Ff and ff, genotype frequency is
Allele frequency	How to calculate allele frequency: $\frac{\text{Number of copies of allele (D) in the population}}{\text{Total number of copies of the gene (D + d) in the population}}$

INHERITANCE PATTERNS IN A POPULATION

Inquiry question 5: Can population genetic patterns be predicted with any accuracy?

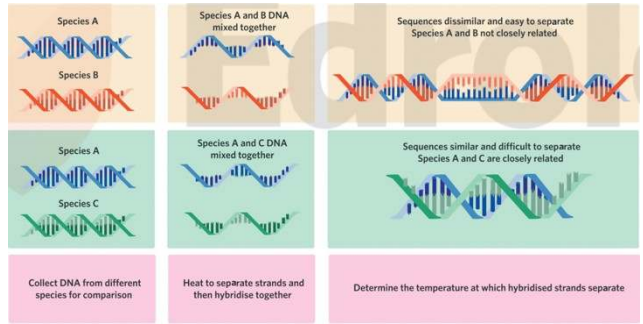
- investigate the use of technologies to determine inheritance patterns in a population using, for example:
 - DNA sequencing and profiling.

DNA sequencing	DNA sequencing is the process of determining the precise order of nucleotides within a segment of DNA. It includes any method or technology used to determine the specific order of adenine, guanine, cytosine, and thymine in a strand of DNA.
DNA profiling	<p>DNA profiling involves the testing of highly variable regions of an individual's DNA that contain short repeating sequences called Short Tandem Repeats (STRs), located in the introns (non-coding regions). The exact number of STRs varies from person to person, because we inherit our DNA from our parents. DNA profiles can be used to:</p> <ul style="list-style-type: none"> - confirm how closely related individuals are. - trace inheritance patterns. - solve crimes.
Polymerase Chain reaction (PCR)	<p>PCR is a technique used to exponentially amplify large numbers of copies of a specific sequence of DNA. The central paradigm to PCR is understanding that all life contains genetic material that is unique to a species, and individuals within a species contain unique genetic sequences. As a result, PCR can be used to amplify unique DNA sequences of any organism which can then be compared to other nucleotide segments from a known source (control or standard).</p> <p>PCR uses:</p> <ul style="list-style-type: none"> - Genetic testing - Medical diagnosis - DNA fingerprinting - Cloning - Evolutionary study
Process of PCR	<p>A PCR machine uses variations in temperature to control the replication process via three steps:</p> <ul style="list-style-type: none"> - Denaturation – DNA sample is heated to separate it into two single strands (95°C for 1 min) - Annealing – DNA primers attach to the 3' ends of the target sequence (55°C for 1 min) - 3. Elongation – A heat-tolerant DNA polymerase binds to the primer and copies the strand of DNA (72°C for 2 min)
Gel Electrophoresis	<p>Gel electrophoresis is a lab technique used to separate mixtures of DNA based on molecular size. The molecules (which contain a charge) are pushed by an electrical field through a gel. The smaller the molecule, the faster and further they move.</p> <p>Gel electrophoresis Process:</p>

	<ul style="list-style-type: none"> - DNA is extracted. - PCR isolation and amplification of DNA. - DNA added to the gel wells. - Electric current applied to the gel. - DNA bands are separated by size. - DNA bands are stained.
Summary	<p>Genetic sequencing is the process by which the exact sequence of nucleotides in a chromosome or gene is determined. Special versions of the four nucleotides, called chain-terminating, nucleotides (ddATP, ddTTP, ddCTP, ddGTP), are each labelled with a different coloured fluorescent marker.</p> <p>Genetic sequencing using PCR and gel electrophoresis During the PCR cycles different length DNA fragments will form due to the chain-terminating nucleotides stopping further sequencing.</p>
Karyotype	The number and visual appearance of the chromosomes in the cell nuclei of an organism or species.
Karyotyping	Standard karyotypes can be used to detect abnormalities in whole chromosome numbers or structural issues e.g., Down syndrome (trisomy: C21), Klinefelter syndrome (trisomy: XXY). Typically requires dividing white blood cells from the patient. Chromosomes are stained, counted, sorted into homologous chromosomes, and analysed. Requires light microscope at magnification of 1000-1500x.

- 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 Genetics	<p>Conservation genetics is the application of genetics to preserve species through maintaining variation within populations so that they are capable of coping with environmental change.</p> <p>Scientists analyse alleles of multiple genes to examine the genetic diversity within a species.</p> <p>When the significant majority of individuals have the same alleles, the population has low genetic diversity. In contrast, when there are multiple alleles of multiple genes within a population, it has high genetic diversity. The organisms that conservation geneticists' study are typically endangered or threatened due to habitat destruction, environmental change and change in population size</p>
Minimum Viable Population	Minimum viable population (MVP) is the ecological threshold that indicates the smallest number of individuals in a species or population capable of surviving in the wild for an ongoing amount of time.

	<p>Small populations are more susceptible to genetic drift than large populations. This occurs when differential breeding leads to the loss of alleles from the gene pool due to the small number of individuals carrying particular alleles. Genetic drift can cause a problem as it occurs through chance, unlike natural selection, where individuals with favourable alleles survive and pass on these desirable traits.</p>
Human Genome Project (HGP)	<p>The Human Genome Project (HGP) was an international effort to decode the entire sequence of the human genome. It was completed in 2003. A complete and accurate sequence of 3 billion DNA base pairs. An estimated 20,000 to 25,000 genes have been identified.</p>
Single nucleotide polymorphism (SNP)	<p>A point mutation in a segment of DNA that occurs in more than 1% of a population.</p>
Haplotype	<p>A group of genes that are inherited together from a single parent. The word is derived from the word's haploid and genotype.</p>
Genome-wide association studies (GWAS)	<p>Multiple GWAS success stories have involved international collaborations utilising results of the HGP, tag-SNPs and the HapMap to share large scale genotyping data.</p> <p>Examples</p> <ul style="list-style-type: none"> - Age-related macular degeneration (blindness) is linked to gene involved in regulating inflammation. - Prostate cancer (PrCa) is the most common cancer in men. 170 common genetic variants have been linked to PrCa. - Parkinson's disease is a degenerative neurological condition that affects the control of body movements. Recent GWAS discovered 35 genes with links to the disease. - Also uncovered genes associated with type 2 diabetes, cardiovascular and Crohn's disease. <p>These gene associations will now be used to in screening, tailored treatments and possible cures.</p>
Population Genetics	<p>The study of genetic composition of populations.</p>
DNA hybridisation	<p>DNA hybridization is a technique that separates the double-stranded DNA of two species into single strands. The single strands from each species are mixed and allowed to bind. They are then re-heated, and the temperature of separation is recorded. The more closely related the species, the stronger the hybridization, the higher the temperature required to separate the hybrid DNA = a more recent common ancestor.</p>  <p>The diagram illustrates the DNA hybridization process in six panels. The top row shows Species A (blue) and Species B (red) DNA being collected, mixed together, and then separated into single strands. The bottom row shows Species A (blue) and Species C (green) DNA being collected, mixed together, and then separated into single strands. The text indicates that Species A and B DNA are 'Sequences dissimilar and easy to separate' (Species A and B not closely related), while Species A and C DNA are 'Sequences similar and difficult to separate' (Species A and C are closely related). The process steps are: 'Collect DNA from different species for comparison', 'Heat to separate strands and then hybridise together', and 'Determine the temperature at which hybridised strands separate'.</p>
Comparative Genomics	<p>Comparative genomics uses computer-based analysis to compare the genome sequences of different species. There are over 1,300 species sequenced including humans (HGP), chimpanzee, rhesus monkey, mouse, rat, dog, E. coli, cow, fruit fly. Results are used to study:</p>

	<ul style="list-style-type: none"> - Evolutionary relationships. - Conserved DNA sequences which indicate genes that are essential to life. <p>Genomic similarities and differences provide scientists with a better understanding of how the appearance and behaviour of organisms has changed over time.</p>
Summary	<p>Technologies including PCR and gel electrophoresis are used in the Sanger method to sequence DNA.</p> <p>Once sequenced, DNA profiling can be used for forensics, to test for genetic diseases and in paternity tests.</p> <p>Conservation genetics is the field of science that aims to understand the dynamics of genes in a population to avoid falling below a MVP.</p> <p>The Human Genome Project (HGP) completely sequenced the DNA profile of humans.</p> <p>GWAS use results of the HGP to compare 'normal' groups with groups with specific diseases to isolate the gene/s responsible.</p> <p>Comparative genomics uses computer-based analysis to compare the genome sequence of different species and determine evolutionary relationships.</p>