

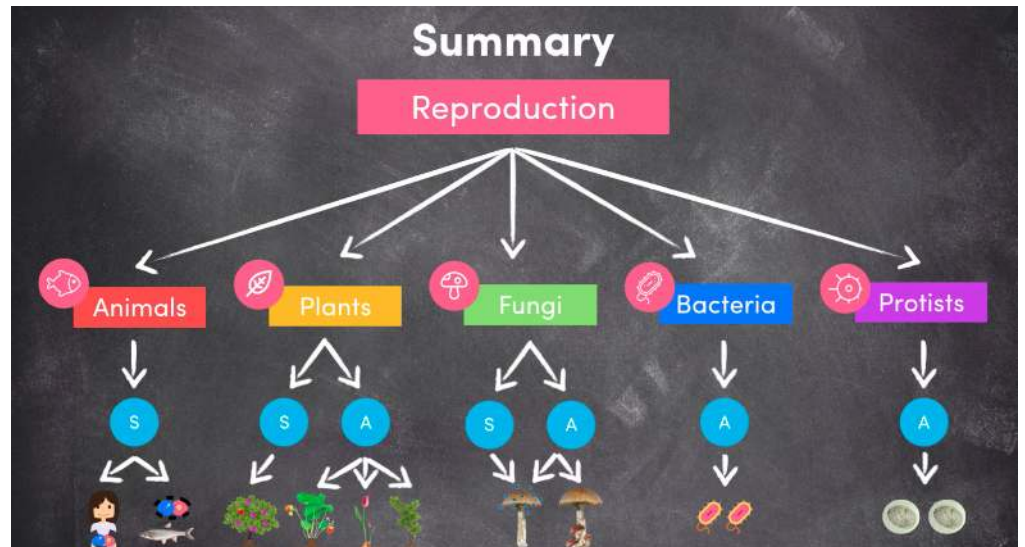
MODULE 5: HEREDITY

Content: Reproduction

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

1.1 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:

(a) animals: advantages of external and internal fertilisation



There are 2 ways that an animal can reproduce sexually, either internal or external

Internal

Egg is fertilised within the female reproductive tracts

External

The egg is fertilised outside of the body

Both male and female release their sex cells directly into the surrounding environment for fertilisation to take place . EG spawning from fish.

(b) plants: asexual and sexual reproduction

Asexual reproduction

- Bulbs
- Runners
- Cuttings

Runners

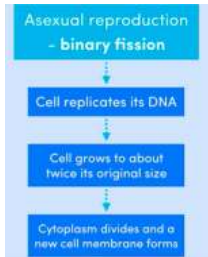
Runners are side branches with little clumps of leaves and roots, called plantlets, distributed along them. These runners grow along the ground , and the roots then dig down and establish the plantlets as individual plants. Eg strawberry

Bulbs

Growth of bulbs underground. From these bulbs, little buds form and grow to form a new plant , eg. daffodils

Cuttings

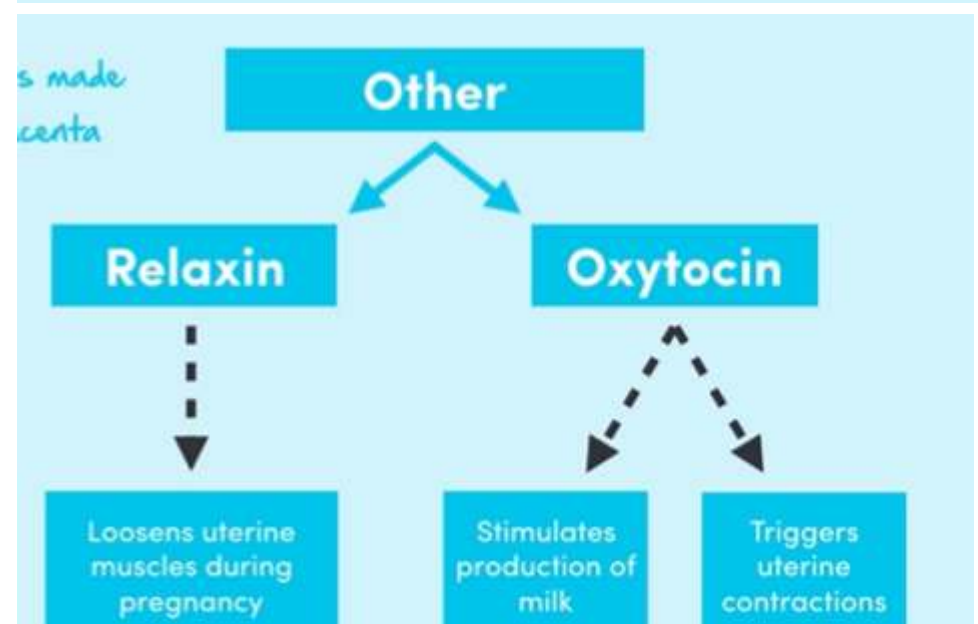
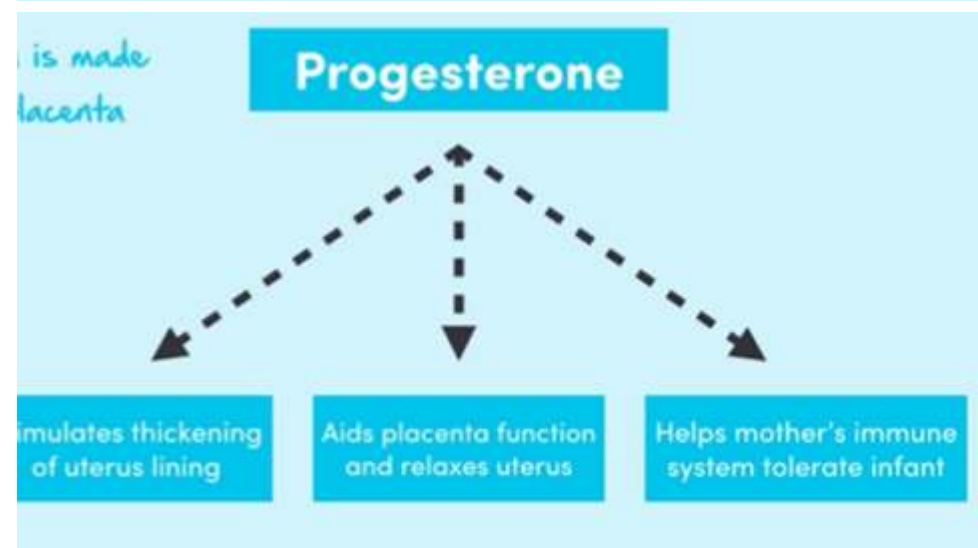
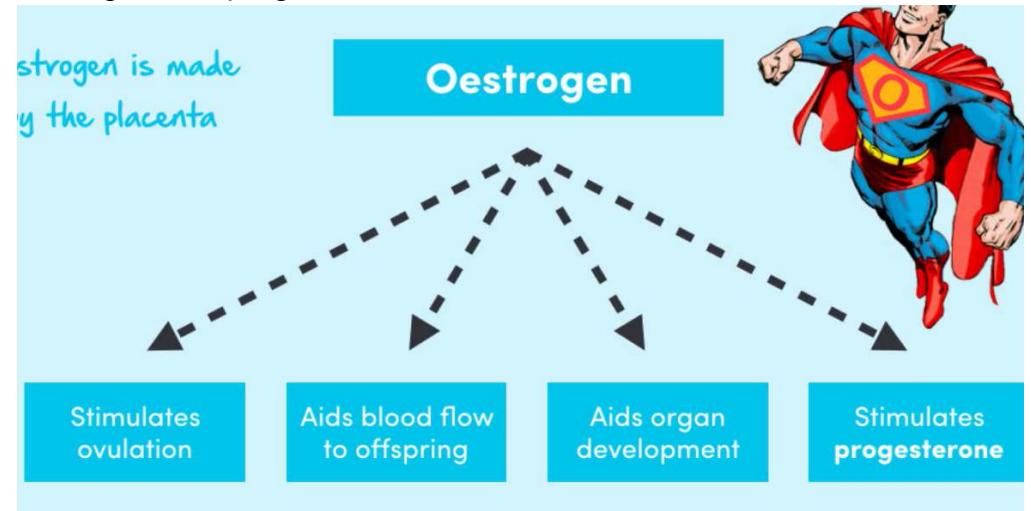
The process of taking cuttings is referred to as vegetative propagation. This is where a branch from the parent plant is cut off, stripped of its lower leaves and planted into the ground , where it will grow and establish itself as a new plant.

	<p><u>Sexual reproduction</u></p> <p>Involves a process called pollination. Pollination refers to the transfer of pollen from one plant to another by insect birds, or wind to one plant to another. Deep inside the flowers are the ovules, the female plant sex cells</p> <ul style="list-style-type: none"> - After fertilisation of the flower develop into fruit, and the ovules turn into seeds
(c) fungi: budding, spores	<p>Fungi is a eukaryotic organisms that can be either unicellular or multicellular , such as mushrooms”</p> <p><u>Asexual</u></p> <p>When condition are good the fungus will reproduce asexually and spread quickly</p> <p>This is done via budding, where the fungus develops a groth and then breaks off to form a new organism or Spores which are released directly into the surrounding environment to spread and grow elsewhere</p> <p><u>Sexual</u></p> <p>When conditions are bad, fungi can use these spores to reproduce sexually, as the spores from two different fungi fuse together to form a single new cell , which will develop to grow into a new fungus. (increasing genetic variation)</p>
(d) bacteria: binary fission	<p>A bacteria are prokaryotic single celled organisms such as E- coli</p> <p><u>Asexual</u></p> <p>Most bacteria reproduce asexually in a process called Binary fission</p>  <pre> graph TD A["Asexual reproduction - binary fission"] --> B["Cell replicates its DNA"] B --> C["Cell grows to about twice its original size"] C --> D["Cytoplasm divides and a new cell membrane forms"] </pre>
(e) protists: binary fission, budding	<p>The protist are such a mixed bag that some can reproduces by binary fission and budding, but even reproduce sexually</p>
1.2 analyse the features of fertilisation, implantation and hormonal control of pregnancy and birth in mammals	<p>FERTILISATION</p> <p>The fusion of and egg and sperm</p> <p>IMPLANTATION</p> <p>The attachments of a fertilised egg to the uterine lining in early pregnancy.</p> <p>After the blastocyst is implanted it becomes an embryo and then a</p>

foetus when it has formed all the basic adult features.

HORMONES

Hormones are chemical messengers produced by the body, which travel in the blood to other cells where they have a specific effect. Hormones that have a significant role during pregnancy include oestrogen and progesterone,



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

- Agriculture is the growth of crops and animals for human needs
- Reproduction can be manipulated using natural breeding, artificial insemination, artificial pollination and cloning
- Overall the impact of manipulating reproduction in agriculture is huge and positive

Content: Cell Replication

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

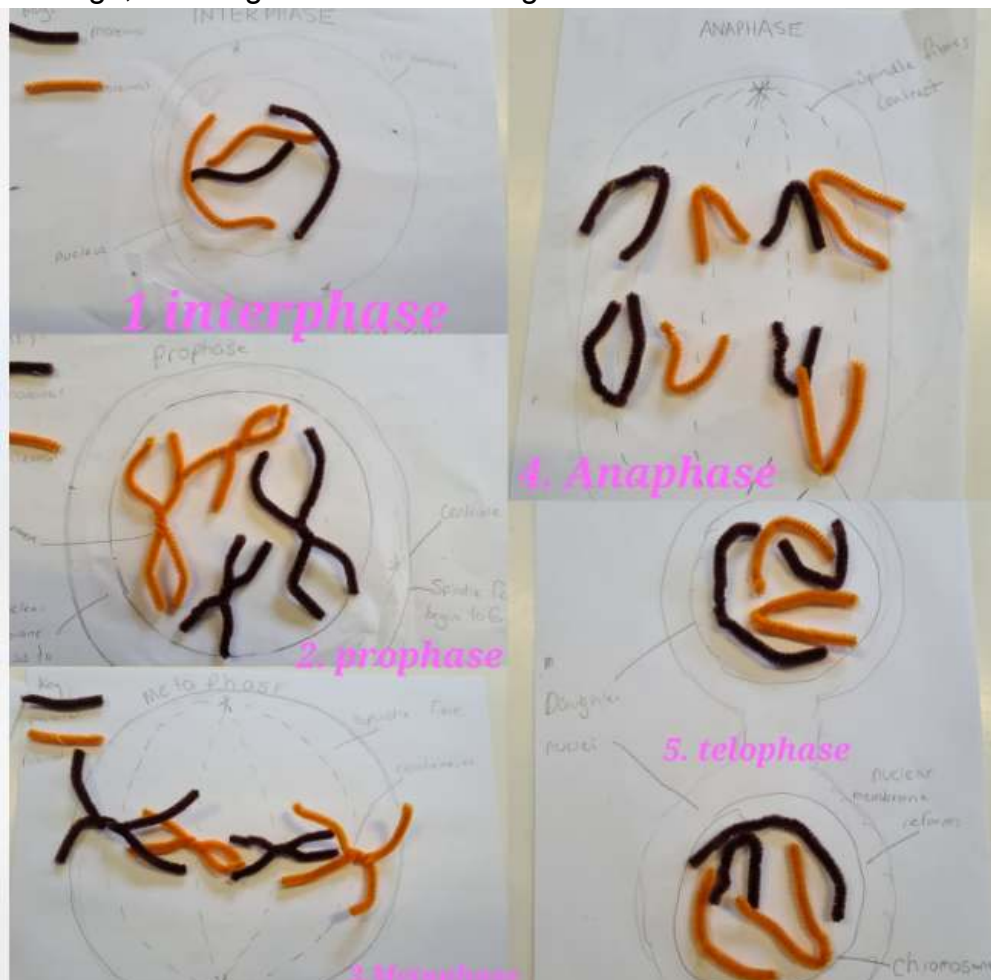
2.1 model the

processes involved in cell replication, including but not limited to:

(a) mitosis and meiosis

MITOSIS

Mitosis function in an organism is the growth and repair of cells. The process of mitosis starts with interphases, where DNA replication occurs. The DNA is currently chromatin and aren't fully formed chromosomes. The next stage in mitosis is Prophase where the Chromatin thickens and becomes Chromosomes, with each sister chromatid containing 2 copies of the DNA. In this stage, the nuclear membrane starts to break down and the spindle fibre and centrioles start to form. In Metaphase, the chromosomes line horizontally along the equator, each of the centromeres connected to a spindle fibre. The Spindle fibres start to retract in **Anaphase**, separating the chromatids, pulling them to the poles of the cell. In **Telophase**, the nuclear division is complete, so the nuclear membrane reform around the two identical nuclei. Resulting in cleavage connecting the two nuclei together. **Cytokinesis** then breaks down that cytoplasmic cleavage, resulting in 2 identical daughter cells.




MEIOSIS



Meiosis 1

Prophase is where the is where chromatin thickens to become x like chromosomes. However, crossing over occurs between homologous chromosomes, transferring genetic information to the other chromosome within the pair. Metaphase is where the chromosomes line up in their homologous pairs, they are independently assorted to other homologous pairs. Anaphases is where the homologous pairs are separated by spindle fibres to the opposite poles of the cell. Telophase is where the two cells start to form creating a cytoplasmic cleavage. Cytokinesis is where the 2 cells separate. Due to Random

	<p>segregation of the chromosomes, this makes the two cells identical compared to the parent cell. These 2 cells are haploid</p> <p>Meiosis 2</p> <p>This starts in metaphase, where the chromosomes in the two cells line up in the centre of the cell. During anaphase, the sister chromatids are pulled to the opposite poles of the cell via spindle fibres. In telophase II the 4 cells start to become visible, but are connected with cleavage. Cytokinesis breaks the cleavage producing 4 identical haploid cells.</p> <p><u>Limitations</u></p> <ul style="list-style-type: none"> - The model is not to scale, affecting the effectiveness of the model as it does not provide an understanding of the actual size of the cells and chromosomes - The model is static and not dynamic, causing the model to lack the ability to show the speed of the process and how long the cell spends in each stage, ultimately decreasing the effectiveness of this model. <p><u>Advantages</u></p> <ul style="list-style-type: none"> - The model effectively simplified the complex process of meiosis, increasing the ability to understand, through the usage of simple drawings with descriptive labels, - By using different coloured pipe cleaners, along with a key, it allowed for a clear understanding of differentiation of paternal and maternal chromosomes. Allowing for a simple understanding of what each chromosome is doing in each stage
<p>(b) DNA replication using the Watson and Crick DNA model, including nucleotide composition, pairing and bonding</p>	<p><u>DNA Replication</u></p> <p>At the beginning of DNA replication, an enzyme called helicase temporarily unwinds the two strands of DNA, creating a replication fork. One new strand is called the leading strand. It is made as one continuous piece. The other strand is made in the opposite direction and is made of multiple pieces that get joined together. It is called the lagging strand. DNA Polymerase assists the process of DNA nucleotides being added to the new strands. The result is in 2 identical DNA molecules. Multiple other enzymes and proteins assist the process of DNA replication.</p> <ul style="list-style-type: none"> - The conservative strand from the original DNA strand is used as a template for the correct complementary bases to attach to.

	<p>The model of DNA replication above shows an accurate visualisation of how DNA replicates itself during Cell division to form 2 identical strands.</p> <p><u>Strengths</u></p> <p>Each different loolie symbolised a component of the DNA structure and the process and remained consistent throughout the diagram, making it reliable. It's a simplified and understandable depiction of the complex, microscopic process.</p> <p><u>Limitations</u></p> <ul style="list-style-type: none"> - The model was static, did not show speed of process - not to scale and no order of the steps
<p>2.2 assess the effect of the cell replication processes on the continuity of species</p>	 <p>Genetic continuity referred to the ongoing survival of species as a result of characteristics being passed from parents</p> <ul style="list-style-type: none"> - It is related to passing consistently accurate genetic information. <p>It is achieved through:</p> <p>DNA Replication: exact replication of DNA is the basic key to genetic continuity</p> <p>Mitosis: Identical 2 daughter cells, each containing the same number as the parent cell (diploid)</p> <p>Meiosis:- Each offspring has all the genes it needs to survive</p> <ul style="list-style-type: none"> - Offspring will be able to mate with other organisms. - Wrong number of chromosomes= unable to mate

Content: DNA and Polypeptide Synthesis

3. *Inquiry question:* Why is polypeptide synthesis important?

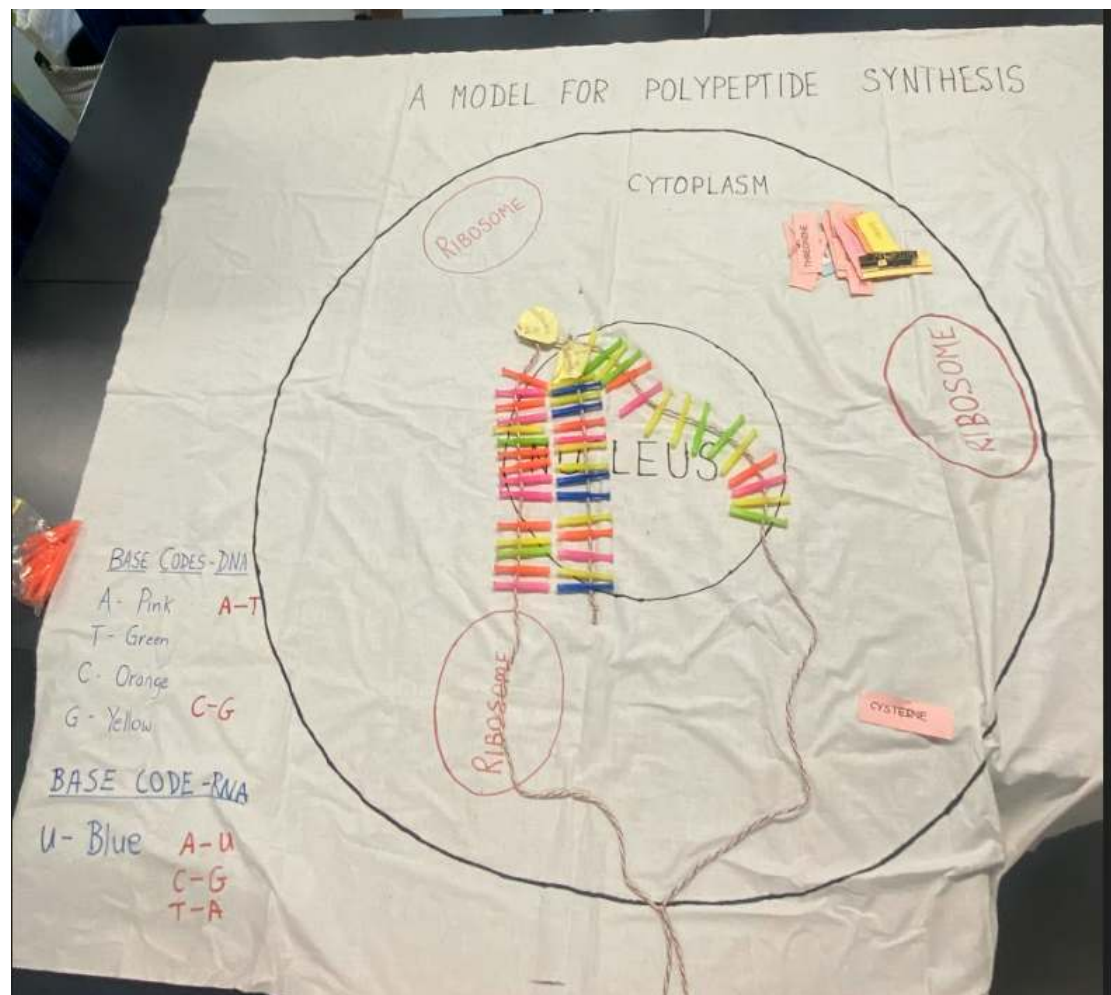
3.1 construct appropriate representations to model and compare the forms in which DNA exists in eukaryotes and prokaryotes

	prokaryote	Eukaryote
Examples	Bacteria and archaea	Animal, plant, fungi
Does this type of cell have DNA?	Yes	yes
Does DNA form chromosomes?	yes	yes
What is the shape of chromosomes?	circular	linear
How many chromosomes?	1	multiple
Where is most of the DNA?	Nucleoid region	nucleus
Where else can DNA be found?	plasmids	Mitochondria, chloroplast

3.2 model the process of polypeptide synthesis, including:

(a) transcription and translation

TRANSCRIPTION



Limitations

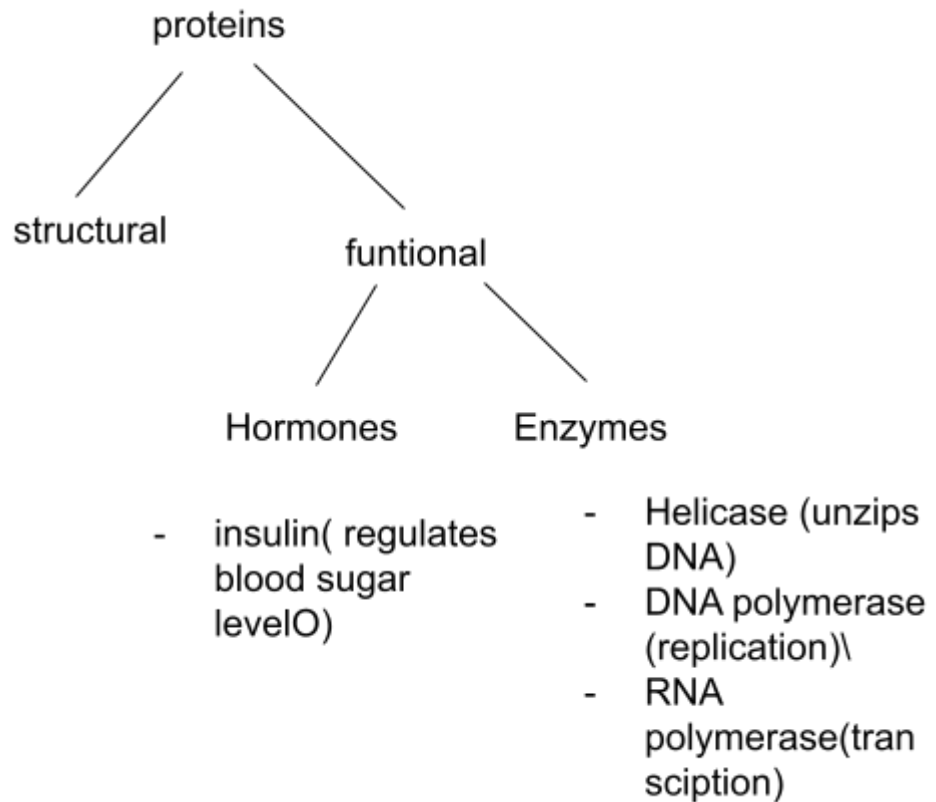
- The model is not to scale, which limits the understanding of the size of the process and all its components
- The model is static- not dynamic, didn't show the component of mRNA to ribosomes along with the length of time it takes for each stage
- The model did not start with a start codon or end with a stop codon which can lead to an inaccurate misconception of how translation occurs.

Advantages

	<ul style="list-style-type: none"> - It simplified the complex stages of transcription and translation through labels - The process of polypeptide synthesis was further simplified through the key and the usages of brightly different coloured pegs, allowing the distinct differentiation between the bases, names of bases and the complementary bases, <p>Having the key it allows for the easy creation of codons and anticodon as the differentiation of the nitrogenous bases were clearly visible and labels</p>
(b) assessing the importance of mRNA and tRNA in transcription and translation	<p><i>The importance of mRNA</i></p> <p>MRNA is important as it transcribes the genetic instructions from the genes on DNA making polypeptides. The mRNA is mobile, it then takes the instructions to the ribosomes where they are translated into proteins. Due to DNA being permanently stationary and unable to leave the nucleus, mRNA is the only way the instructions can be taken to the ribosome. Without mRNA polypeptide synthesis would not occur, leaving the body without fundamental, structural or functional proteins.</p> <p><i>The importance of tRNA</i></p> <p>tRNA is important as it carries a triplet basis complementary to the codons on mRNA, called anticodons, as well as the corresponding amino acid. As the tRNA anticodon binds with its complementary pair of mRNA codons, it ensures the correct sequence on the polypeptide chain. This allows it to create the correct protein needed</p>
(c) analysing the function and importance of polypeptide synthesis	
(d) assessing how genes and environment affect phenotypic expression	<p>The term phenotype in this context includes the structure, behaviour and physiology of an organism.</p> <p>Many variations arise as a result of an interaction between the two – the environment can influence how genes are expressed..</p> <p>although genes may be direct determinants of phenotype, gene expression can be enhanced or masked by factors in the environment.</p> <p>Some results from the study of epigenetics suggest that the environment may chemically modify DNA in individuals and in this way affect gene expression. This chemical modification is not a change in the sequence of bases in the genome (as in mutations), but instead seems to involve chemical markers or tags being added to DNA.</p> <p>E.G Both genes AND the environment affect an organism's phenotype.</p> <p>e.g. A human can't grow to their full height if they are undernourished.</p>

e.g. hydrangeas produce blue flowers in acidic soil, and pink flowers in alkaline soil

3.3 investigate the structure and function of proteins in living things/



Protein structure

- Large complex macromolecules made of one or more polypeptide chains
- If primary stage (twisted) amino acids are joined by peptide bonds)

PROTEIN FUNCTION

Proteins carry out most of the functions that are essential for life.

- Enzymes are all protein molecules.
- Structural Molecules, such as in muscle fibres, skin, hair and bone matrix are proteins. Eg. collagen
- Many “Special Molecules” are proteins e.g.
 - haemoglobin (the oxygen carrier in blood)
 - chlorophyll (absorbs light for photosynthesis)
 - antibodies (which help fight disease)
 - Hormones

Types of Structural proteins

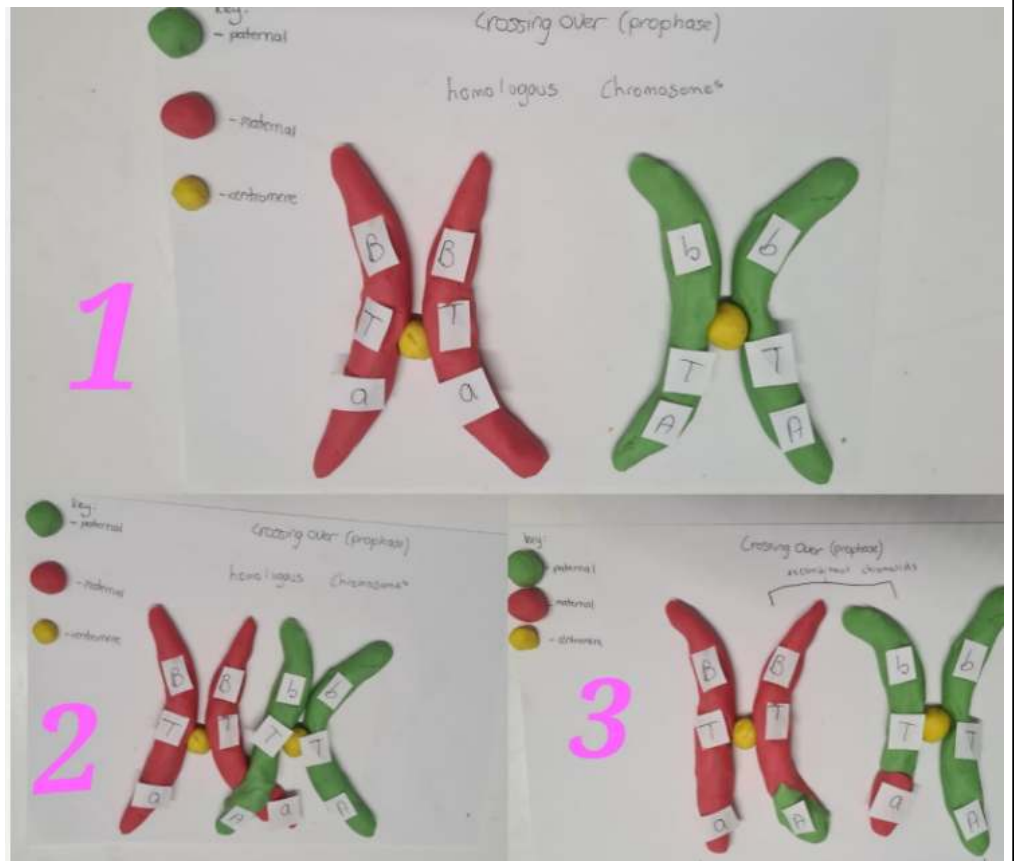
- Support
- Structural proteins are often fibrous and stringy

	<ul style="list-style-type: none"> - Found in consecutive tissues such as skin and cartilages, called <i>collagen</i> - tubulin is a protein in microtubules form cytoskeletons - <u>Movement</u> - <u>contractile</u> proteins also occur in cells, allowing movement to occur - E.g in muscle, <i>actin</i> slides along <i>myosin</i> to contract muscle <p><u>Types of functional proteins</u></p> <ul style="list-style-type: none"> - <u>Regulating metabolic functioning</u> - Enzymes: particularly important in gene functioning, replicating, repairing and transcribing dna. E.g : helicase, DNA & RNA polymerase - Hormones: chemical messengers between cells that can regulate metabolic functions. E.g insulin regulates blood sugar levels - <u>Cell communication</u> - Signalling: communicate messages to a cell about the environment , trigger responses. - biological recognition: receptors(a protein) allow the body to recognize foreign cells (e,g antibodies) - <u>Storage and transport</u> - storage : store chemicals needed for the organism. Eg. ferritin store iron, albumin(eggs) and casein(milk)
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Content: Genetic Variation

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

4.1 conduct practical investigations to predict variations in the genotype of offspring by modelling meiosis, including the crossing over of homologous chromosomes, fertilisation and mutations	CROSSING OVER
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Crossing over occurs when the arms of a homologous pair of chromosomes cross over. Where the arms meet is called a chiasma. At the chiasma, the arms break off their original chromosomes, exchanging the genetic material between the maternal and paternal chromosomes

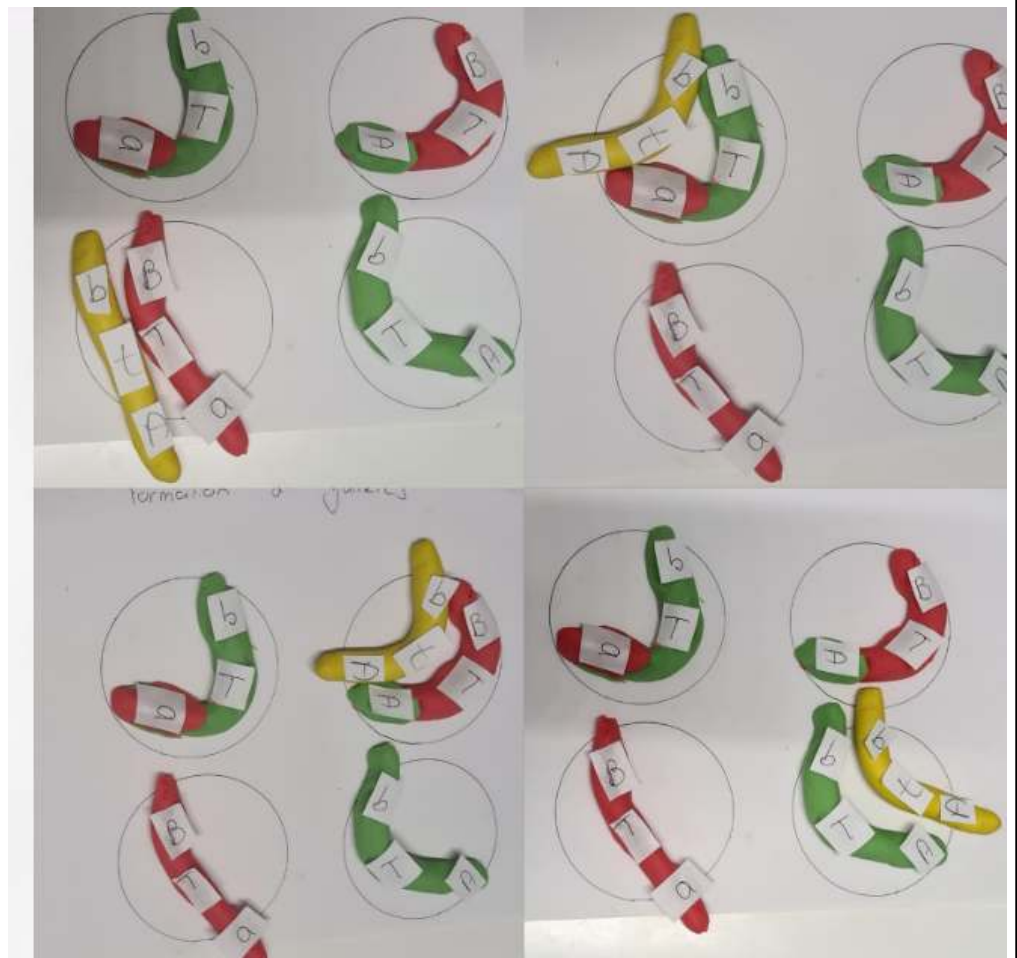
Advantages

- Key
- Simplified process

Limitations

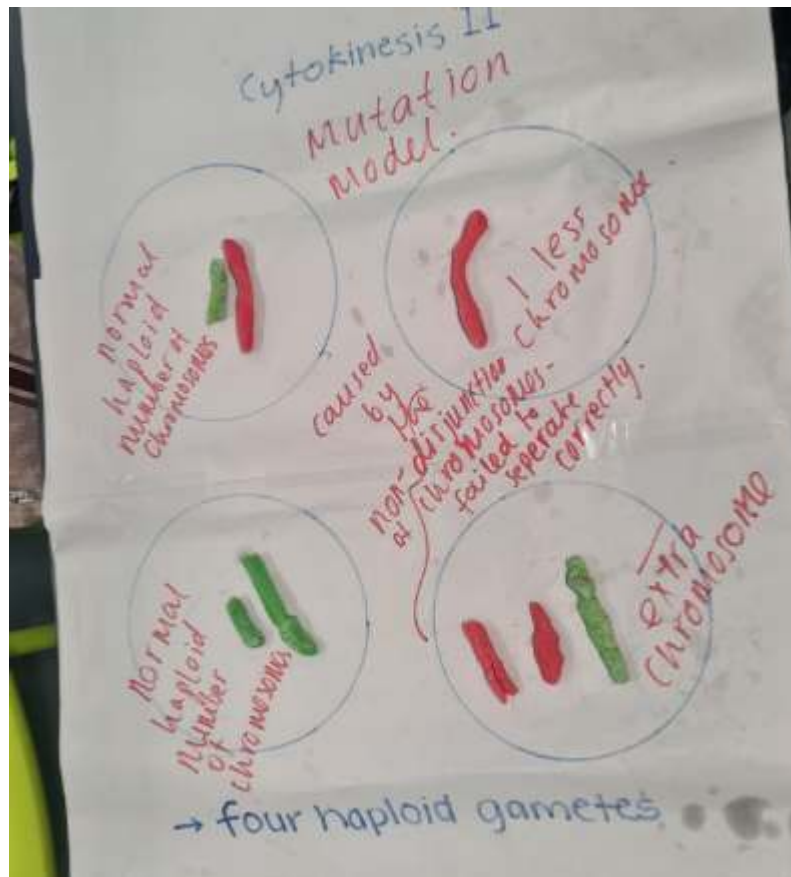
- Static
- Not to scale

FERTILISATION



Fertilisation is when one haploid paternal gamete(sperm) combines with a haploid maternal gamete (egg). Fertilisation restores the cell to a diploid number of chromosomes. Due to independent assortment in metaphase and crossing over., it increases the genetic variation ,causing each gamete to be genetically unique. Furthermore, the fertilisation process further increases variation as the possible genotype is determined from which 2 unique gametes combine.

MUTATIONS



Mutation can further contribute to genetic variation in an individual and genetic viability within a population.

Mutations in meiosis are when the separation of either chromosomes in anaphase 1 or the separation of c=sister chromatids in anaphase 2. In anaphase one, mutations can occur when rather than separating the homologous pairs into one of the new cells, both chromosomes in the homologous pair go into one cell. In anaphase 2, mutations can occur when rather than separate the sister chromatids, both go into one cell. Leaving some cells with $2N-1$ or $2N+1$

4.2 model the formation of new combinations of genotypes produced during meiosis, including but not limited to:

(a) interpreting examples of autosomal, sex-linkage, codominance, incomplete dominance and multiple alleles

AUTOSOMAL

Patterns in the expression of characteristics which are found on **Autosomes**.

- Autosomes only code for non-sexual characteristics(eg eye colour)
- Patterns of Autosomal inheritance include dominant/recessive, codominance and incomplete dominance

Dominant /recessive

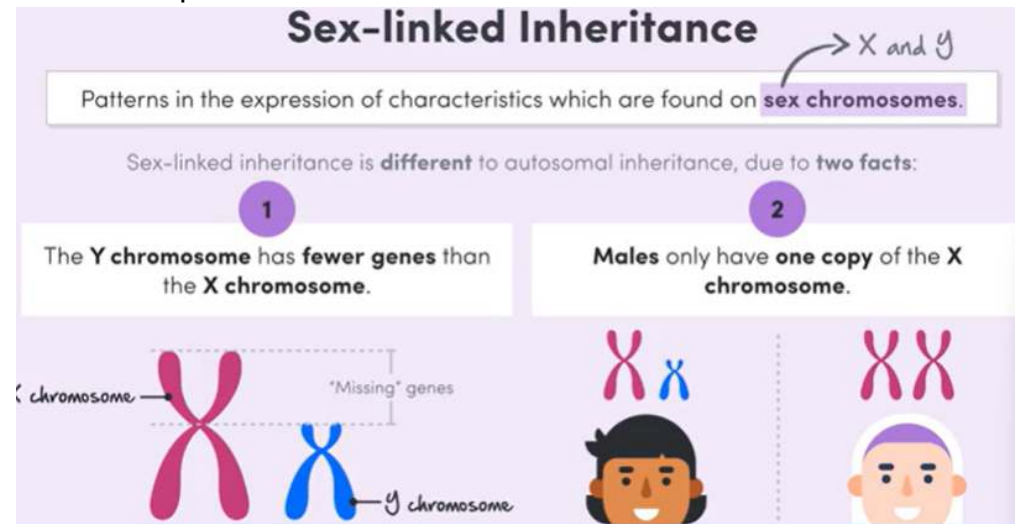
- Dominant allele is always expressed
 - . heterozygous
 - . homozygous
- Recessive allele is only expressed when no dominant allele is present

. homozygous recessive

SEX-LINKAGE

Sex linkage applies to the genes that are located on the sex chromosomes

Inheritance patterns differ between males and females



*14

CODOMINANCE

Both alleles are fully expressed in the organism's phenotype.
The heterozygote appear to have a **"Mixed phenotype"**

Eg roan cattle, have patches of white hair and red hair

INCOMPLETE DOMINANCE

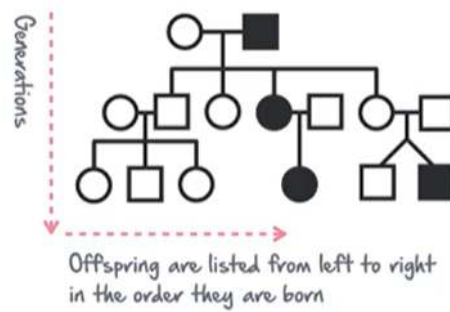
The dominant allele is only partially expressed
The heterozygote had an intermediate phenotype and has a blended effect between the two phenotypes

Eg pink snapdragons are an offspring from red and white snapdragons

(b) constructing and interpreting information and data from pedigrees and Punnett squares

Pedigrees

A pedigree is a diagram which shows how traits are passed on over many generations



SYMBOL	MEANING
○	Female, without trait
●	Female, with trait
□	Male, without trait
■	Male, with trait
—	Mating pair
	Offspring

IDENTIFY PATTERNS → DETERMINE GENOTYPES → PREDICT PHENOTYPES

Reading pedigrees

Step 1: Check the key

Step 2 : look at the sex

- *Autosomal* : number of affected females is around the same and number of affected males
- *Sex-linked* : Almost all affected individual are males

Step 3: Look at the generations

- *Dominant*: Usually appears in every generation
- *Recessive* if the parent are affected, all children will have affected phenotype

Step 4: check your estimate

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

(a) examining frequency data

Allele Frequency

It is the relative proportion of a particular allele in a population. So , it can be represented during the following equation:

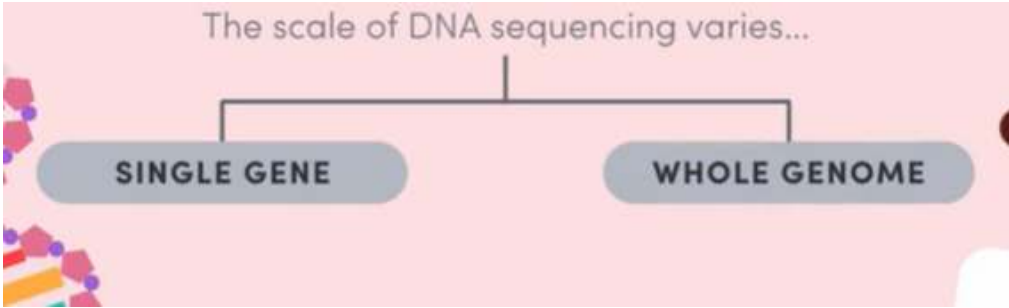
$$\text{Allele frequency} = \frac{\text{Number of an allele in a population}}{\text{Total number of alleles in the population}}$$

- You can work out allele frequencies by taking a **sample** of the population, as long as that sample is big enough for us to say that it is likely to be representative.
- Allele frequencies can be calculated from information about alleles, or by looking at phenotypes so long as we know the relationship between genotype and phenotype

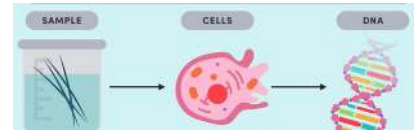
	<div>Example</div> <table><tr><th>Generation</th><th>1st</th><th>Later – 7th</th><th>Logic</th></tr><tr><td>Phenotypic Frequencies: Normal Frogs: 10/12 Albino Frogs: 2/12</td><td>0.82 0.17</td><td>8/12 = 0.6 4/12 = 0.3</td><td>Count the number of normal and albino frogs in each generation. Calculate the phenotypic frequency by dividing the number of each type of frog by the total number of frogs. Round to two decimal places</td></tr><tr><td>Genotypic Frequencies: AA Aa aa</td><td>AA= 7/12 =0.58 Aa= 3/12= 0.25 aa=2/12= 0.16</td><td>AA= 3/12=0.25 Aa=5/12=0.41 aa= 4/12=0.33</td><td>Count the number of each type of genotype (AA, Aa and aa) in each generation. Calculate the genotypic frequencies by dividing the number of each type of genotype by the total number of frogs.</td></tr><tr><td>Allele Frequencies A a</td><td>A= 17/24=0.70 a =7/24= 0.29</td><td>A= 11/24=0.45 a= 13/12 = 0.54</td><td>Count the number of each type of allele (A and a) in each generation. Calculate the allele frequency by dividing the number of each type of allele by the total number of alleles (12 frogs x 2 alleles per frog = 24).</td></tr></table>	Generation	1 st	Later – 7 th	Logic	Phenotypic Frequencies: Normal Frogs: 10/12 Albino Frogs: 2/12	0.82 0.17	8/12 = 0.6 4/12 = 0.3	Count the number of normal and albino frogs in each generation. Calculate the phenotypic frequency by dividing the number of each type of frog by the total number of frogs. Round to two decimal places	Genotypic Frequencies: AA Aa aa	AA= 7/12 =0.58 Aa= 3/12= 0.25 aa=2/12= 0.16	AA= 3/12=0.25 Aa=5/12=0.41 aa= 4/12=0.33	Count the number of each type of genotype (AA, Aa and aa) in each generation. Calculate the genotypic frequencies by dividing the number of each type of genotype by the total number of frogs.	Allele Frequencies A a	A= 17/24=0.70 a =7/24= 0.29	A= 11/24=0.45 a= 13/12 = 0.54	Count the number of each type of allele (A and a) in each generation. Calculate the allele frequency by dividing the number of each type of allele by the total number of alleles (12 frogs x 2 alleles per frog = 24).
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(b) analysing single nucleotide polymorphism (SNP)Inheritance Patterns in a Population																	

Content: Inheritance Patterns in a Population

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

<p>5.1 investigate the use of technologies to determine inheritance patterns in a population using, for example:</p> <p>-DNA sequencing and profiling</p>	<p>Dna sequencing</p> <p>Dna sequencing is the process of determining the sequence of nucleotides in a piece of DNA</p>  <p>The final sequence can be visualised using EITHER</p> <ul style="list-style-type: none"> - gELL ELECTROPHORESIS - CAPILLARY ELECTROPHORESIS - <p>STEPS IN DNA SEQUENCING</p> <ol style="list-style-type: none"> 1. DNA structure 2. PCR 3. Electrophoresis <p>Steps in Gel electrophoresis sequencing</p> <ol style="list-style-type: none"> 1. Collect a DNA sample
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2. Extract dna FROM SAMPLE (chemicals are added to break open cells, dna is then separated from other cell components)
3. **Amplify DNA** (involved using PCR to make lots of copies of the DNA if sample was too small to provide enough NDA)
4. **Perform Sanger sequencing reaction:** separately identify the position of each nucleotide. Use 4 = normal chain determining nucleotidespecial PCRS-
5. **Determine DNA sequences:** run all 4 PCR reaction on the gel electrophoresis , to determine the lengths if the fragments in each reaction



Steps in Capillary sequencing

1. Collect a DNA sample
2. Extract dna FROM SAMPLE (chemicals are added to break open cells, dna is then separated from other cell components)
3. **Amplify DNA** (involved using PCR to make lots of copies of the DNA if sample was too small to provide enough NDA)
4. **Perform Sanger sequencing reaction:** separately identify the position of each nucleotide. Use 4 = normal chain determining nucleotidespecial PCRS- **BUT WITH FLUORESCENT CHAIN DETERMINING NUCLEOTIDES**

Determine DNA sequence

- Run all 4 PCR reactions on a **capillary electrophoresis gel**
 - Smaller fragments move towards the end of the tube faster
 - As each fragment passes the laser, its fluorescent tag lights up
 - Fluorescence is measured by detector & recorded to produce an **electropherogram**

1 Collect a DNA sample, e.g. hair, saliva, blood

2 Extract DNA from sample and separate it from other chemicals, e.g. proteins

3 Amplify DNA using PCR, if necessary

4 Perform 4 Sanger sequencing reactions, using chain-terminating nucleotides

5A

ELECTROPHORESIS GEL

Read the complementary base of the reaction tube with the smallest fragment, and then work your way back towards the wells

5B

CAPILLARY GEL

Read the complementary base of the fluorescent chain-terminating nucleotides, from left to right

In this lesson, we'll look at the main applications of DNA sequencing:

1. Testing for genetic diseases and disorders.
2. Biological research, at the molecular level.
3. Providing evidence for evolution.
4. Personal identification.

Profiling

DNA profiling refers to the process of analysing DNA variations , for the purpose of identification

Short tandem repeats

A string of repeating nucleotide units , where the **number of units varies** between people .

	<p>Result: people have STRS of different lengths , DNA profiles are based on this</p> <p><u>Making DNA profiles</u></p> <ol style="list-style-type: none"> 1. Collect DNA sample 2. Extract DNA from sample and separate it from other chemicals 3. Amplify STR fragments using a PCR with primers which flank the STR regions 4. Determine length of STR fragments using gel/ capillary electrophoresis 5. Interpret the electrophoresis gel/ electrophoresis gram <p>This can be used for</p> <ul style="list-style-type: none"> - Determining parentage (a person's DNA profile considered of a combination of their parents alleles) - identification(when victim/ suspect is the person whose DNA profile matches the one from the crime scene
<p>5.2 investigate the use of data analysis from a large-scale collaborative project to identify trends, patterns and relationships, for example:</p> <p>(a) the use of population genetics data in conservation management</p>	
<p>(b) population genetics studies used to determine the inheritance of a disease or disorder</p>	
<p>(c) population genetics relating to human evolution</p>	

	prokaryote	Eukaryote
Examples	Bacteria and archaea	Animal, plant, fungi
Does this type of cell have DNA?	Yes	yes
Does DNA form chromosomes?	yes	yes

What is the shape of chromosomes?	circular	linear
How many chromosomes?	1	multiple
Where is most of the DNA?	Nucleoid region	nucleus
Where else can DNA be found?	plasmids	Mitochondria, chloroplast