The LOGY UACE BIOLOGY NOTES

INHERITANCE AND EVOLUTION

Competency: The learner appreciates the transmission of traits from one generation to the next, and the **mechanisms that drive change in a gene pool,** by analyzing the concepts of inheritance and evolution, so as to make informed decisions regarding inheritable conditions, for genetic engineering, conservation biology, and health

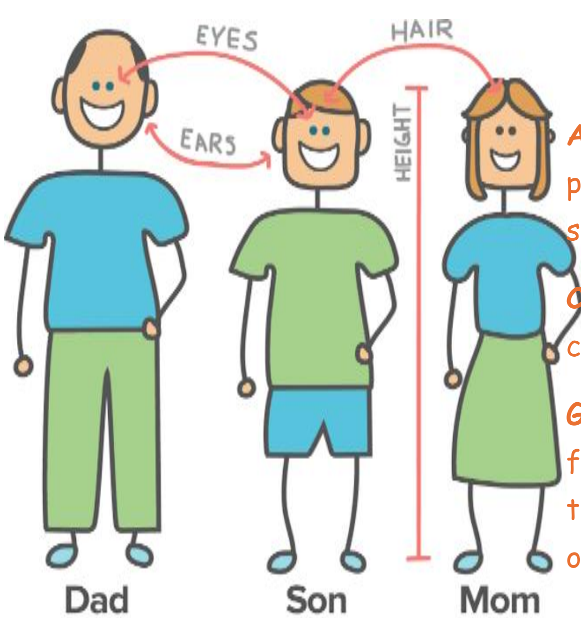
Learning outcomes

a) **Analyses the structural and functional significance of nucleic acids in meiosis and mitosis, their role in cellular functions, and how mutations in nucleotide sequences can contribute to disease (cancer).**

Introduction

Inheritance reference to the **transfer of traits** from generations to generations or from parents to the offspring through **genes**

May Do you think how this happens?......…………………….



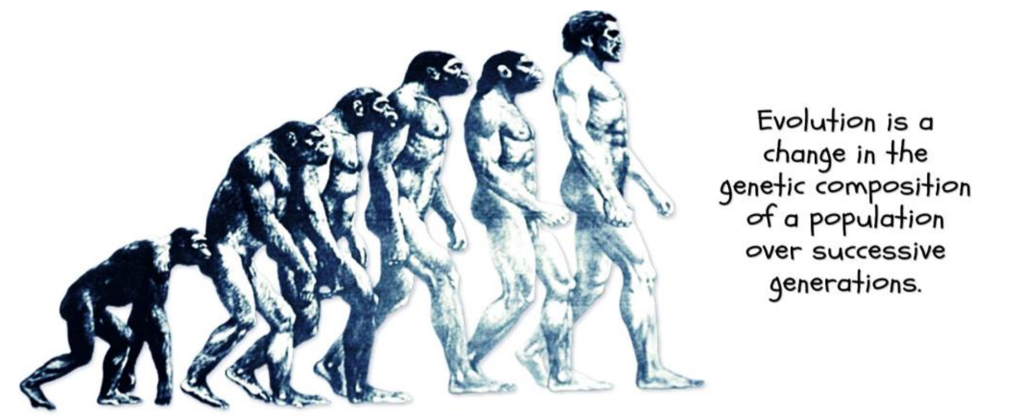
A gene is a section of DNA that codes for a functional protein, DNA is a part of chromosomes; DNA belongs to special series of **chemicals of life** called **nucleic acids**

Chromosomes are thread like structures in the nucleus of a cell that contain the genetic information of the organisms

Genetic information is the hereditary material, primarily found in DNA, that determines the characteristics and traits of an organism and is passed down from parents to offspring

Evolution could be simply defined as descent with modification a simple term used by Darwin; this implies that the modifications take place in genes, this is due to mutations and natural selection.

In more detailed way, **evolution refers to a long-term process in which the population of organisms change from primitive forms to more advanced ones**



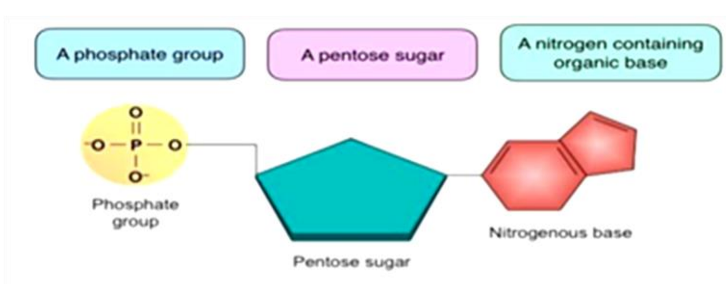
Let's now turn to the first learning outcomes:

NUCLEIC ACIDS

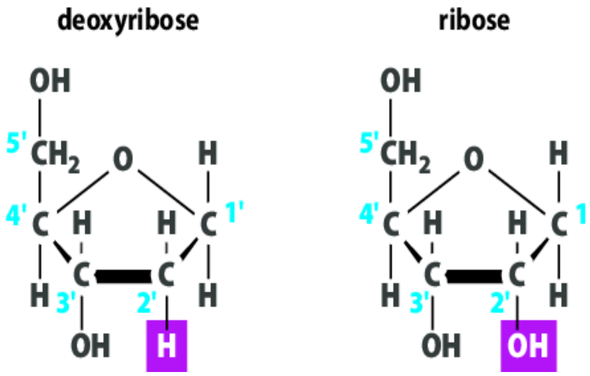
These are **polymers of nucleotides**, they include **DNA and RNA,** these differ in **nitrogen based and pentose sugar as well number of chains or strands.**

NUCLEOTIDES

These are building blocks of nucleic acids; we can describe them by looking at their composition; **Components of nucleotides.**



Pentose sugar; this may be **Ribose** or **deoxyribose**



Identify the difference between the two sugars

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Which of the a above pentose sugars is in DNA or RNA

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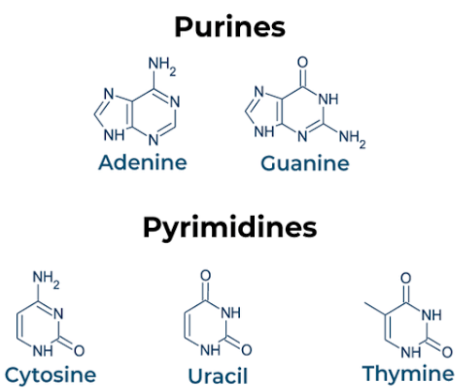
NITROGENOUS BASE/NITROGEN BASE/ ORGANIC BASE

This is a nitrogen containing cycling base or alkaline molecule,

There are two types of nitrogen bases ie

Purines which contain **single rings** **pyrimidines** which contain **two fused rings**



Identify the difference between uracil and thymine

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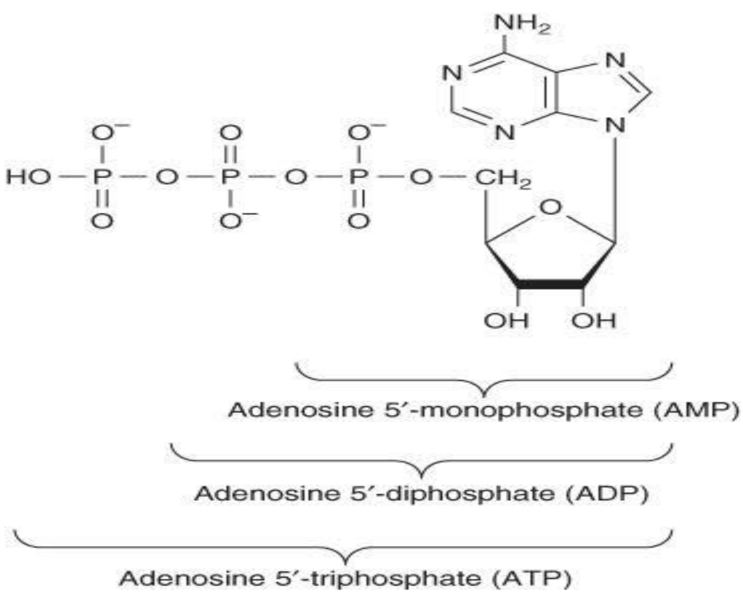
All the nitrogen bases can be found in both DNA and RNA **except uracil which is found only in RNA and never in DNA** and **thymine which is found in DNA only and never in RNA**

PHOSPHATE GROUP

This gives the nucleic acids their acidic character, tell the reason why nucleic acids are acidic yet they **contain nitrogen bases**

Some nucleotides that exist singly (**mononucleotides**) eg ATP, AMP, ADP, **may contain one or more phosphate groups.**



Use your note book

Explain the structure of ATP, ADP and AMP

Research and describe the structure of NAD, NADPH, and FAD

Outline the functions of nucleotides in living organisms

DNA (DEOXYRIBONUCLEIC ACID)

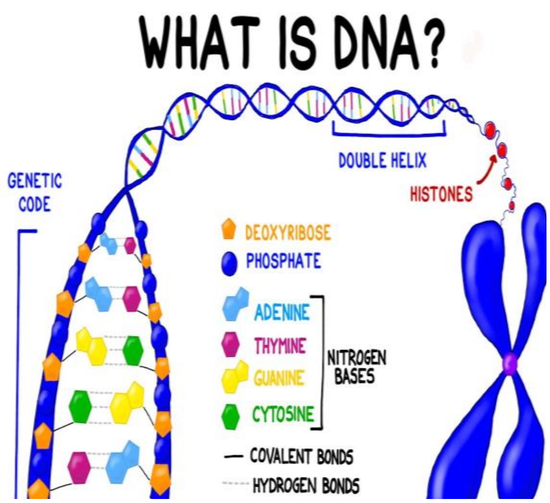
DNA has been called the **"molecule of life"** as **it plays a key role in genetic inheritance and protein synthesis;** resulting into gene expression; thus influences all the physical and behavioral characteristics of organisms.

DNA; is made up of deoxyribose sugar; adenine (A), thymine (T), guanine (G), and cytosine (C) as the nitrogen bases together with phosphate groups.

STRUCTURE OF DNA

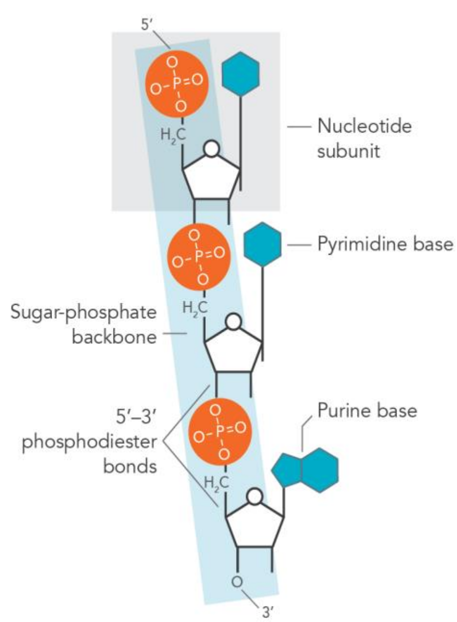
It is a **double helix**; containing two **antiparallel spiral** polynucleotide chains; linked by hydrogen bonds between nitrogen base pairs formed, following the base pairing rule (**complementary base pairing rule**); where **purines pair with pyrimidines**; that is; **cytosine pairs with guanine**, and **adenine pairs with thymine** in the opposite strand (polynucleotide chain); this implies that the ratio of **A: T = 1:1**, and the ratio of **C: G = 1:1**, in DNA; however the **ratio of adenine and thymine to guanine and cytosine vary between species.**

During base pairing; the bases are joined by hydrogen bonds whereby there are **two (2) hydrogen bond between adenine and thymine**, and three **(3) hydrogen bonds between guanine and cytosine.**



NOTE

The sugar phosphate backbone held by phosphodiester bonds

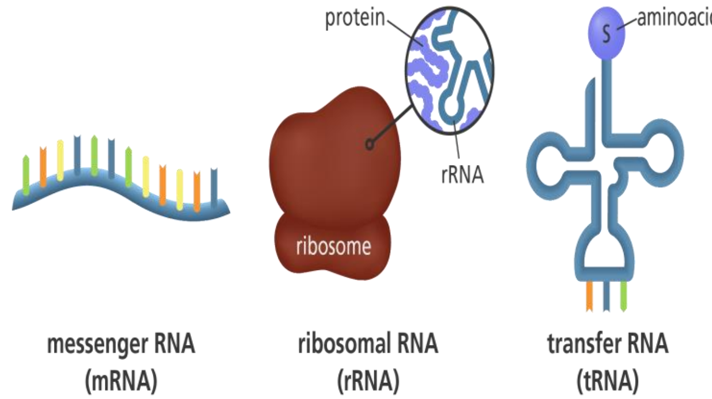


ADAPTATIONS OF DNA AS A HERITARY MATERIAL

- Its location in the nucleus protects it from being affected by cytoplasmic chemicals and enzymes.
- It's extremely large to carry vast information and instructions inform of base sequences.
- The two strands are joined by weak hydrogen bonds which can separate easily during DNA replication and formation of mRNA for protein synthesis.
- It can replicate hence can be transferred from generation to generation without change.
- It's highly coiled to fit in the small space with in the nucleus.
- Complementary base pairing ensures accuracy during DNA replication and protein synthesis

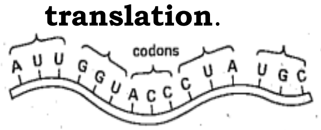
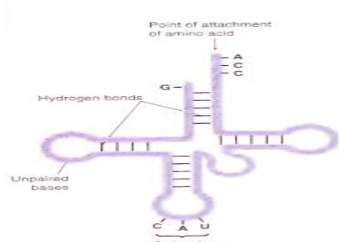
RIBONUCLEIC ACID (RNA)

This usually contains **a single polynucleotide chain** (strand); coiled into an **alpha-helix**; its sugar phosphate backbone has **ribose as its pentose sugar;** the nitrogen bases are similar to those in DNA **except thymine which is replaced by uracil**.

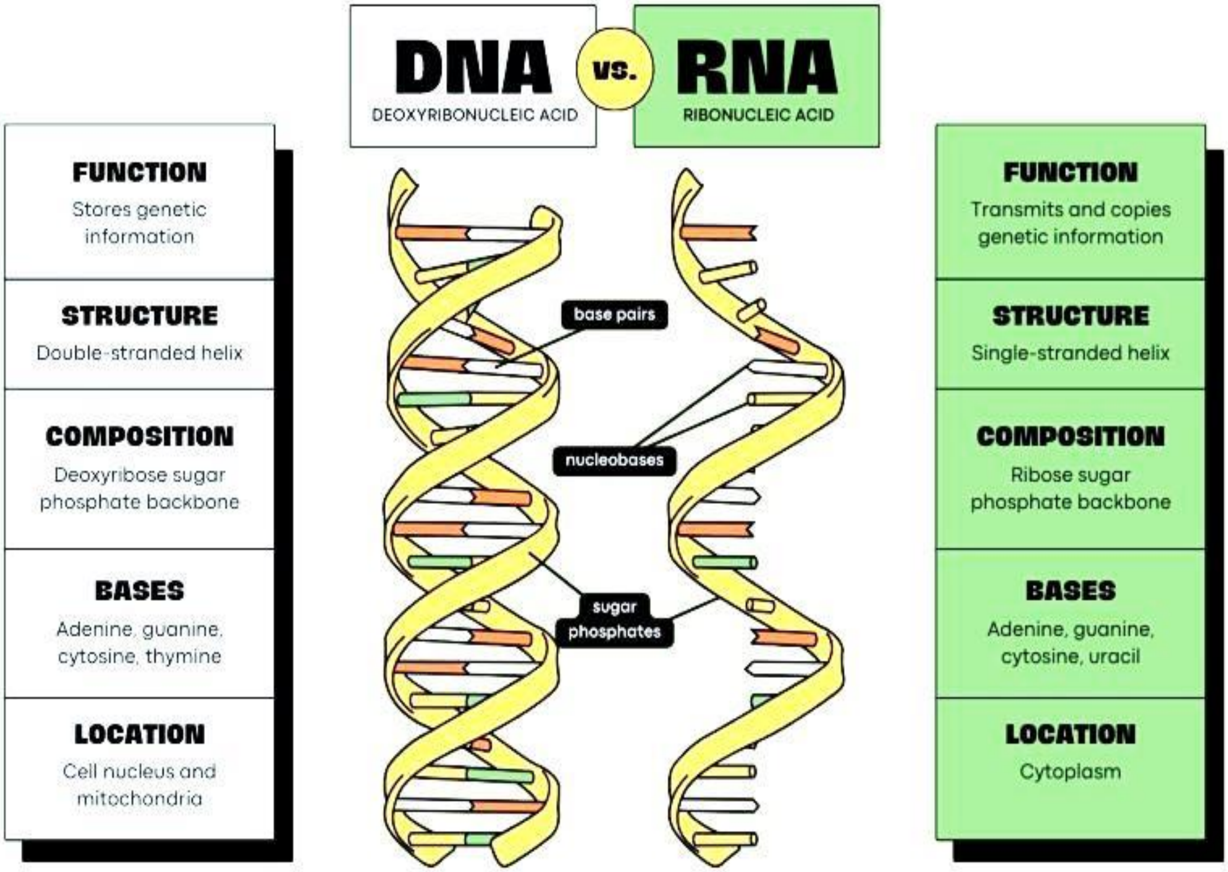


RNA occurs in **three main forms**; these differ in length, shape and function, though they have the same basic structure.

- Messenger RNA **(mRNA)**
- Ribosomal RNA **(rRNA)**
- Transfer RNA **(tRNA)**

mRNA	rRNA	tRNA
✓ Single, helical stranded polynucleotide, ✓ It accounts for about 3%-4% of the total RNA molecules in a cell. ✓ It is synthesized from the nucleus through **transcription**; but transported to the cytoplasm and attaches to ribosomes for **translation**. 	✓ Partly single stranded and double stranded helices; having thousands of nucleotides; (it is a polynucleotide); these are wound into a complex structure; ✓ It is synthesized from the nucleolus in a region called **nucleolar organiser** under the control of the nucleoli, but enters the cytoplasm where it binds with protein molecules to form ribosomes. ✓ It accounts for over 80% of the total RNA molecules in the cell.	✓ It is a single stranded polynucleotide, having 75-90 nucleotides; wound into a double helix with three prominent bulges; the helix is held by hydrogen bonds; ✓ One of the free ends of all tRNA, ends with nucleotides with **ACC;** forming **the amino acid binding site.** ✓ There are about 20 different tRNA molecules which differ by nitrogen bases at the anticodon loop; these are complementary to the codons on the mRNA. ✓ It accounts for about 10%-15% of the total RNA molecules in the cell 

Comparison between DNA and RNA



DNA contains an organism's genetic code or "blueprint." But it can't function alone. It needs RNA to translate and copy the instructions from the cell nucleus to the ribosomes in the cytoplasm. These are the key differences between DNA and RNA.

Outline the similarities between DNA and RNA

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DNA AS A HEREDITARY MOLECULE

DNA is described as a hereditary molecule, as **it contains information that is responsible for physical and behavioral characteristics of all organisms (genes)**; these are passed on from generation to generation (inheritance), the information is in form of codes; sequences of nitrogen bases.

Transfer of DNA to the offspring follows DNA replication during gamete formation in meiosis as well as during mitosis; this is possible due to **complementary base pairing**; as there are many nucleotides in the nucleus; *when the hydrogen bond are broken, the attracted nucleotides complementary pair with those on single strands forming new exact copies of the parent DNA molecule.*

DNA REPLICATION

DNA replication is controlled by a number of enzymes including **helicases, ligases and DNA polymerase.**

It starts by **DNA helicase** attaching its self on the parent DNA molecule at points called **replication origin**; causing it to split by **breaking the hydrogen bonds between base pairs**; this forms a **replication fork**

This is followed by unwinding by completely breaking the hydrogen bond between base pairs; forming separate strands of the parent DNA molecule; which act as **template strands** or molds over which a new strand is copied. To avoid the strands from rejoining, one of the parent strands is partly broken by enzyme **topoisomerase;** this is rejoined after unwinding is complete. The unwinding process is energy requiring hence **uses ATP**.

The separate strands are maintained by DNA binding proteins.

Assembling the leading strand; DNA polymerase attaches itself on the strand that runs in 3'-5' and then a complementary strand is synthesized alongside this strand **continuously**; the **synthesized is the leading strand and runs in 5'-3**, direction; because the **DNA polymerase is moving in the same direction as the unwinding enzyme/ direction of the replication fork.**

Assembling the leading strand; since DNA polymerase works in the 5'-3' direction, the antiparallel parental DNA strand that runs in the 5'-3' direction is copied in small fragments that run in 5'-3 directions; these are called **Okazaki fragments** (named after Reiji Okazaki); these are **joined by DNA ligase** to form a continuous strand.

Assembling the synthesized strands involves attracting free nitrogen bases from the nucleus by complementary bases on the template strand; these are held in position by DNA polymerase until hydrogen bonds are formed between them and then formation of phosphodiester bonds.

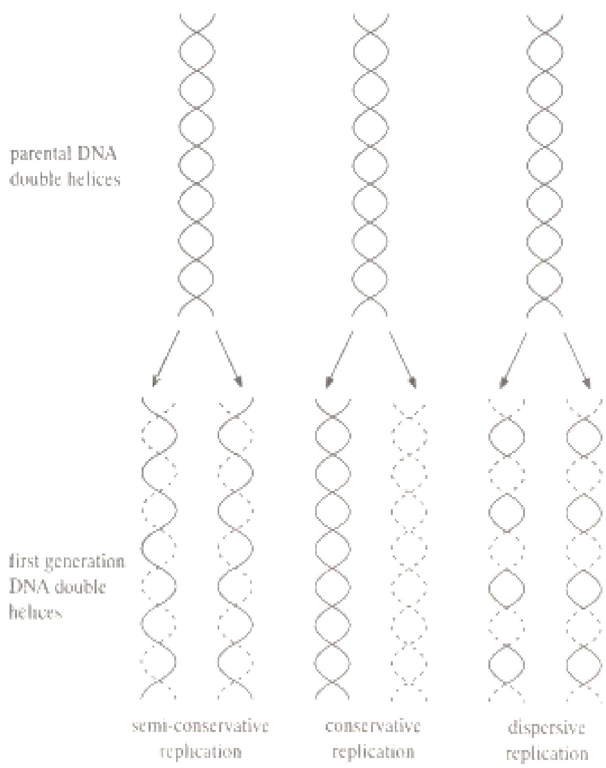
DNA replication is accomplished by removing mismatched nucleotides and replacing them with correct ones; an activity done by DNA polymerase.

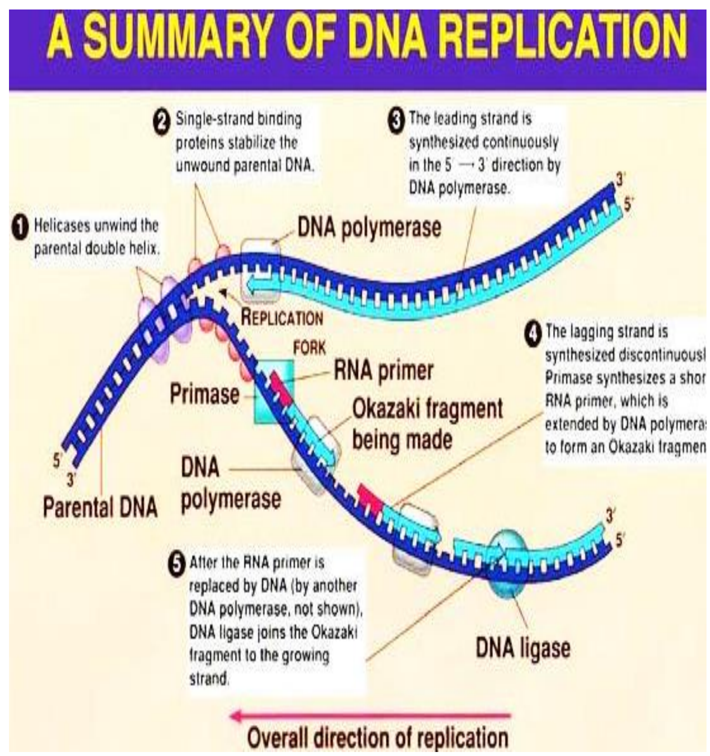
NOTE: synthesis of new strands begins with **primers by primase enzyme** which are short fragments that are then removed; by polymerase enzyme

- From the above, DNA replication can be described to be **semi-discontinuous** as one strand (leading strand) is synthesized continuously and the other (lagging strand) is synthesized from short fragments.
- It can also be described to be **semi-conservative** as one of the strands in the new DNA molecule is the parental strand and the other; is the synthesized strand.

Other DNA replication models are; **conservative**; which postulate that the parental DNA molecule is retained a new DNA molecule comprises of only synthesized strands; by rejoining the template strands.

Dispersive; in which the new DNA molecule has some sections having completely the parental strands and the other sections having only sections of the synthesized strands; as a result of breaking the parental DNA molecule and randomly mixing it with fragments of the newly synthesized DNA fragments





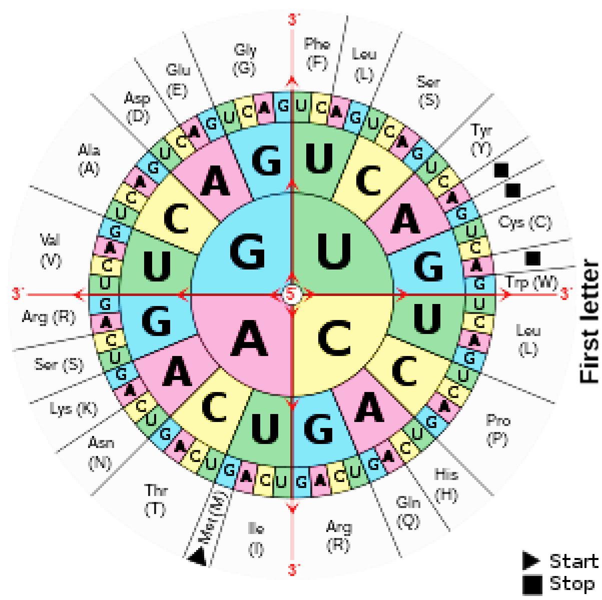
THE GENETIC CODE

Genetic information is stored in DNA in a sequence of nitrogen bases. The **set of rules** that transform this information into a functional protein is referred to as the **genetic code.**

The genetic code can be described from **a triplet of bases in the mRNA molecule known as the codon.**

Since there are only 4 bases (adenine (A), guanine (G), cytosine (C), and uracil (U)), there are only 64 possible combinations of three bases; **$4^3 = 64$,**

Some of these codons are **amino acid codes** while others are not (only three); since there are only 20 amino acids, 64 codons are more than enough to code for the amino acids; **thus one amino acid may be coded by more than one codon.**



Second letter

First letter	U	C	A	G	Third letter
U	UUU } Phe UUC UUA } Leu UUG	UCU UCC } Ser UCA UCG	UAU } Tyr UAC **UAA Stop** **UAG Stop**	UGU } Cys UGC **UGA Stop** UGG Trp	U C A G
C	CUU CUC } Leu CUA CUG	CCU CCC } Pro CCA CCG	CAU } His CAC CAA } Gln CAG	CGU CGC } Arg CGA CGG	U C A G
A	AUU AUC } Ile AUA AUG Met	ACU ACC } Thr ACA ACG	AAU } Asn AAC AAA } Lys AAG	AGU } Ser AGC AGA } Arg AGG	U C A G
G	GUU GUC } Val GUA GUG	GCU GCC } Ala GCA GCG	GAU } Asp GAC GAA } Glu GAG	GGU GGC } Gly GGA GGG	U C A G

Notes

The code must be read in 5’-3’

From the above table and wheel, characteristics of the genetic code are;

1. It is **triplet of bases on mRNA**, each triplet codes for a specific amino acid, though one amino acid may be coded by more than one triplet, and some triplets do not code for amino acids these are stop codons;
   - This implies **each triplet of bases in the polynucleotide of DNA codes for an amino acid in the polypeptide chain**.

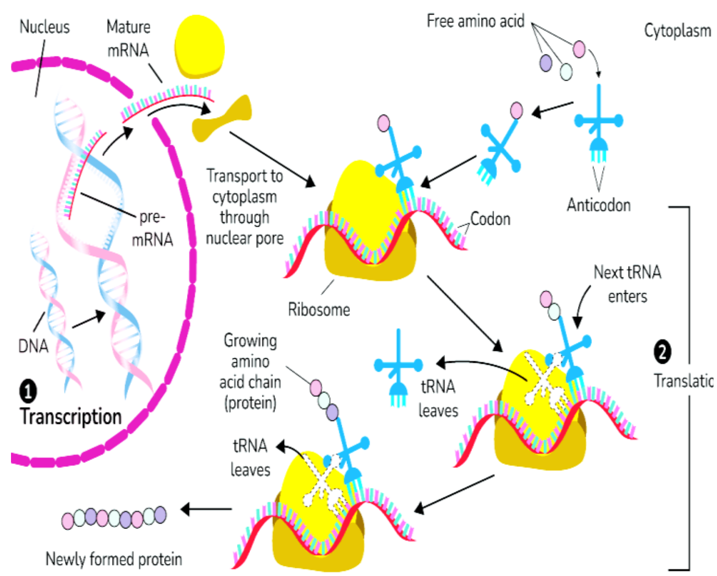
2. The code is also **non-overlapping** in that each triplet is read separately. For example, CUGAGCUAG is read as **CUG-AGC-UAG** and not CUG-UGA-GAG-AGC etc., where each triplet overlaps the previous one, in this case by two bases. Overlapping would allow more information to be provided by a given base sequence, but it limits flexibility. Some viruses, with limited amounts of DNA, may use overlapping codes, but this is very rare.
3. The code is **punctuated;** having start and stop codons; the start codon is AUG; which codes for methionine, while the stop codons are UAA, UAG and UGA; which are non-amino acid codes; they are also called **nonsense codons.**
4. The code is **degenerate**; as there are more than one codon for one amino acid; this implies that one amino acid can be coded by more than one triplet (codon).
5. It is a **universal code;** the triplets code for the same amino acids in all living organisms; and they are precisely the same for all organisms.

In some cases only the first two bases of the codon are relevant. Valine for instance is coded for by GU*, where * can be any of the four bases. Some amino acids have up to six codons. Arginine, for example, has CGU, CGC, CGA, CGG, AGA and AGG; it is significant in cases of mutation effects, by still coding the same amino acid; this is the degenerate character of the code.

PROTEIN SYNTHESIS

DNA makes RNA and RNA makes protein

Protein synthesis includes the process by which a polypeptide is formed from amino acids following the instructions from the genetic code;



The sequence in which the amino acids are assembled during the formation of the protein is based on the arrangement of the codons on mRNA which is formed from a section of DNA known as the gene; therefore the process of **protein synthesis can also be called *gene expression*.**

Protein synthesis involves the following processes;

1. Amino acid synthesis
2. Transcription (mRNA formation)
3. Amino acid activation
4. Translation (formation of a polypeptide chain)

Amino acid synthesis

Formation of amino acids usually occurs in the chloroplasts and mitochondria of plant cells; this involves a series of processes;

i. Absorption of nitrates from the soil; this can be active transport;
ii. Conversion of nitrates to amino groups (reduction); ($-NH_2$)
iii. Combination of these amino groups with a carbohydrate skeleton (e.g. a-ketoglutarate from Krebs cycle); this forms amino acid glutamic acid;

iv. **Transamination;** this transfer of the amino group from one carbohydrate skeleton to another; by this all the 20 amino acids are formed.

Animals obtain their amino acids from plants they eat; however **they can synthesize non-essential, amino acids by transamination;** these are 11 in total; the other nine are essential and must be obtained from diet.

Transcription

Transcription is the process by which a complementary mRNA copy is made from the specific region of the DNA molecule which codes for a polypeptide; this is a **gene.**

It begins with unwinding of the DNA double helix at a region called the **cistron**; by enzyme **helicase;** through breaking the hydrogen bonds between the nitrogen bases. This exposes free potions of DNA strands; the strand of the DNA which is used as a template to synthesize mRNA called the **antisense/non-coding strand**. While the other complementary DNA strand which bears the same sequence as the mRNA is called the **coding/ Sense strand**

Then a large enzyme, **RNA polymerase attaches** its self on the promoter region of the non-coding strand (template strand/ transcribing strand) and build up the mRNA by adding complementary RNA-nucleotides; to the corresponding nitrogen base on the template strand**; (the base adenine attracts RNA-nucleotides with a base uracil, thymine attracts RNA-nucleotides with a base adenine, guanine attracts RNA-nucleotides with a base cytosine while cytosine attracts RNA-nucleotides with a base guanine);** this is complementary base pairing; RNA polymerase moves along the template strand **(cistron / gene)** in 5'-3' direction until it reaches special sequences called the **terminator region**; where it stops adding RNA-nucleotides, and detaches off; this the **primary transcript**

The formed mRNA strand leaves the template strand as the transcribed region of DNA joins back into a helix.

After mRNA has been formed it is processed before it leaves the nucleus; this involves removal of **introns** these are nucleotide base sequences that do not code for a protein; but are part of the cistron. The remaining nucleotide base sequences of the cistron that codes for the functional protein are the **exons;** these are joined together **(spliced together)** to form a **mature mRNA** which then exist the nucleus through the nuclear pore as it is too large to diffuse out.

Amino acid activation

This is the process by which an amino acid is attached to tRNA molecule having a corresponding anticodon to the codon on the mRNA that code for the amino acid; using energy from ATP. It takes place in the cytoplasm of the cell

It begins by the amino acid combining with ATP; and the combines with tRNA to form **amino acid-transfer RNA complex** known as the **amino-acyl tRNA**; the reaction is controlled by the enzyme **amino-acyl tRNA synthetase.**

The energy in ATP is used to form the peptide bond during translation

Translation (formation of a polypeptide chain)

This is a process by which **nucleotide sequences on mRNA are used to form an amino acid sequence in a polypeptide.**

This takes place in the **cytoplasm;** when the mRNA reaches the cytoplasm it is attached on the small subunit of ribosomes at its 5' end.

The **starting codon of the mRNA is usually AUG** which is called **the start codon;** and codes for **amino acid methionine;**

The amino-acyl tRNA molecule with anticodon UAC attaches to the large subunit of ribosomes; this because the anticodons on the anticodon loop are complementary to codons on mRNA; this assembles amino acid methionine; **thus most polypeptides have methionine as their start amino acid;** though **if methionine is not the start of the polypeptide, it can be removed from the polypeptide chain after translation.**

The ribosome then moves one codon along the mRNA and then the amino-acyl tRNA molecule with anticodon complementary to the codon is attached to the large subunit of the ribosome; assembling the next amino acid in the polypeptide chain; the ribosome holds in position the mRNA, tRNA and the associated enzymes controlling the process until a **peptide bond forms between the adjacent amino acids**.

The tRNA molecule which was previously attached to the polypeptide chain now **leaves the ribosome and passes back to the cytoplasm to be reconverted into a new amino-acyl-tRNA molecule;**

This sequence of the ribosome 'reading' and 'translating' the mRNA code continues until it comes to a codon signaling 'stop'. **These terminating codons are UAA, UAG and UGA** (they donot code for any amino acid) they are also called nonsense codons. At this point the polypeptide chain, now with its **primary structure as determined by the DNA,** leaves the ribosome and translation is complete.

- Several ribosomes may become attached to a molecule of mRNA; this whole structure is known as a **polyribosome or polysome**; these speed up the process of translation producing the protein the protein in a short time.

Post translation modification

After translation the polypeptide moves through the lumen of RER; into the Golgi apparatus; where it is modified; by combining it with other polypeptides (to form quaternary protein structure), folding (to form secondary or tertiary protein structure) or combining with carbohydrate fragments to form glycoproteins; this results into **a functional protein.**

MITOSIS AND MEIOSIS

(CELL DIVISION)

Introduction

The cell theory maintains that all organisms are made of cells and all **cells arise from preexisting cells**; you learnt this in cytology; you may be interested in knowing how this takes place;

In this chapter you are going to learn that;

i. Cells arise by splitting preexisting cells—that is, by cell division.
ii. There are two kinds of cell division mitosis and meiosis
iii. The significance of cell division
iv. Negative outcomes if cell division is out of control

HOW CELLS REPLICATE?

For life on Earth to exist, cells must replicate. The general requirements for cellular replication are **(1) copy the DNA (deoxyribonucleic acid),** (2) **separate the copies,** and (3) **divide the cytoplasm to create two complete cells.**

This chapter focuses on eukaryotic cell replication, which is responsible for three key events:

1. Growth, The trillions of genetically identical cells that make up your body are the product of mitotic divisions that started in a single fertilized egg.

2. Wound repair, when you suffer a wound, cellular replication generates the cells that repair your skin.

3. Reproduction when yeast cells grow in bread dough or in a vat of beer, they are reproducing by cellular replication. In yeasts and other single-cell eukaryotes, mitotic division is the basis of asexual reproduction. Asexual reproduction produces offspring that are genetically identical with the parent. Formation of gametes in sexual reproduction also involves cell division (meiosis)

As studies of cell division in eukaryotes began, biologists found that certain chemical dyes made **threadlike structures** visible within nuclei.

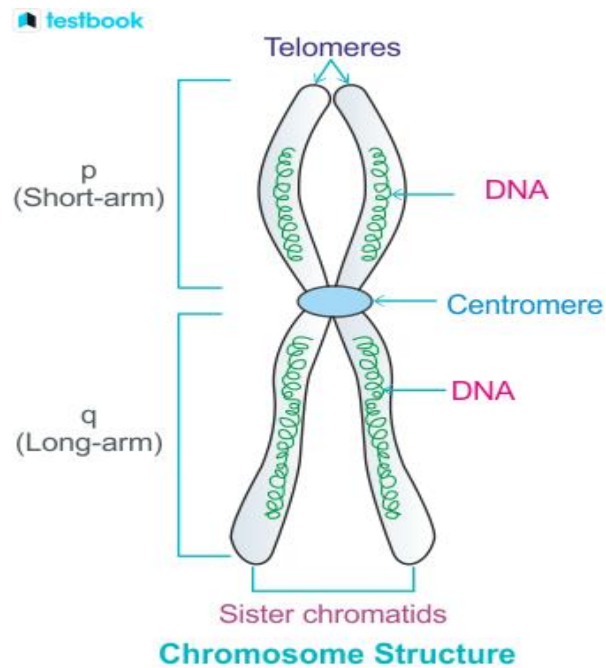
Flemming; introduced the term mitosis, from the **Greek mitos** ("thread"), to describe this process.

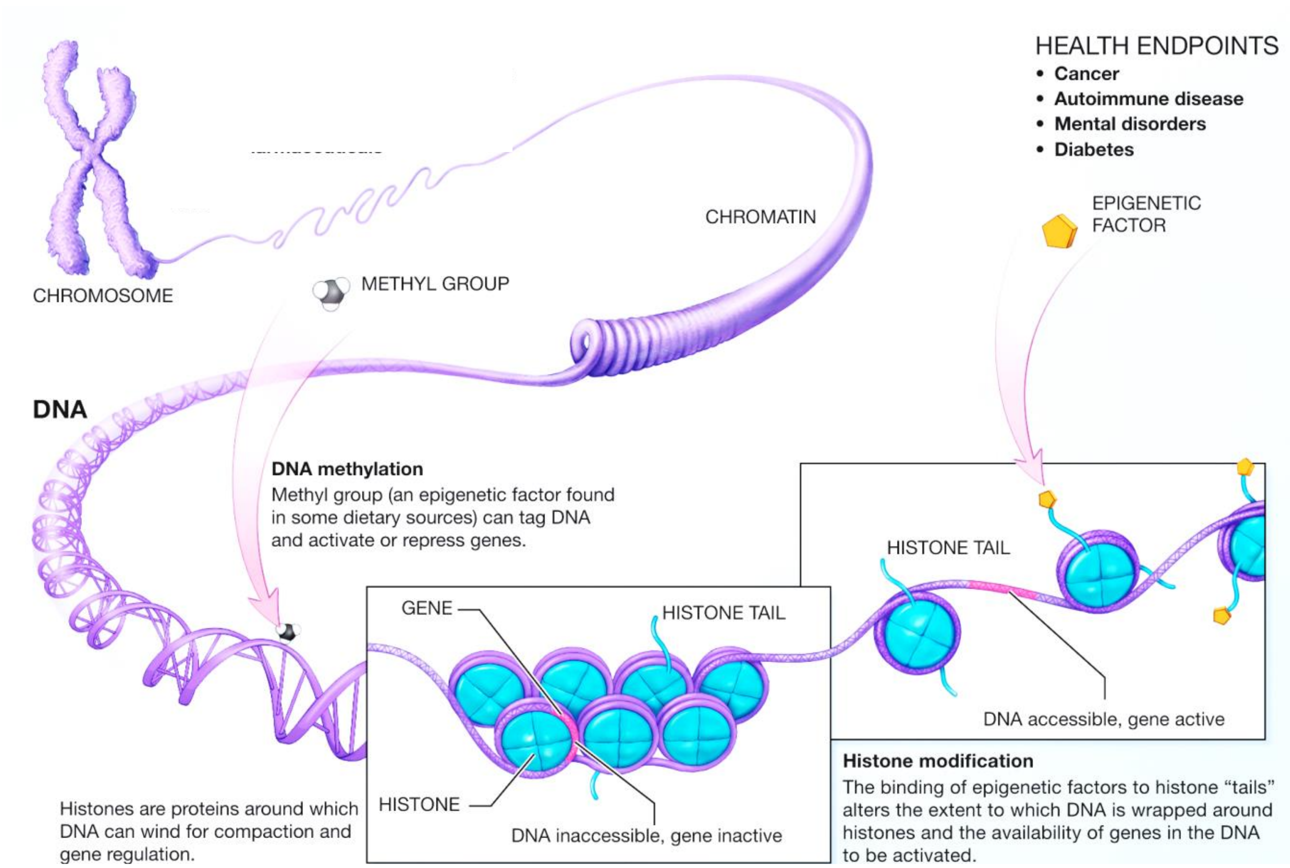
In 1888 Wilhelm Waldeyer coined the term chromosome ("colored-body") to refer to these threadlike structures; A chromosome consists of a single, long DNA double helix that is wrapped around proteins, called histones, in a highly organized manner. DNA encodes the cell's hereditary information, or genetic material. A gene is a length of DNA that codes for a particular protein or ribonucleic acid (RNA) found in the cell.

The chromosomes

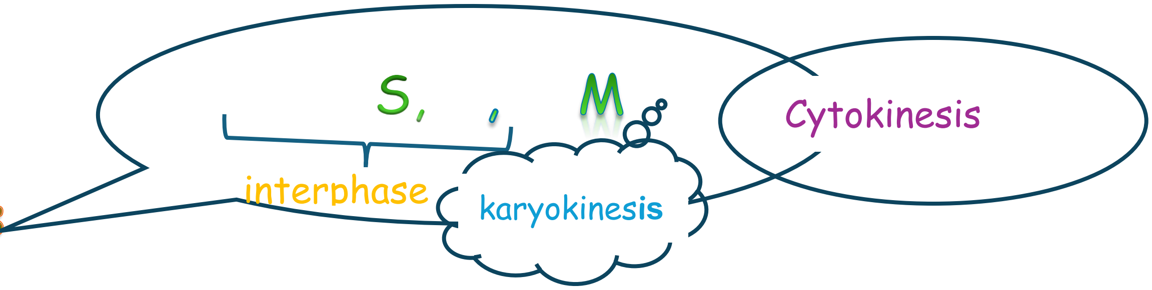
Chromosomes are thread-like structures located within the nucleus of cells that carry genetic information in the form of DNA.

They are essentially packages of DNA, with the long, thin DNA molecules wrapped around proteins for efficient packaging and organization. Humans typically have 23 pairs of chromosomes (46 total) in each cell, with 22 pairs of autosomes and one pair of sex chromosomes (XX for females and XY for males).





Mitosis



Complete cell division cycle is called the **cell cycle;** the cell cycle involves four phases: **M phase** and **an interphase** consisting of the **G1, S, and G2 phases**.

Mitosis is a type of cell division where one cell divides into **two identical daughter cells.** It's a crucial process for **growth, repair, and asexual reproduction in eukaryotic organisms.** The process involves a series of well-defined phases that ensure **each new cell receives a complete and accurate copy of the parent cell's genetic material**.

1. Interphase

Interphase is the stage of the cell cycle where a cell spends most of its time. It's **the period of growth and DNA replication** before cell division (mitosis or meiosis). Interphase is not just a resting phase, **it's a period of intense activity where the cell performs its normal functions, grows, and prepares its genetic material for division;** Interphase is the longest phase of the cell cycle, typically accounting for about 90% of the total cycle time.

Sub-phases:

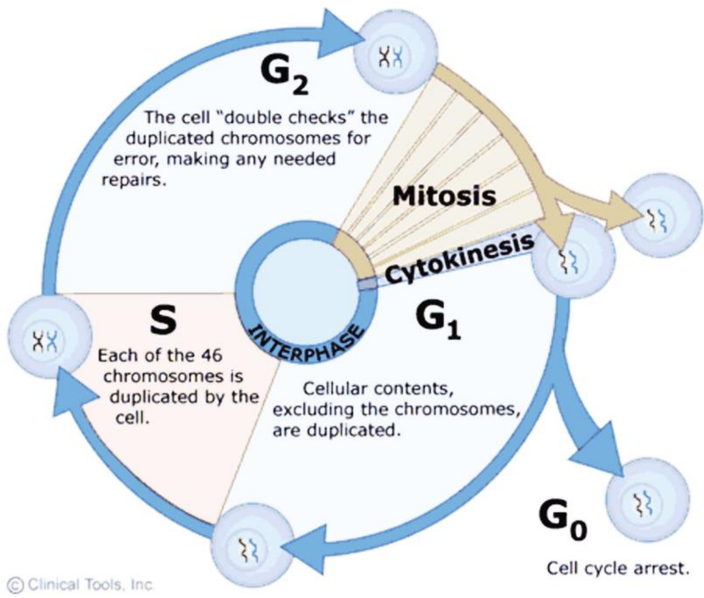
G1 (Gap 1): Intensive cellular synthesis, including new cell organelles. Cell metabolic rate is high. Cell growth occurs. Some substances are produced to inhibit or stimulate onset of next phase as appropriate.

S (Synthesis): DNA replication occurs, creating identical sister chromatids; Protein molecules called histones are synthesized and cover each DNA strand. Each chromosome becomes two chromatids. At this stage the cell is 4n (tetraploid)

G2 (Gap 2): The cell continues to grow by intensive cellular synthesis. **Mitochondria and chloroplasts divide**. Energy stores increase. Mitotic spindle begins to form.

Significance: Interphase is crucial for ensuring that the cell is ready for division, with a complete and accurate copy of the genetic material.

SUMMARY

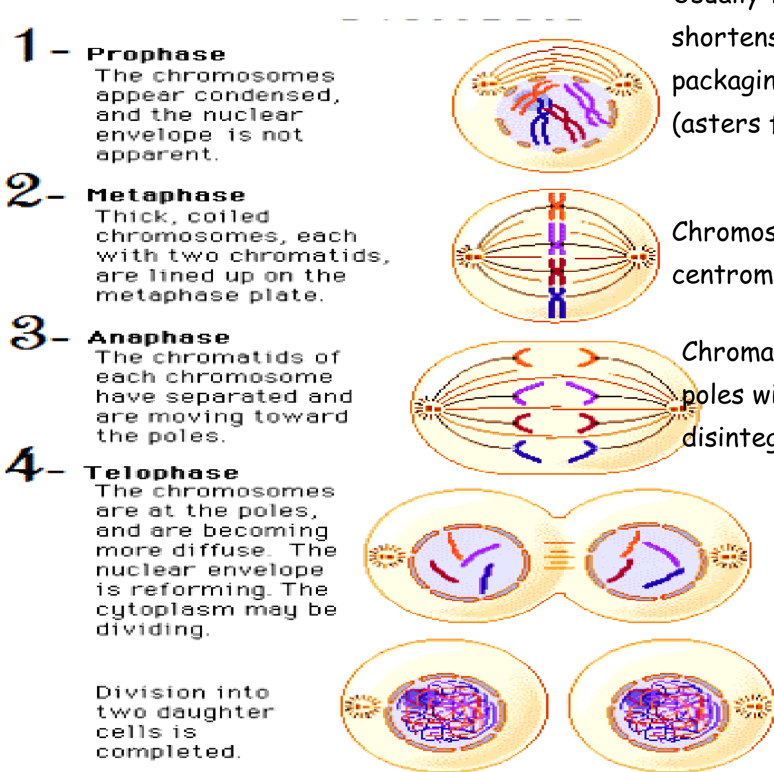


Cytokinesis is the division of the cytoplasm; to give rise to two of daughter cells

Karyokinesis; is the process of nuclear division during cell division, specifically mitosis or meiosis, where the nucleus divides to form two daughter nuclei.

2. M-phase

This involves karyokinesis; mitosis which takes place in four sub phases as described in the figure below;



Usually the longest phase of M-phase Chromosomes shortens and thickens by coiling and tighter packaging of their components. Spindle fibres (asters form); nucleus and nucleolus disappear

Chromosomes align on the spindle equator by their centromeres attached on the spindle fibres

Chromatids are separated and chromatids move to the poles with their centromere leading this is due to disintegration of microtubules in asters

Significance of mitosis

Genetic stability, Mitosis produces two nuclei which have the same number of chromosomes as the parent cell. Since these chromosomes were derived from parental chromosomes by the exact replication of their DNA, they will carry the same hereditary information in their genes. Daughter cells are genetically identical to the parent cell and no variation in genetic information can therefore be introduced during mitosis.

Growth, The number of cells within an organism increases by mitosis and this is the basis of growth in multicellular organisms

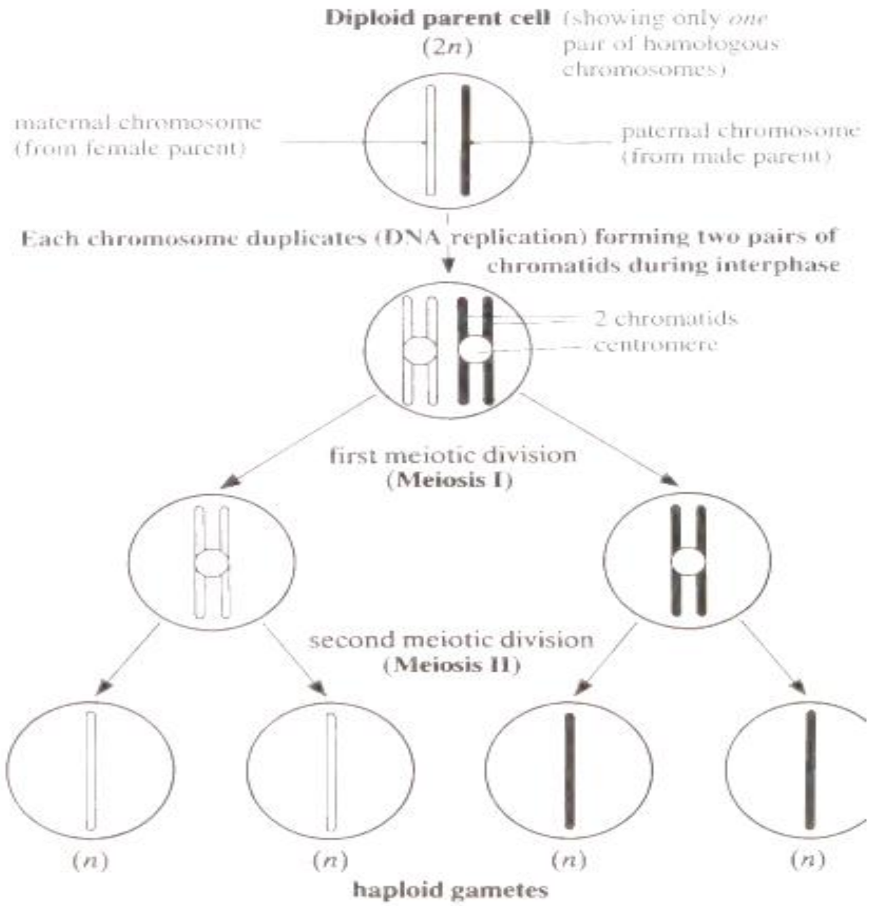
Cell replacement, Replacement of cells and tissues also involves mitosis. Cells are constantly dying and being replaced, an obvious example being in the skin.

Regeneration, some animals are able to regenerate whole parts of the body, such as arms in starfish. Production of the new cells involves mitosis.

Asexual reproduction, Mitosis is the basis of asexual reproduction, the production of new individuals of a species by one parent organism. Many species undergo asexual reproduction.

MEIOSIS

Meiosis is a type of cell division during which a single cell divides twice and produces four haploid daughter cells. These four daughter cells contain half the amount of genetic material and are known as our sex cells (gametes). Gametes are sex cells in organisms, where sperm are found in males and eggs are found in females.

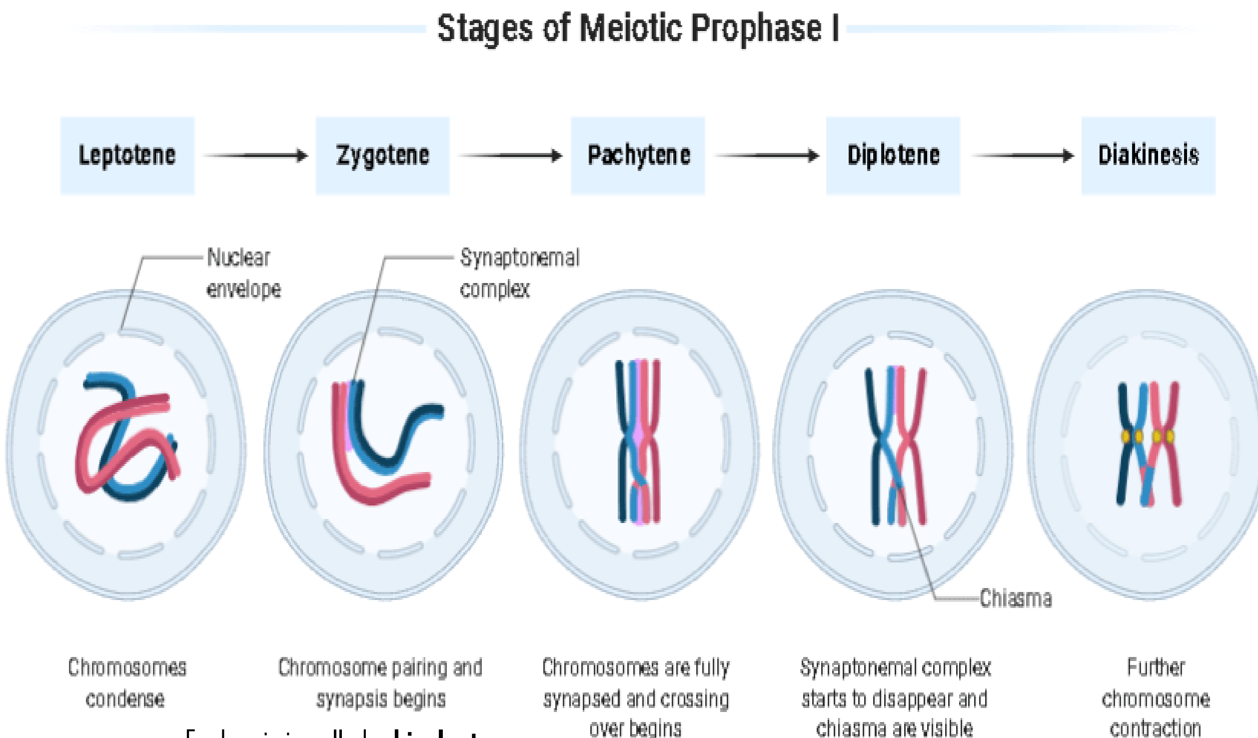


Meiosis I

This is the first meiotic division; in which the homologous chromosomes are separated; crossing over may take place; the sharing of segments between non-sister chromatids of a bivalent.

Prophase I

The longest phase of meiosis; it is occurs in five steps as described below;



Each pair is called **a bivalent.** One of the pair comes from the male parent and one from the female parent.

The non-sister chromatids of homologous chromosomes join; at chiasmata;

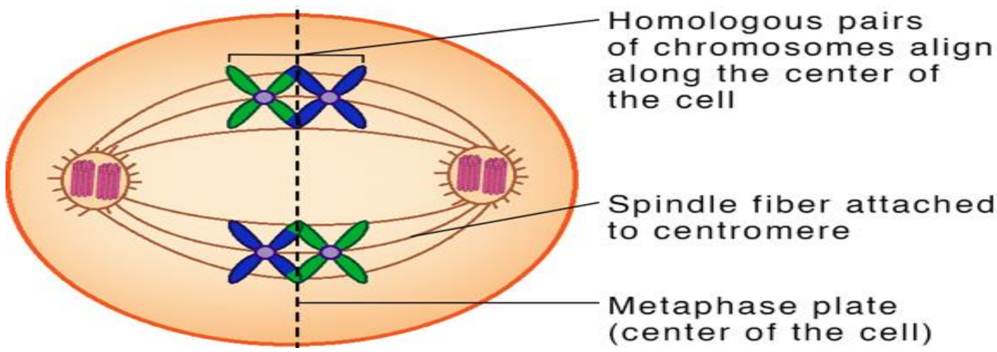
Chromatids of homologous chromosomes continue to repel each other and bivalents assume particular shapes depending upon the number of chiasmata.

By the end of prophase I;

- All chromosomes are fully contracted
- The centrioles (if present) have migrated to the poles;
- The nucleoli and nuclear envelope have dispersed;
- Lastly the spindle fibres form.

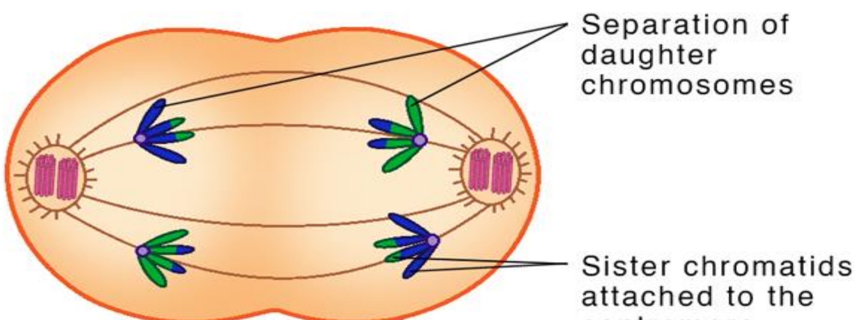
Metaphase I

The bivalents become arranged around the equator of the spindle, attached by their centromeres.

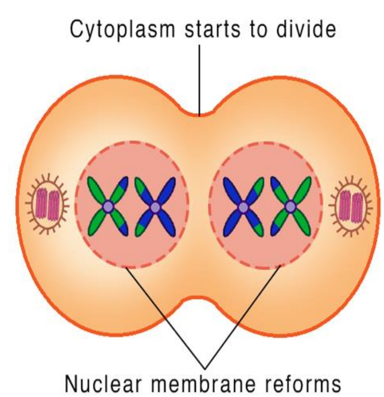


ANAPHASE I

Spindle fibres pull homologous chromosomes, centromeres first, towards opposite poles of the spindle. This separates the chromosomes into two haploid sets, one set at each end of the spindle.



Telophase I



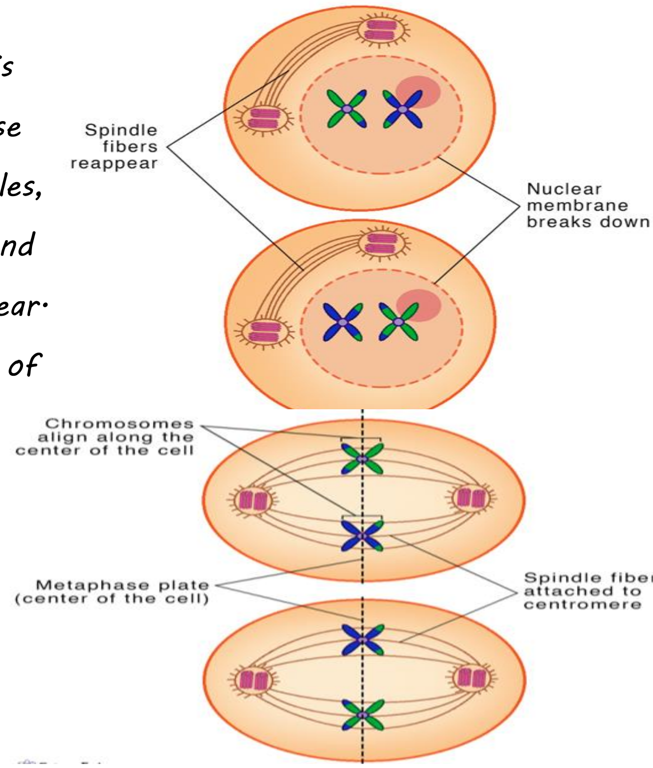
Homologous chromosomes reach their opposite cells marking the end of meiosis l; the chromosomes still have two chromatids which have to separate in meiosis ll; the cells are genetically different; nuclear envelope reforms; and spindle fibres disappear; in plants there is no interphase the cell just precedes to prophase of meiosis ll

INTERPHASE II; this stage is present usually only in animal cells and varies in length. No further DNA replication occurs

MEOISIS II

This stage is similar to mitosis;

Prophase II This stage is absent if interphase II is absent. The nucleoli and nuclear envelopes disperse and the chromatids shorten and thicken. Centrioles, if present, move to opposite poles of the cells and at the end of prophase II new spindle fibres appear. They are arranged at right-angles to the spindle of meiosis I.

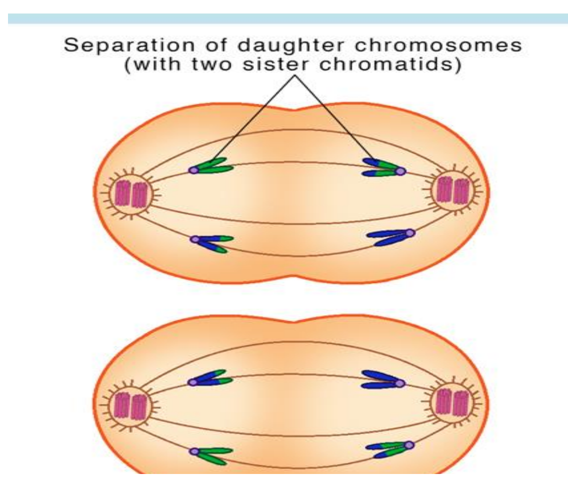


METAPHASE II

Chromosomes line up separately around the equator of the spindle.

They are arranged in ***a plane perpendicular to that in meiosis I***

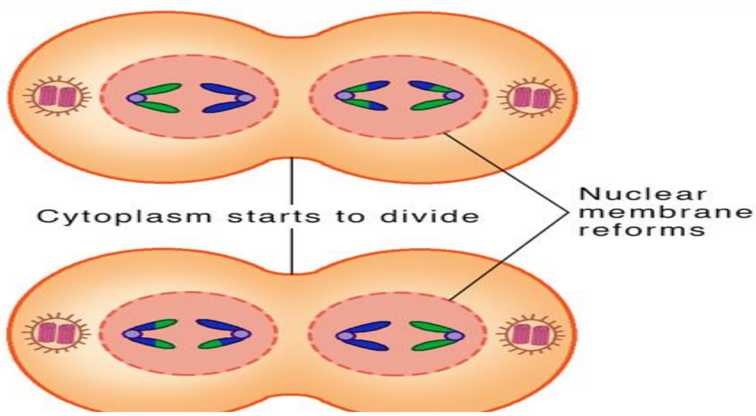
Anaphase II



The centromeres divide and the spindle fibres pull the chromatids to opposite poles, centromeres first

TELOPHASE II

As telophase in mitosis but **four haploid daughter cells** are formed. The chromosomes uncoil, lengthen and become very indistinct. The spindle fibres disappear the centrioles replicate. Nuclear envelopes re-form around each nucleus which possess half of the parent the cell number chromosomes (haploid). Subsequent cleavage (animals) or cell wall formation (plants) will produce four daughter cells from the original single parent cell.



SIGNIFICANCE OF MEIOSIS:

- Provides ***constancy of the chromosome number*** from generation to generation by reducing the chromosome number from diploid to haploid. thereby producing haploid gametes.
- Allows ***random assortment*** of maternal and paternal chromosomes between the gametes.
- Relocates segments of maternal and paternal chromosomes by crossing over of chromosome segments, which "shuffles" the genes and produces a ***recombination of genetic material***.

- Facilitates stable sexual reproduction
- Recombination and independent assortment of homologous chromosomes allow for a greater diversity of genotypes in the offspring.

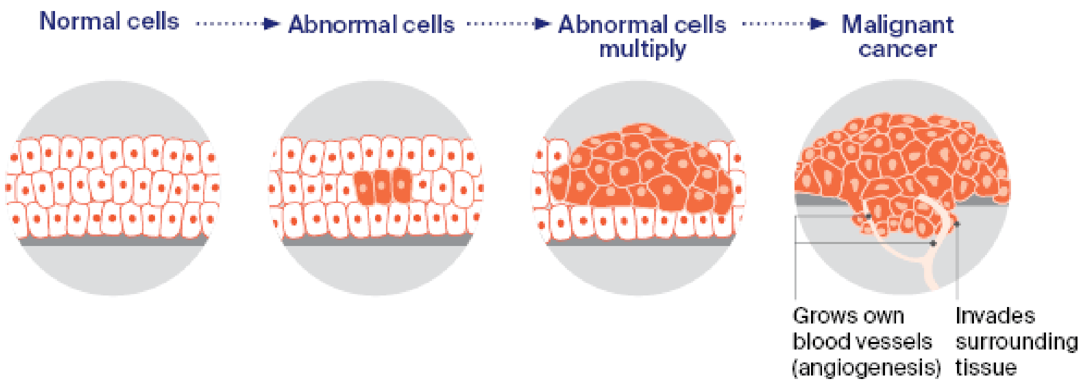
Variation; which is a basis for natural selection

Comparison of mitosis and meiosis I.

	Mitosis	*Meiosis*
Prophase	Homologous chromosomes remain separate No formation of chiasmata No crossing over	Homologous chromosomes pair up Chiasmata form Crossing over may occur
Metaphase	Pairs of chromatids line up on the equator of the spindle	Pairs of chromosomes line up on the equator
Anaphase	Centromeres divide Chromatids separate Separating chromatids identical	Centromeres do not divide Whole chromosomes separate Separating chromosomes and their chromatids may not be identical due to crossing over
Telophase	Same number of chromosomes present in daughter cells as parent cells Both homologous chromosomes present in daughter cells if diploid	Half the number of chromosomes present in daughter cells Only one of each pair of homologous chromosomes present in daughter cells
Occurrence	May occur in haploid, diploid or polyploid cells Occurs during the formation of somatic (body) cells and some spores. Also occurs during the formation of gametes in plants	Only occurs in diploid or polyploid cells Occurs during formation of gametes or spores

CANCER; CAUSES, RISK FACTORS, PREVENTION AND MANAGEMENT

Cancer is a large group of diseases that can start in almost any organ or tissue of the body when abnormal cells grow (divide) uncontrollably, go beyond their usual boundaries to invade adjoining parts of the body and/or spread to other organs (metastasize).



CAUSES OF CANCER

It is caused by a number of factors that activate cancer causing genes; (oncogenes) these factors are collectively known as carcinogens;

Everyone almost has oncogenes which can be activated by carcinogens resulting into cancer that may develop in different body organs depending on the carcinogen exposed to;

- Genetics; due to mutations which may be inherited or acquired; affecting the cell's genome
- Radiations; eg X-rays and UV radiations from the sun
- Chronic infectious diseases such herps, HPV
- Hormonal imbalances such as excess estrogen linked to breast cancer

Use your note book

Explain each of the above cancer treatment methods

b) LEARNER Assess gene technology techniques, their applications in various fields, and the associated ethical implications

BIOTECHNOLOGY

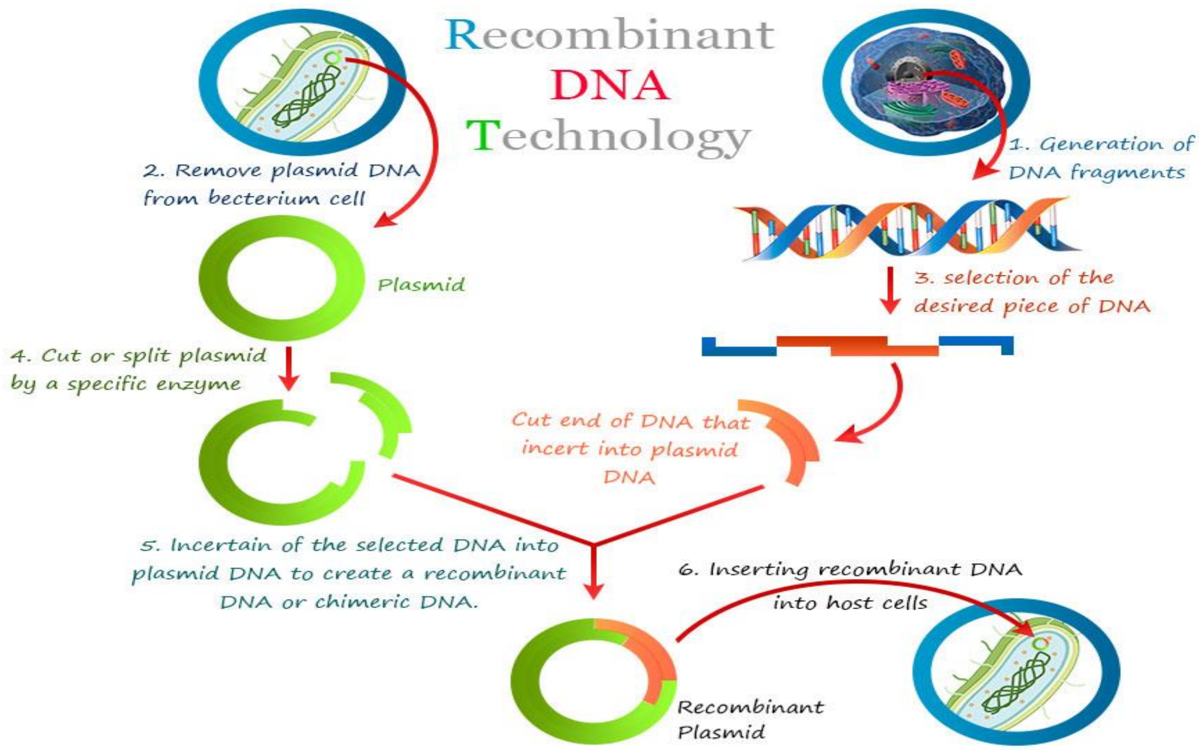
This involves techniques that employ manipulation of cells or cellular components to produce desired products or roganisms

This includes;

Cell cloning, production of GMOs, polymerase chain reaction recombinant DNA technology

Recombinant DNA technology

Recombinant DNA technology comprises altering genetic material outside an organism to obtain enhanced and desired characteristics in living organisms or as their products. This technology involves the insertion of DNA fragments from an organism to another organism of a different species, having a desirable gene sequence via appropriate vector; such as bacterium; this produces hybrid DNA called recombinant DNA



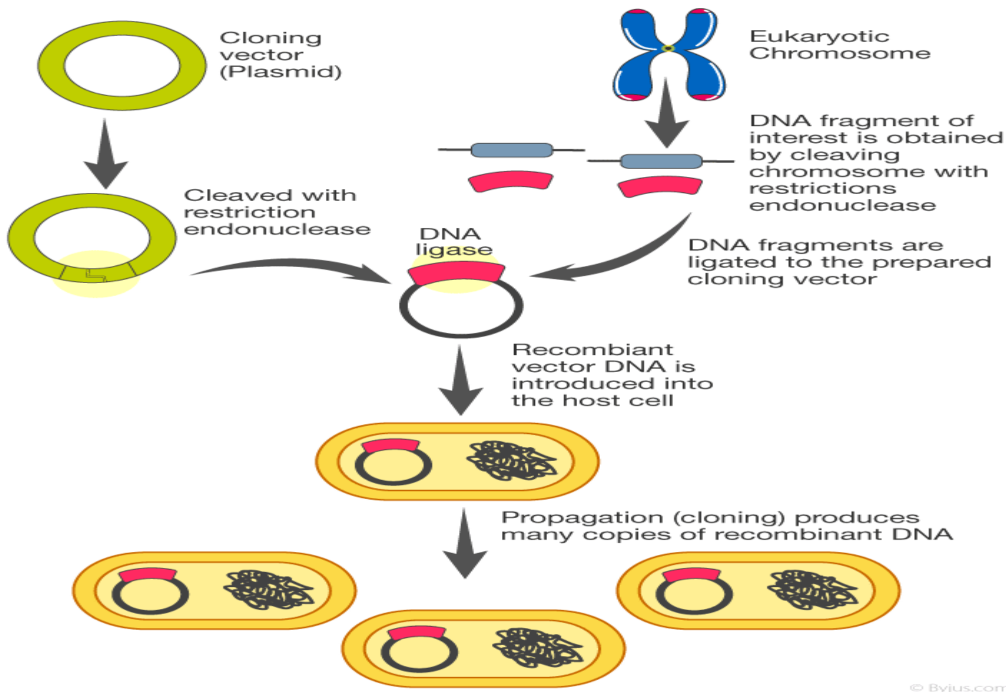
USE YOUR NOTE BOOK

Describe the various applications of recombinant DNA technology;

Hint; production of hormones such as insulin; gene therapy; production of GMOs with desired qualities such as pest resistance

Gene cloning

Is a molecular biology technique which is used for the creation of **exact copies or clones of a particular gene or DNA**.



Significance of gene cloning

- ✓ DNA cloning can be used to make proteins such as insulin with biomedical techniques.
- ✓ It is used to develop recombinant versions of the non-functional gene to understand the functioning of the normal gene. This is applied in gene therapies also.
- ✓ It helps to analyse the effect of mutation on a particular gene.

Polymerase chain reaction (PCR)

The polymerase chain reaction (PCR) is **an in vitro (outside the cell)** DNA synthesis reaction that uses DNA polymerase to replicate a spe cific section of DNA over and over. It generates many identical copies of a particular region of DNA.

NOTE opposite of in vitro is in vivo

Steps of the PCR

Step 1 (selection)

The researcher creates a reaction mix containing an abundant supply of the **four deoxyribonucleoside triphosphates (dNTPs) a DNA sample that includes the gene of interest, many copies of the two primers, and a heat-resistant DNA polymerase called Taq polymerase**.

Primers are short fragments of nucleic acids onto which polymerase enzyme attaches;

Step 2 (denaturation)

The reaction mix is heated **to 95°C**. At this temperature, the double-stranded template DNA denatures (separates), forming single-stranded templates

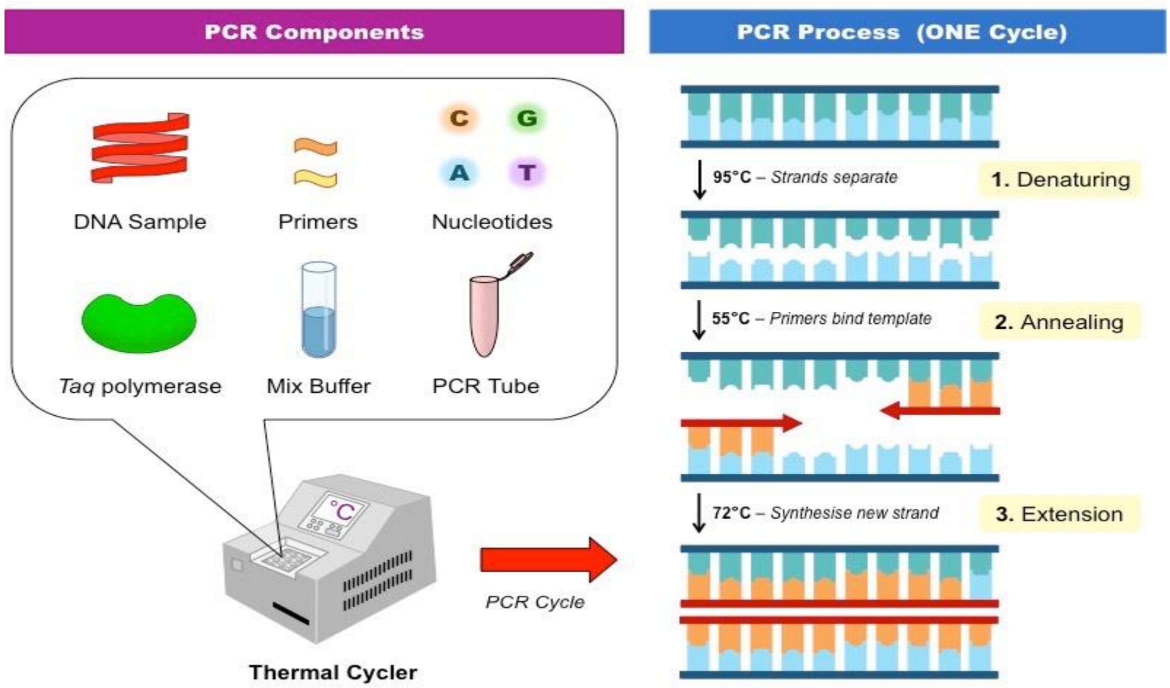
Step 3 (annealing)

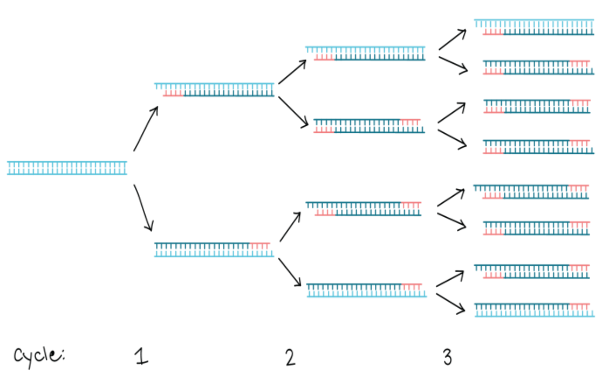
The mixture is allowed to cool **to 50–60°C**. In this temperature range, the primers bind, or anneal, to complementary portions of the single-stranded template DNA. This step is called **primer annealing.**

Step 4 (extension)

The reaction mix is heated to **72°C**. At this temperature, **Taq polymerase efficiently synthesizes the complementary DNA strand from the dNTPs,** starting at the primer. This step is called extension.

Continue repeating steps 2 through 4 until the necessary number of copies is obtained.



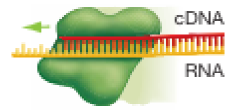
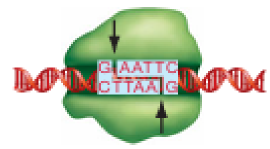
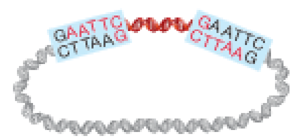
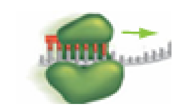


Use your note book; math application

If each cycle doubles the amount of DNA present; how many copies can be formed after **10 cycles of the PCR**

FACT; Taq polymerase is a DNA polymerase found in the thermophilic ("heat-loving") bacterium *Thermus aquaticus*, which was discovered in a hot spring in Yellowstone National Park.

Some Common Tools Used in Genetic Engineering

Tool	Description	How Used	Illustration
Reverse transcriptase	Enzyme that catalyzes synthesis of a complementary DNA (cDNA) from an RNA template.	Many applications, including making cDNAs used in constructing a genetic library.	
Restriction endonucleases	Enzymes that cut DNA at a specific sequence—often a palindromic sequence that is six base pairs long.	Allows researchers to cut DNA at specific locations. Cuts in palindromic sites create "sticky ends."	
DNA ligase	An enzyme that catalyzes the formation of a phosphodiester bond between nucleotides on the same DNA strand.	Ligates (joins) sequences that were cut with a restriction endonuclease. Gives researchers the ability to splice fragments of DNA together.	
Plasmids	Small, extrachromosomal circles of DNA found in many bacteria and in some yeast.	After a target gene is inserted into a plasmid, the recombinant plasmid serves as a vector for transferring the gene into a bacterial or yeast cell, so the gene can be cloned.	
Taq polymerase	DNA polymerase from the bacterium *Thermus aquaticus*. Catalyzes synthesis of DNA from a primed DNA template; remains stable at 95°C.	Responsible for the "primer extension" step in the polymerase chain reaction. Heat stability allows enzyme to be active even after the 95°C denaturation step of PCR.	

THE LOGY INTERACT

a) Make a report showing the applications of gene technology in genetically modified organisms (GMOs), synthetic insulin production, and vaccine development.

b) In groups, discus the ethical, social, and environmental implications of gene technology, particularly GMOs.

The third part of it

c) Apply Mendelian principles to predict inheritance patterns and utilise mathematical models to analyse allele frequencies and genotype distributions within populations.

MENDEL AND THE GENE

INTRODUCTION

In the city where Gregor Mendel lived, there was particular interest in how selective breeding could result in hardier and more productive varieties of sheep, fruit trees, and vines; and an agricultural society had been formed there to promote research into making selective breeding more efficient. Mendel was an active member of this society, and the monastery he belonged to was also devoted to scientific teaching and research

He used garden peas in his experiments where he controlled mating between peas; by removing either the **gynoecium or androecium from experimental flowers** allowing either cross or self-pollination to occur

Use your notebook

Explain the possible characteristics about peas that made them better for genetic experiments

……

Vocabulary Used in Mendelian Genetics

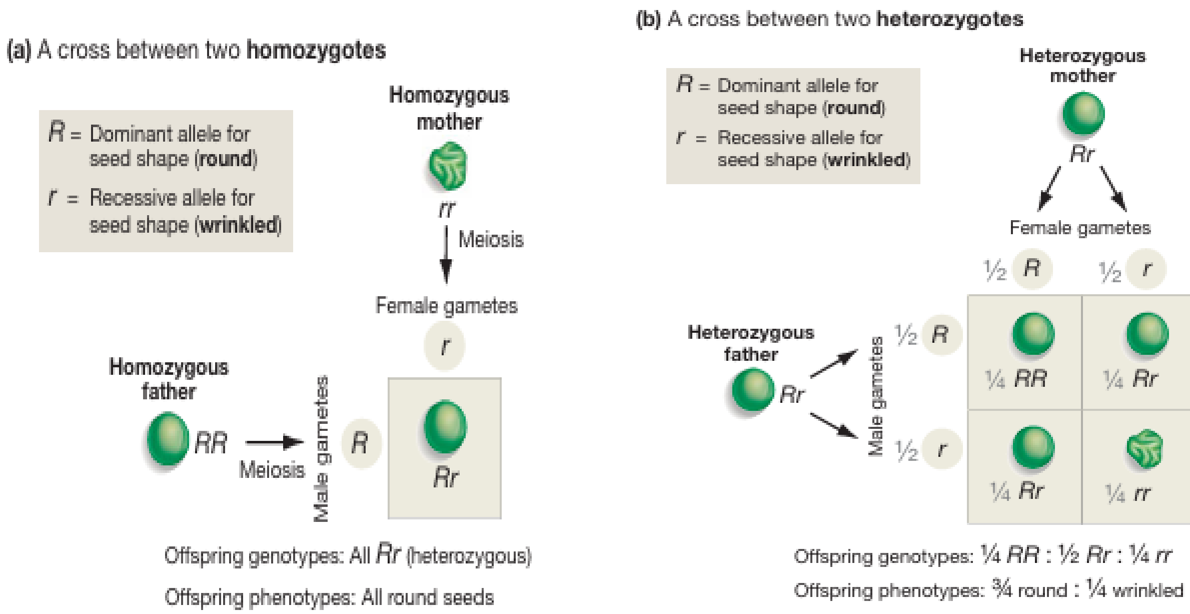
Term	Definition	Example or Comment
Gene	A hereditary factor that influences a particular trait.	This definition will become more precise in later chapters.
Allele	A particular form of a gene.	The two alleles in a diploid may be the same or different.
Genotype	A listing of the alleles in an individual.	In diploids, the genotype lists two alleles of each gene; in haploids, the genotype lists one allele of each gene.
Phenotype	An individual's observable traits.	Can be observed at levels from molecules to the whole organism; influenced, not dictated, by the genotype.
Homozygous	Having two of the same allele.	Refers to a particular gene.
Heterozygous	Having two different alleles.	Refers to a particular gene.
Dominant allele	An allele that produces its phenotype in heterozygous and homozygous form.	Dominance does not imply high frequency or high fitness.
Recessive allele	An allele that produces its phenotype only in homozygous form.	Phenotype "recedes" or disappears in heterozygous individuals.
Pure line	Individuals of the same phenotype that, when crossed, always produce offspring with the same phenotype.	Pure-line individuals are homozygous for the gene in question.
Hybrid	Offspring from crosses between homozygous parents with different genotypes.	Hybrids are heterozygous.
Reciprocal cross	A cross in which the phenotypes of the male and female are reversed compared with a prior cross.	If reciprocal crosses give identical results, the sex of the parent does not influence transmission of the trait.
Testcross	A cross between a homozygous recessive individual and an individual with the dominant phenotype but an unknown genotype.	Usually used to determine whether a parent with a dominant phenotype is homozygous or heterozygous.
X-linked	Referring to a gene located on the X chromosome.	X-linked genes and traits show different patterns of inheritance in males and females.
Y-linked	Referring to a gene located on the Y chromosome.	In humans, Y-linked genes determine male-specific development.
Autosomal	Referring to a gene located on any non-sex chromosome (an autosome) or a trait determined by an autosomal gene.	Mendel studied only autosomal genes and traits.

Mendel's first law and monohybrid inheritance

A monohybrid cross is a genetic cross between two parents that differ in **only one inherited trait**, or locus, to study the inheritance of a single gene.

The process involves **purebred homozygous parents** with contrasting forms of a single gene (alleles) to produce offspring that are heterozygous for that trait; a monohybrid cross allows for the prediction of the genotypic ratio (e.g., 1:2:1) and phenotypic ratio (e.g., 3:1) of the offspring, which is key to understanding Mendelian inheritance and the law of segregation;

Consider a cross involving round and wrinkled seeds;

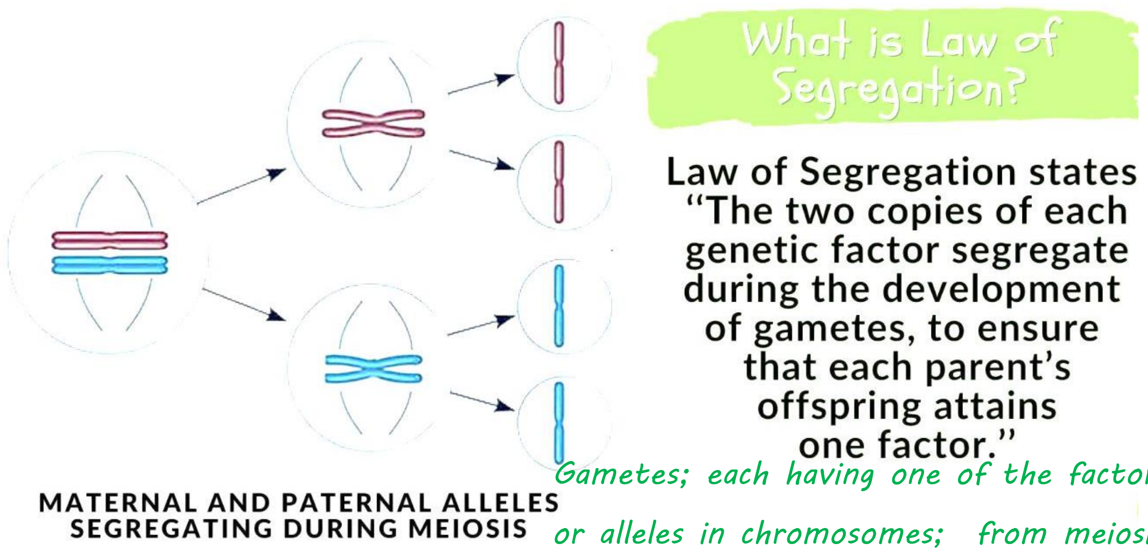


SUMM/ lendel's Model to Explain the Results of a Cross between Pure Lines*

Mendel's Claims	Comments
1. Peas have two copies of each gene and thus may have two different alleles of the gene.	This also turns out to be true for many other organisms.
2. Genes are particles of inheritance that do not blend together.	Genes maintain their integrity from generation to generation.
3. Each gamete contains one copy of each gene (one allele).	This is because of the principle of segregation—the members of each gene pair segregate during the formation of gametes.
4. Males and females contribute equally to the genotype of their offspring.	When gametes fuse, offspring acquire a total of two of each gene—one from each parent.
5. Some alleles are dominant to other alleles.	When a dominant and a recessive allele for the same gene are found in the same individual (a heterozygote), that individual has the dominant phenotype.

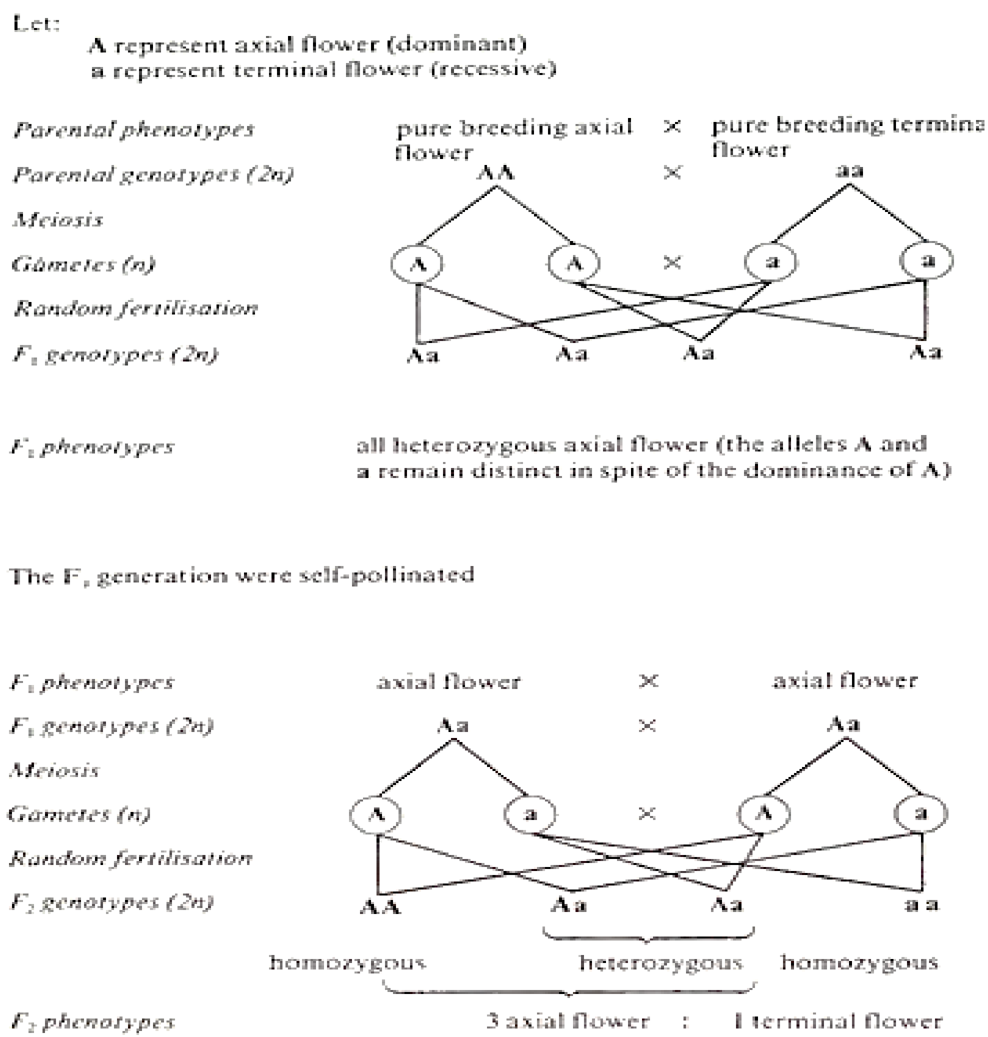
The Principle of Segregation

It states that; ***the characteristics of an organism are determined by internal factors which occur in pairs. Only one of a pair of such factors can be represented in a single gamete*- the factors are alleles**



Gametes; each having one of the factors or alleles in chromosomes; from meiosis

Consider a cross involving plants having axial and terminal flowers

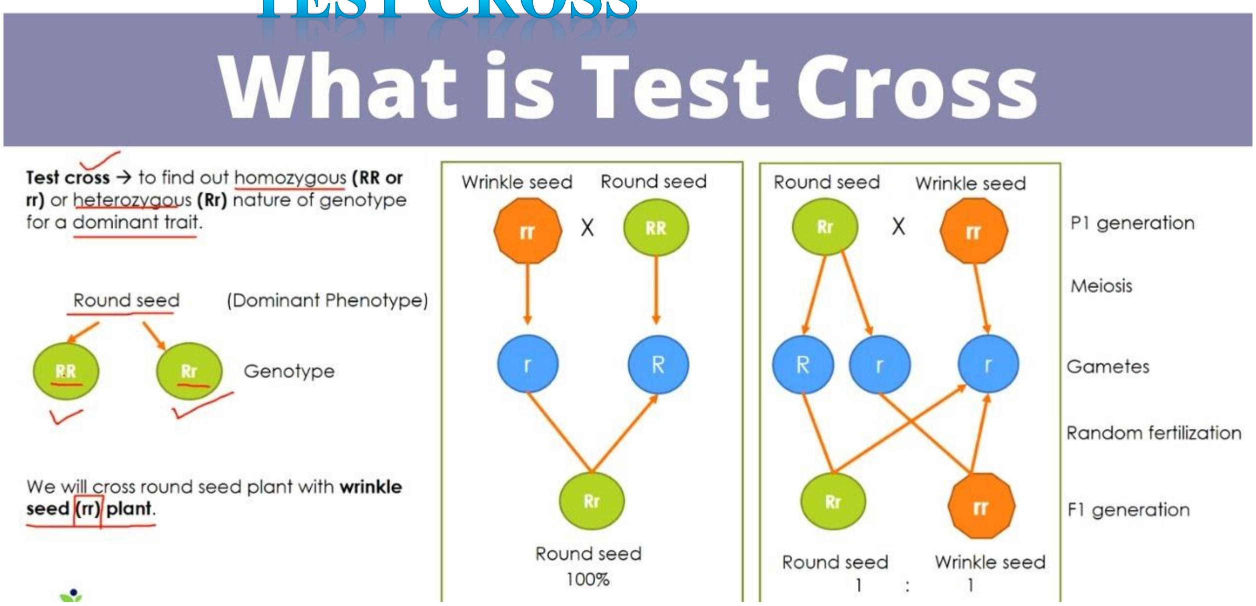


Full genetic explanation of one of Mendel's monohybrid crosses. (2n represents the diploid condition, n represents the haploid condition;

Explanations based on this example are given in the table below

Genetic term	Explanation	Example
gene	The basic unit of inheritance for a given characteristic	flower position
allele	One of a number of alternative forms of the same gene responsible for determining contrasting characteristics	A or a
locus	Position of an allele within a DNA molecule	
homozygous	The diploid condition in which the alleles at a given locus are identical	AA or aa
heterozygous	The diploid condition in which the alleles at a given locus are different	Aa
phenotype	The observable characteristics of an individual usually resulting from the interaction between the genotype and the environment in which development occurs	axial, terminal
genotype	The genetic constitution of an organism with respect to the alleles under consideration	AA, Aa, aa
dominant	The allele which influences the appearance of the phenotype even in the presence of an alternative allele	A
recessive	The allele which influences the appearance of the phenotype only in the presence of another identical allele	a
F_1 generation	The generation produced by crossing homozygous parental stocks	
F_2 generation	The generation produced by crossing two F_1 organisms	

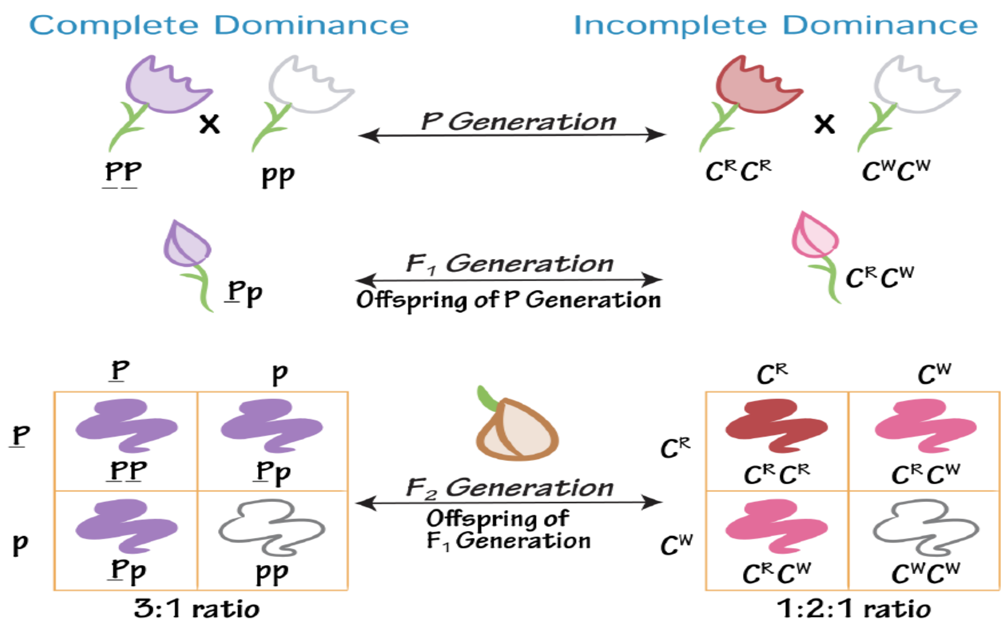
TEST CROSS



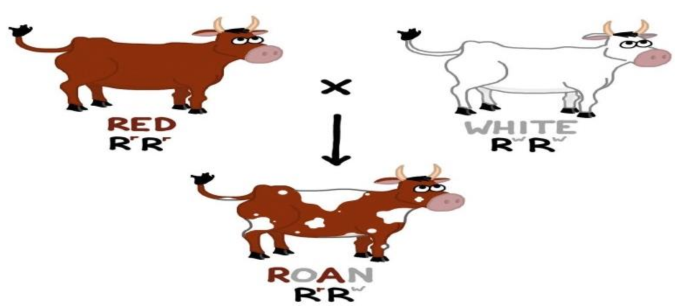
In some cases mendel's first law is an exception; I.e does not apply

1. Co-dominance and incomplete dominance

First let's relate incomplete dominance and complete dominance



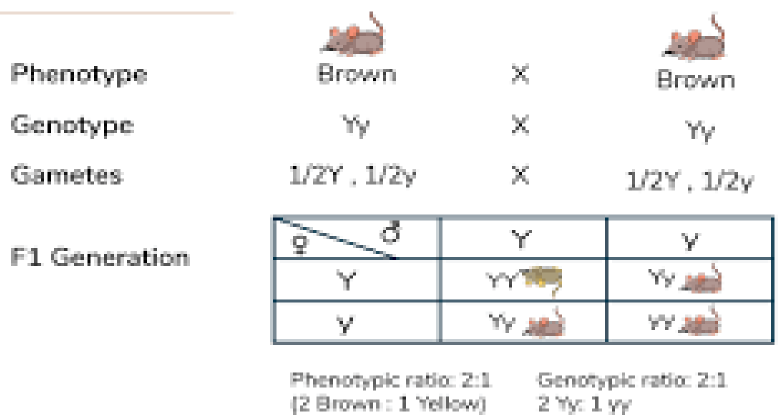
Co-dominance has the same effect only that the heterozygotes have phenotypes for both parents think of roan cows



2. Lethal genes/ alleles

Lethal genes, or lethal alleles, are genes that **cause an organism's death or significantly reduce its viability**. These genes are typically mutations in essential genes required for an organism's survival, growth, or development.

Lethal genes can be dominant, meaning they cause death even if only one copy is present, or recessive, requiring two copies to cause death. They can also be conditional, only acting under specific environmental conditions, or act at different life stages, from embryonic death to late-onset lethality



Note the effect on phenotypic ratios

The Y allele is dominant for color and recessive for lethality

3. Multiple genes

A single characteristic may appear in several different forms controlled by three or more alleles, of which any two may occupy the same gene loci on homologous chromosomes; these are called **multiple alleles; eg blood group inheritance in humans**

Example of Multiple Alleles

Human blood type is controlled by three alleles – I^A, I^B and i. Alleles I^A and I^B are dominant over i, but I^A and I^B are codominant

Phenotype (blood type)	Genotypes
A	$I^A I^A$ or $I^A i$
B	$I^B I^B$ or $I^B i$
AB	$I^A I^B$
O	ii

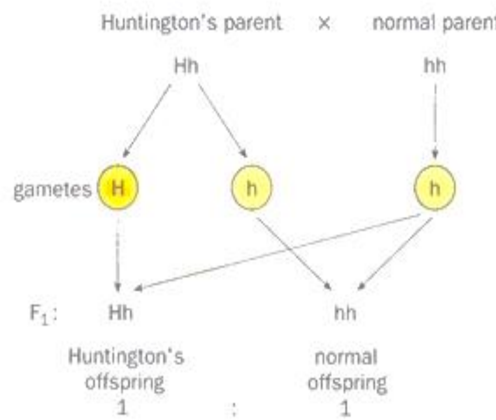
In some books; **I** is written as I^O **(most acceptable)**

Biology around us

Huntington's disease

Huntington's is a rare inherited condition which affects about 1 in 20000 people in Britain. The symptoms are particularly distressing. The cells of the brain degenerate and the patient's coordination is affected.
The patient becomes moody and depressed, memory becomes affected, they eventually become totally disabled and ultimately die.
The most sinister aspect of Huntington's is the fact that the symptoms only become apparent when patients are in their 30s or 40s.
By this time the person may well have had a family and so unintentionally passed the defective allele on to the next generation.

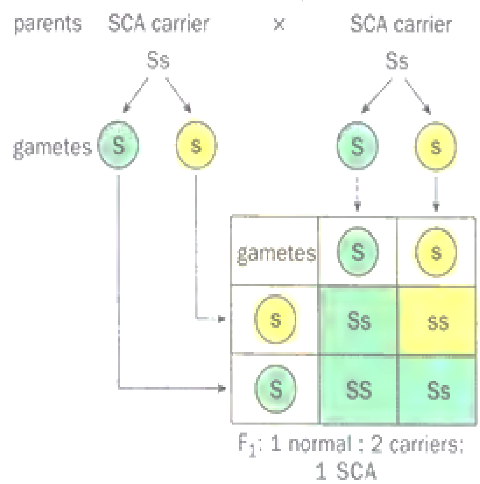
Huntington's disease is caused by a dominant allele.
So only ***one*** allele is necessary to give the disease.
So all heterozygous people are sufferers.



Sickle cell anaemia

Sickle cell anaemia (SCA) is a human blood disease caused by the inheritance of a recessive allele.
The red blood cells are malformed, taking on a crescent or sickle shape.
The sickle cells get stuck in capillaries and inhibit the circulation of oxygen to the tissues.
This can lead to death at an early age.

SCA is due to the recessive allele not coding for the correct form of haemoglobin.
A sufferer must have inherited two recessive alleles from its parents.
Carriers or heterozygous individuals have some normal red blood cells and some sickle cells, so they are not so badly affected.



USE YOUR NOTE BOOK

What daily challenges can be solved by applying knowledge of monohybrid inheritance?

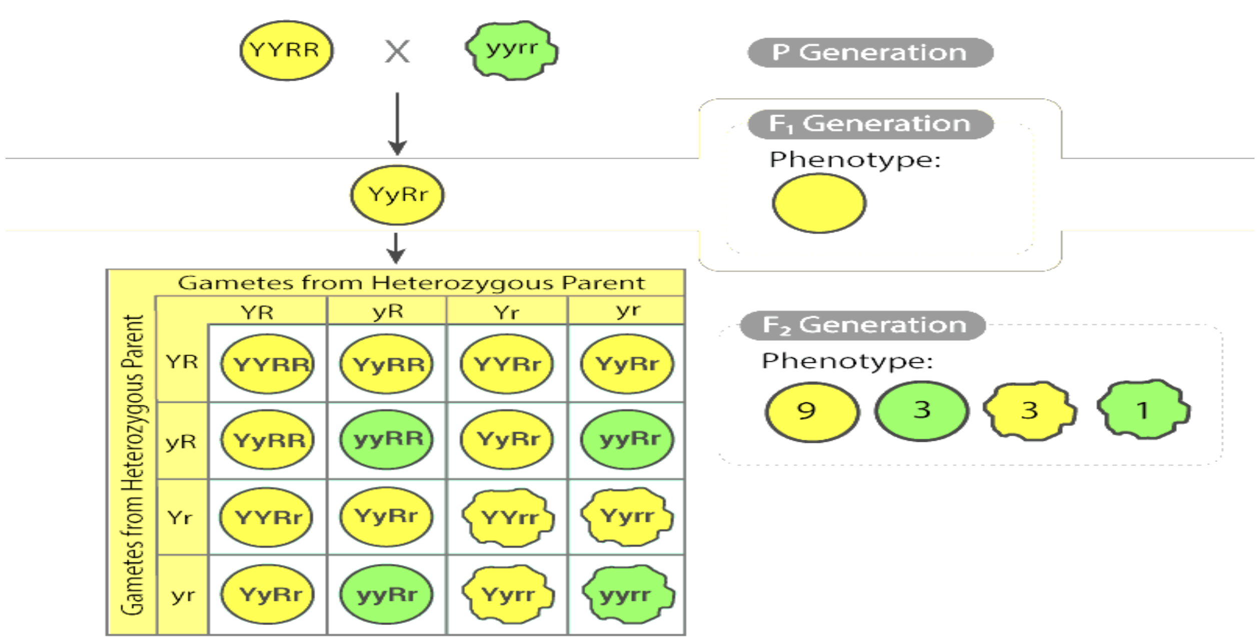
Mendel's second law and dihybrid inheritance

Is the inheritance of **two characteristics**, each controlled by a different gene (allele) at a different locus

Mendel decided to cross homozygous pea plants with the **two dominant characters (round and yellow seeds)** with homozygous plants with the **two recessive characters (wrinkled and green).** He found that all the F_1, generation had round yellow seeds. He then self-pollinated flowers from the F_1; (sometimes called 'selfing'). When he collected and counted the seeds he found the following:

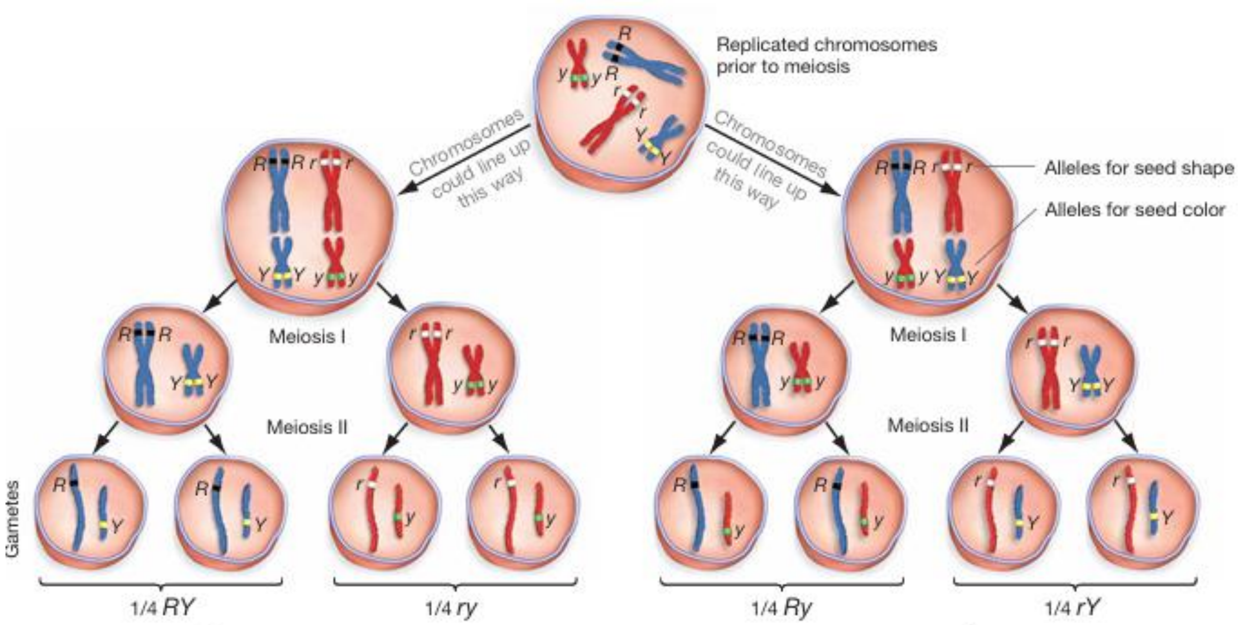
	round yellow	round green	wrinkled yellow	wrinkled green
	315	108	101	32
approx. ratio	9:	3:	3:	1

explanations for his results are shown below using genetic symbols



The results of the dihybrid cross led Mendel to formulate his **second law of inheritance:** ***the law of independent assortment***.

Either of a pair of alleles (factors) may combine randomly with either of another pair.



PRINCIPLE OF INDEPENDENT ASSORTMENT: The genes for seed shape and seed color assort independently, because they are located on different chromosomes.

Think!!!!!!

► A dihybrid test cross

Mendel wanted to cross F_1 plants from his dihybrid cross. He wanted to be sure that the heterozygous plants could produce four different types of gametes.
As with a monohybrid test cross, he crossed the F_1 plants with homozygous recessive plants that produced wrinkled green seeds. These plants were **double recessive** because they were recessive for both characters. Here are Mendel's results:

gametes	(RY)	(Ry)	(rY)	(ry)
(ry)	RrYy	Rryy	rrYy	rryy

- round yellow 57
- round green 51
- wrinkled yellow 49
- wrinkled green 53

The results were close to the ratio of 1:1:1:1 that we would expect to get from the cross.

Summary

(1) Each characteristic of an organism is controlled by a pair of alleles.

(2) If an organism has two unlike alleles for a given characteristic, one may be expressed (the dominant allele) to the total exclusion of the other (the recessive allele).

(3) During meiosis each pair of alleles separates (segregates) and each gamete receives one of each pair of alleles (the principle of segregation).

(4) During gamete formation in each sex, either one of a pair alleles may enter the same gamete cell of (combine randomly) with either one of another pair (the principle of independent assortment).

(5) Each allele is transmitted from generation to generation as a discrete unchanging unit.

(6) Each organism inherits one allele (for each characteristic) from each parent.

A summary of the similarities between events occurring during meiosis and fertilization and Mendel's hypotheses

Meiosis and fertilisation	*Mendel's hypotheses*
Diploid cells contain *pairs* of chromosomes(homologous chromosomes)	Characteristics are controlled by *pairs* of factors
Homologous chromosomes *separate* during meiosis	Pairs of factors *separate* during gamete formation
One homologous chromosome passes into each gamete cell	Each gamete receives *one* factor
Only the *nucleus* of the male gamete fuses with the egg cell nucleus	Factors are transmitted from generation to generation as *discrete units*
Homologous pairs of chromosomes are restored at fertilisation, each gamete (♂ and ♀) contributing *one* homologous chromosome	Each organism inherits *one* factor from each parent

GENE INTERACTIONS

d) *Examine different forms of allele interactions (autosomal linkage, multiple alleles, codominance and incomplete dominance), including their examples and influence on phenotypic expression*

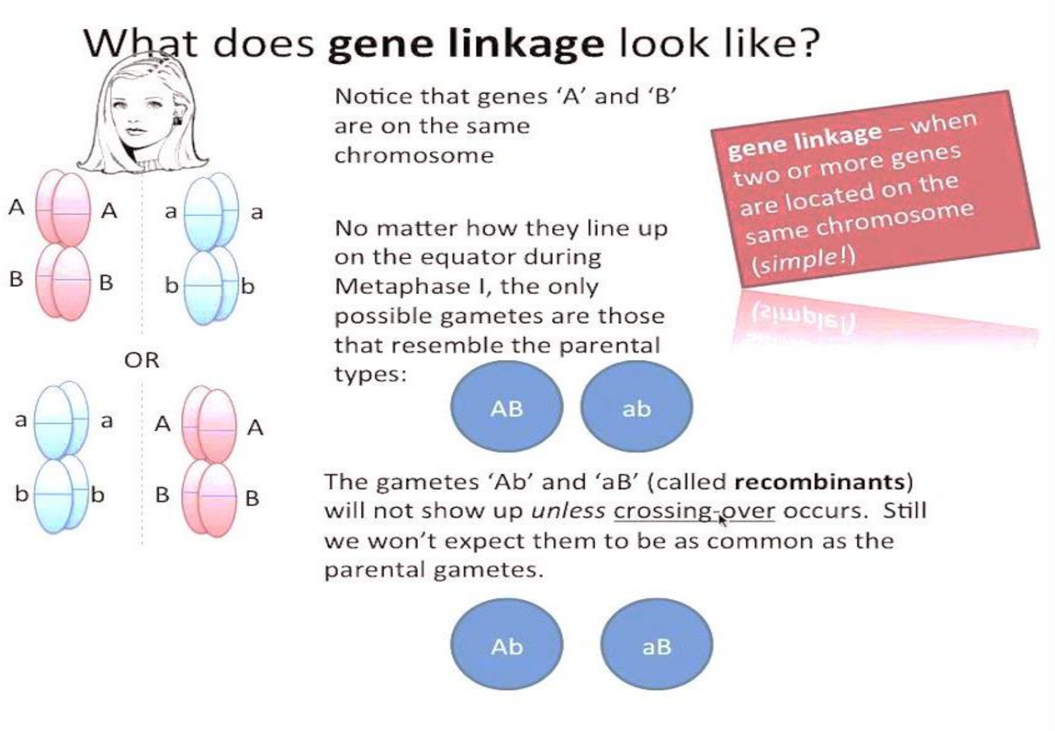
Linkage

Linkage is the tendency of particular **alleles of different genes to be inherited together**. Linkage is seen when genes are on the same chromosome. Notice that the terms **linkage and sex-linkage** have different meanings. *If genes are linked, it means that they are located on the same chromosome.* If *a gene is sex-linked, it means that it is located on a sex chromosome* but says nothing about its location relative to other genes. **QUICK TIP**

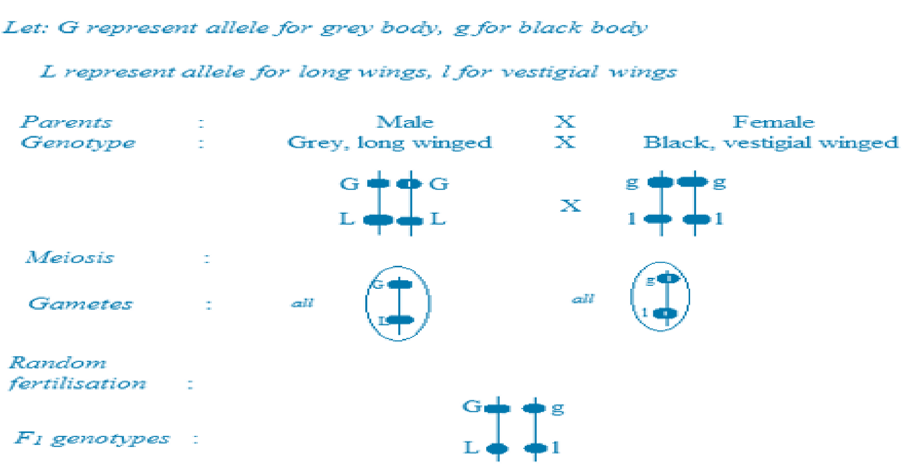
*Genes situated on the **same chromosome** are said to be **linked**. All genes on a single chromosome form a linkage group and usually pass into the same gamete and are inherited together.*

Therefore linkage is a condition where genes are located on the same chromosome and thus inherited as a unit.

As a result of this, genes belonging to the same linkage group usually do not show independent assortment.

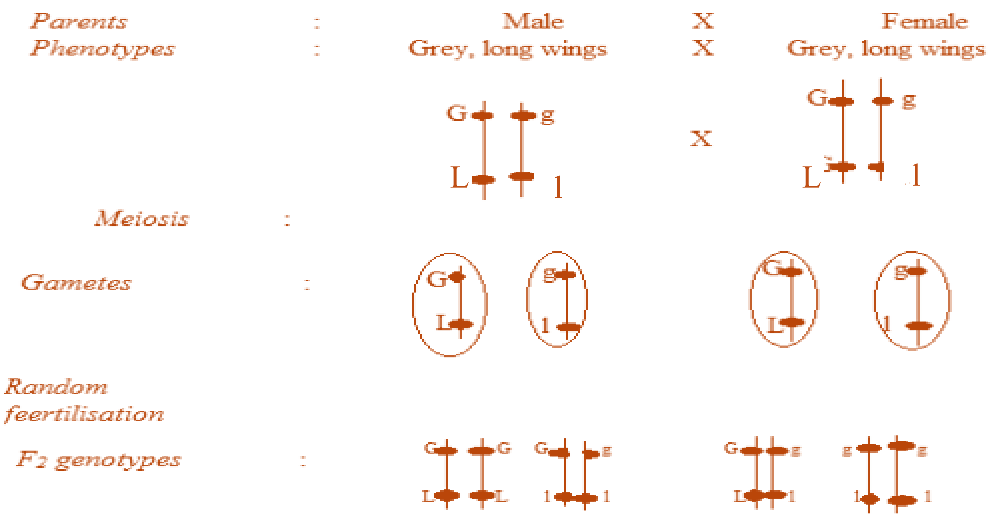


In drosophila, the alleles for grey body and long wings are dominant to those for black body and vestigial wings respectively. If pure breeding grey bodied long winged drosophilae are crossed with pure breeding black bodied vestigial winged drosophila; all in the F1 are grey with long wings. Surprisingly in the F2, a 3:1 ratio of grey long winged and black vestigial winged (the original parental) phenotypes are obtained as follows



F1 phenotypes: all grey with long wings

Selfing F_1Obtaining F_2 generation:



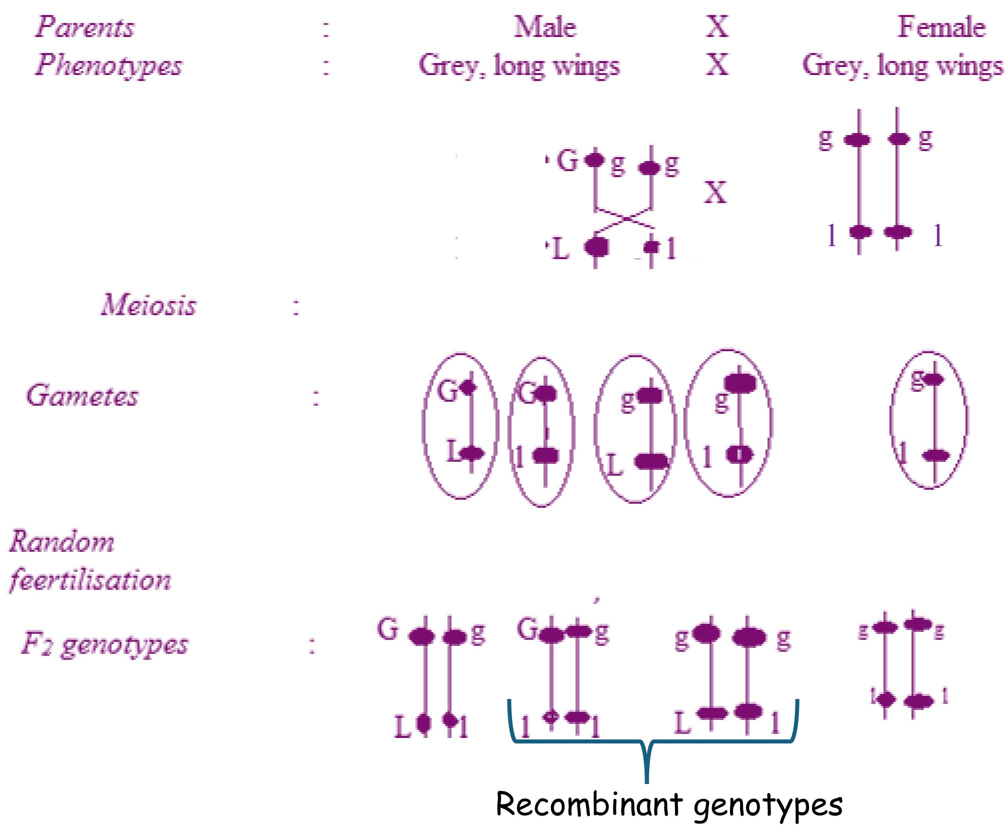
Phenotypic ratio is 3 grey long winged: 1 black vestigial winged.

CROSSING OVER

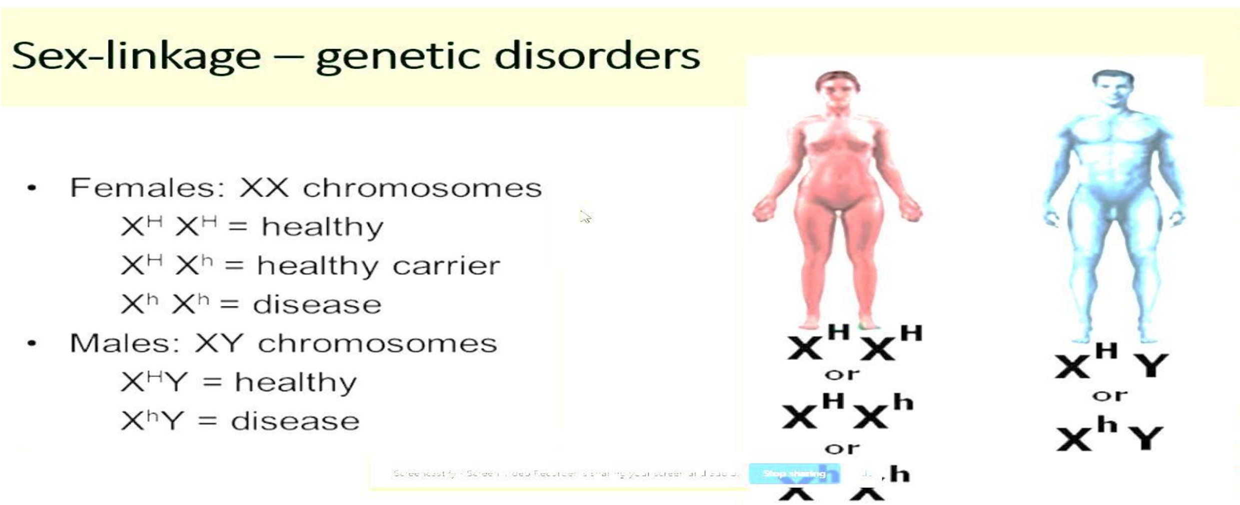
- Crossing over refers to **the interchange of parts between non-sister chromatids of homologus chromosomes during meiotic prophase (pachytene).** In other words, crossing over results from **exchange of genetic material between non-sister chromatids involving breakage and reunion at precise point.**
- The term crossing over was first used **by Morgan and Cattell in 1912.**

To his disappointment; even after performing the test cross about the above cross several times, Morgan never obtained the predicted outcomes. He instead obtained approximately equal numbers of the parental phenotypes with significantly few recombinant phenotypes also in approximately equal numbers as summerised below.

i. 41.5% grey, long winged
ii. 41.5% black, vestigial winged
iii. 8.5% grey, vestigial winged
iv. 8.5% black, long winged



Sex linkage refers to the carrying of genes on the sex chromosomes. These genes determine body characters and have nothing to do with sex. The X chromosome carries many such genes, the Y chromosome has very few. Features linked on the Y chromosome will only arise in the heterogametic (XY) sex, i.e. males in mammals, females in birds. Features linked on the X chromosome may arise in either sex.

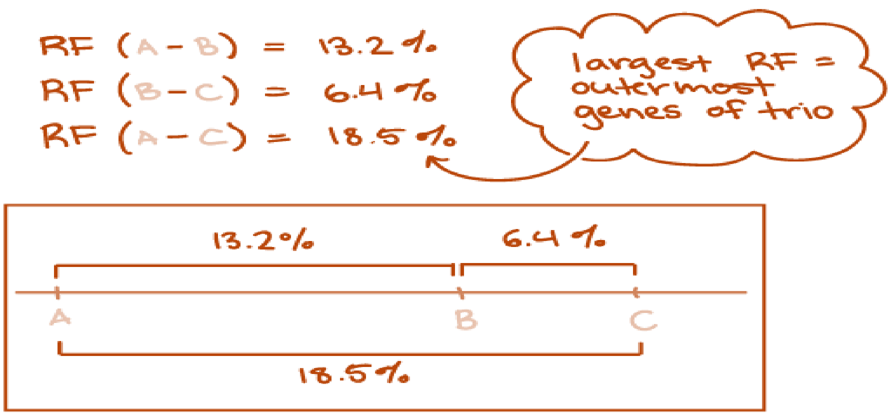


Cross over values and gene maps

During crossing over, the frequency of crossovers which take place was found to be dependent on the distribution and arrangement of chromosomes. This is given by the cross over value/frequency aka recombination frequency. This is calculated as a percentage ration of recombinants to the total number of offsprings.

$$CoV = \frac{Number\ of\ recombinants}{Total\ number\ of\ offsprings} x100$$

The COV also indicates the relative distance between linked genes and the possibility of successful crossing over during meiosis, in the above case the distance between adjacent genes is 17 units. These values can also be used to position genes along the chromosome a process called gene mapping.



Multiple alleles

These are three or more forms of the same gene occurring at the same locus. These alleles control the same contrasting character.

An example is inheritance of blood A, B, AB, and O blood group system in humans.

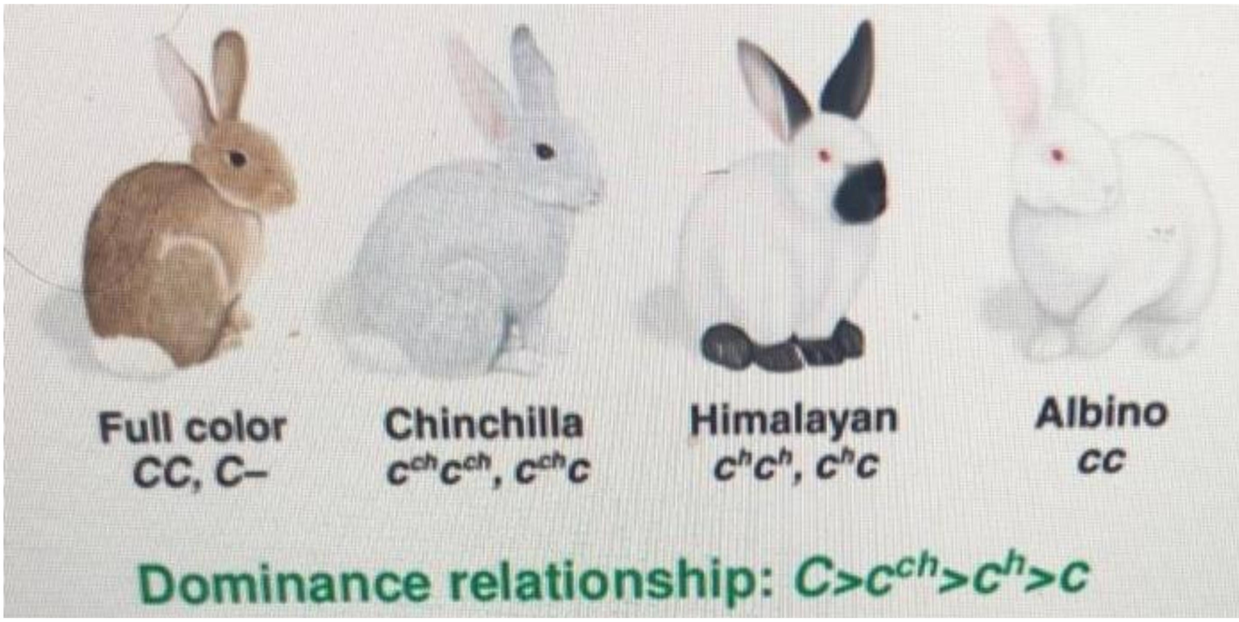
A and B are codominant while o is recessive

Blood Type	Genotype	
A	i^Ai i^Ai^A	AA AO
B	i^Bi i^Bi^B	BB BO
AB	i^Ai^B	AB
O	ii	OO

Blood Groups				
Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
A	I^AI^A or I^Ai	A	A, AB	A, O
B	I^BI^B or I^Bi	B	B, AB	B, O
AB	I^AI^B	A and B	AB	A, B, AB, O
O	ii	none	A, B, AB, O	O

Other examples include;

Fur color inheritance in rabbits



Codominance and incomplete dominance; covered in previous activities.

POPULATION GENETICS

Population genetics is the study of genetic variation within and between populations, and how this variation changes over time due to evolutionary forces like natural selection, mutation, genetic drift, and gene flow.

The **Hardy-Weinberg Principle** states that in a large, randomly mating population **not affected by mutation, migration, natural selection, or genetic drift**, the **allele and genotype frequencies remain constant** from generation to generation.

Term	Definition
Gene Pool	The complete set of alleles present in a population.
Allele	The proportion of a specific allele among all alleles of

Frequency	a gene in a population.
Genotype Frequency	The proportion of a specific genotype among all individuals in a population.
Genetic Equilibrium	A condition where the allele and genotype frequencies in a population remain unchanged over generations.
Evolution	A change in the genetic composition (allele frequencies) of a population over time.

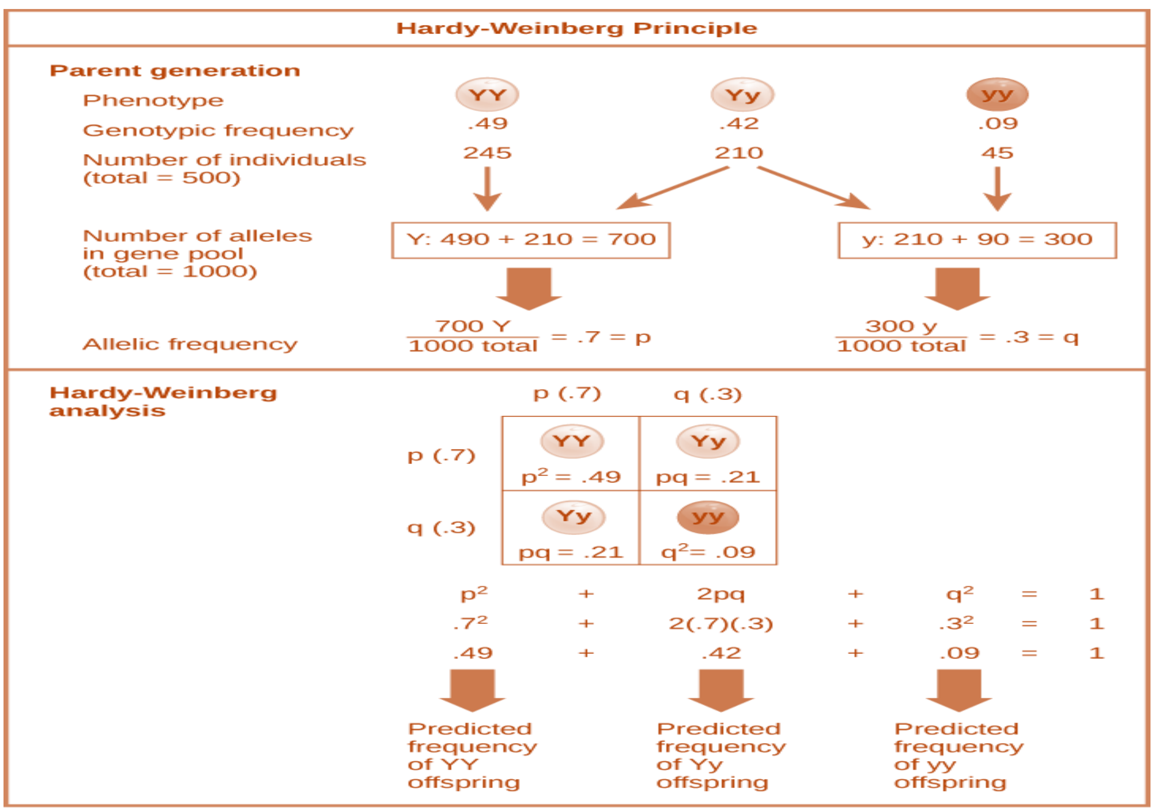
Hardy-Weinberg Equations

$p^2 + 2pq + q^2 = 1$

$p + q = 1$

p = frequency of the dominant allele in a population

q = frequency of the recessive allele in a population



Conditions for Hardy-Weinberg Equilibrium	
Condition	**Consequence if Condition Does Not Hold**
1. No mutations	The gene pool is modified if mutations occur or if entire genes are deleted or duplicated.
2. Random mating	If individuals mate within a subset of the population, such as near neighbors or close relatives (inbreeding), random mixing of gametes does not occur and genotype frequencies change.
3. No natural selection	Allele frequencies change when individuals with different genotypes show consistent differences in their survival or reproductive success.
4. Extremely large population size	In small populations, allele frequencies fluctuate by chance over time (a process called genetic drift).
5. No gene flow	By moving alleles into or out of populations, gene flow can alter allele frequencies.

Use you note book

A population of 1,000 butterflies lives on an isolated island. The color of their wings is controlled by a single gene with two alleles:

- B (dominant) = blue wings
- b (recessive) = white wings

In this population, 360 butterflies have white wings.

Assume:

- The population is large.
- There is random mating.
- No migration, mutation, or natural selection occurs (initially).

 Tasks:

1. Is this population in Hardy-Weinberg equilibrium?

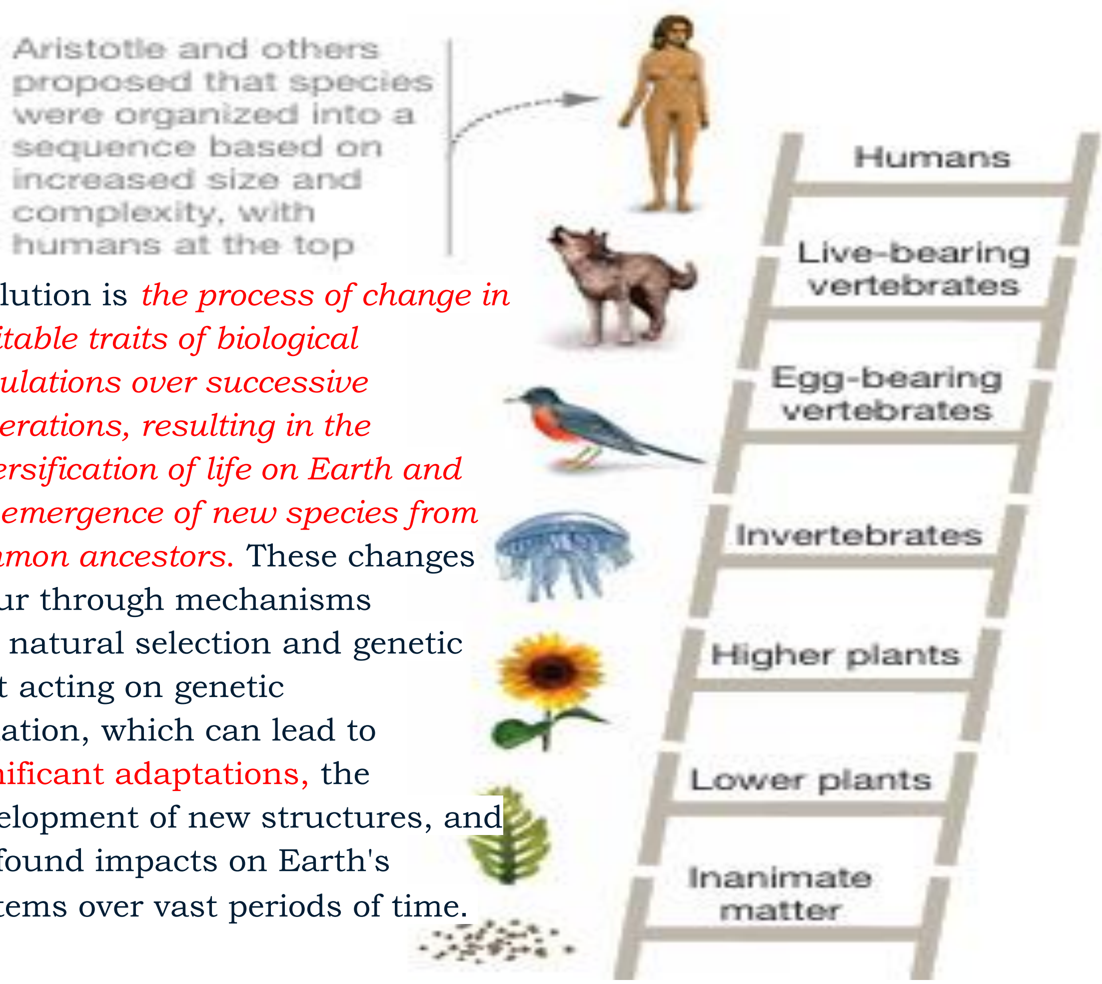
2. What are the allele frequencies of B and b?

3. What are the expected genotype frequencies (BB, Bb, bb) under equilibrium?

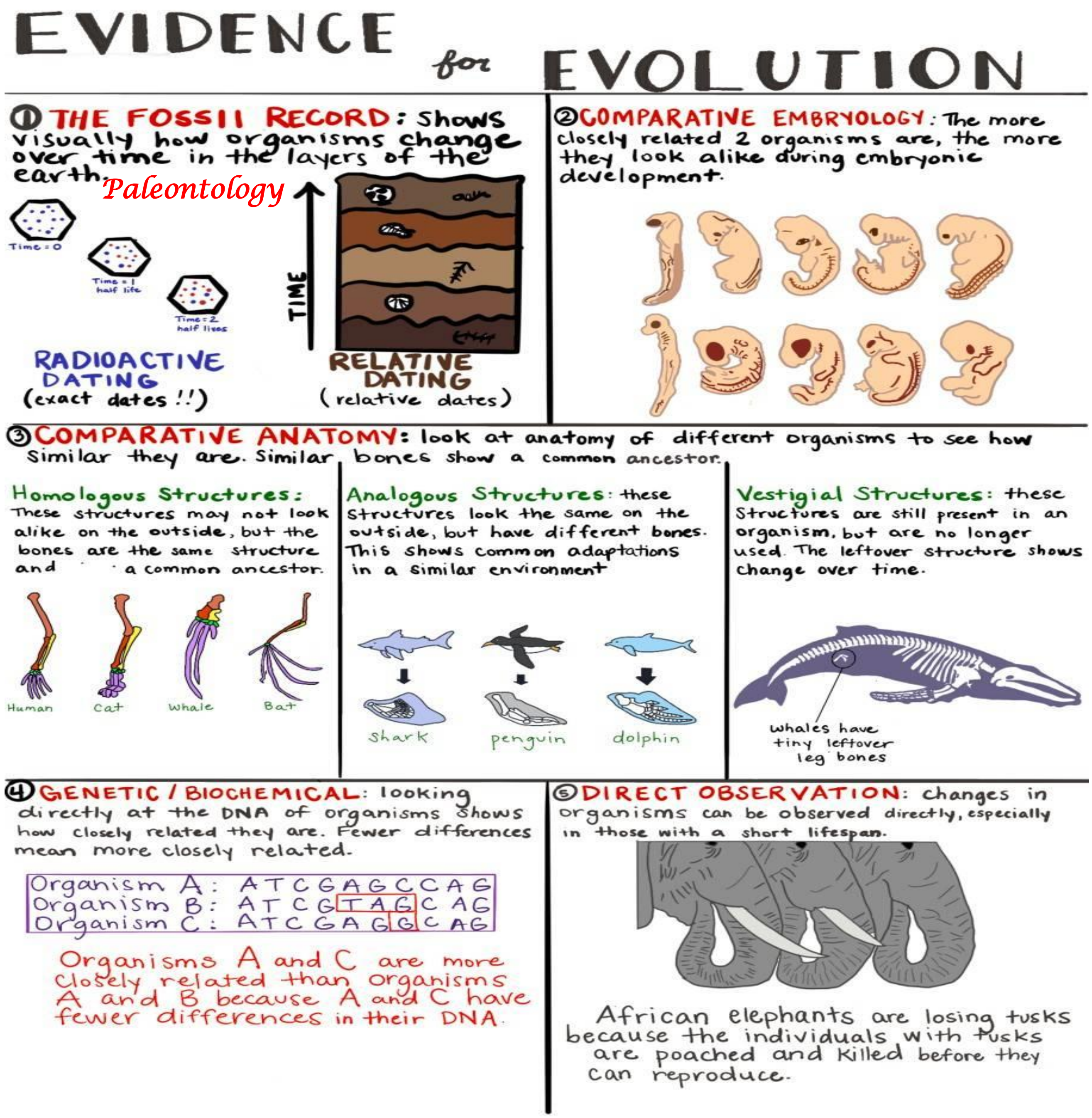
4. What would cause this equilibrium to be disrupted?

EVOLUTION

***e)* Key learning outcome; Analyze evolutionary advancements in key life processes (circulation, reproduction, gaseous exchange, coordination, movement, and excretion), as well as their suitability for survival across different species**



Evolution is *the process of change in heritable traits of biological populations over successive generations, resulting in the diversification of life on Earth and the emergence of new species from common ancestors.* These changes occur through mechanisms like natural selection and genetic drift acting on genetic variation, which can lead to significant adaptations, the development of new structures, and profound impacts on Earth's systems over vast periods of time.



Use your note book;

Discuss the theories of evolution and their limitations

Change through time continues and can be measured directly. Evidence from the fossil record and living species indicates that life is ancient, that species have changed through the course of Earth's history, and that species continue to change. The take-home message is that species are dynamic—not static, unchanging, and fixed types, as claimed by Plato, Aristotle, and advocates of special creation.

As organisms change to give rise to new species they advance; with changes in their anatomy to occupy various ecological significance; though some get extinct.

The advancements are summarized in the table below.

1. Circulation

Group	Advancement	Adaptation Benefit
Single-celled organisms **(e.g., Amoeba)**	No circulatory system; diffusion suffices	Small size allows diffusion of gases/nutrients directly across membrane
Invertebrates **(e.g., insects)**	Open circulatory system	Low energy cost, sufficient for small, active organisms
Fish	Single-loop closed circulatory system (2-chambered heart)	Efficient blood flow for aquatic gas exchange (gills)
Amphibians	Double-loop, 3-chambered heart	Allows partial separation of oxygenated/deoxygenated blood
Mammals and birds	Double-loop, 4-chambered heart	Complete separation of oxygenated and deoxygenated

		blood supports high metabolic demand

2. Reproduction

Group	Advancement	Adaptation Benefit
Bacteria	Binary fission (asexual)	Rapid reproduction in favorable conditions
Amphibians	External fertilization	Requires moist environment; less parental care
Reptiles	Internal fertilization, amniotic egg	Adapted for dry land, protects embryo
Birds	Amniotic egg with hard shell	Protection from desiccation, enables terrestrial reproduction
Mammals	Internal fertilization, live birth (vivipary), parental care	High survival of fewer offspring, development inside body ensures protection

3. Gaseous Exchange (Respiration)

Group	Advancement	Adaptation Benefit
Single-celled organisms	Diffusion across membrane	Effective for small size, aquatic environments
Insects	Tracheal system (spiracles + tubes)	Delivers oxygen directly to tissues, supports high activity
Fish	Gills with countercurrent exchange	Maximizes O_2 uptake in water
Amphibians	Skin + lungs	Dual respiration allows survival in water and on land
Mammals	Lungs with alveoli and diaphragm	Large surface area for gas exchange, supports high

		metabolism

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4. Coordination (Nervous and Hormonal)

Group	Advancement	Adaptation Benefit
Cnidarians **(e.g., jellyfish)**	Nerve net	Basic movement and response to stimuli
Arthropods	Centralized brain and nerve cords	Rapid response, complex behaviors
Vertebrates	Brain with specialized regions + endocrine system	Advanced processing, memory, learning, and homeostasis

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5. Movement (Locomotion)

Group	Advancement	Adaptation Benefit
Protozoa	Pseudopodia, cilia, flagella	Locomotion in aquatic environments
Annelids	Segmented body and muscles	Coordinated movement via hydrostatic skeleton
Arthropods	Jointed limbs, exoskeleton	Precise and rapid movement, body protection
Fish	Fins and streamlined body	Efficient swimming
Birds	Wings, lightweight skeleton	Adaptation to flight
Mammals	Limbs specialized for running, climbing, swimming, etc.	High adaptability to diverse environments

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6. Excretion

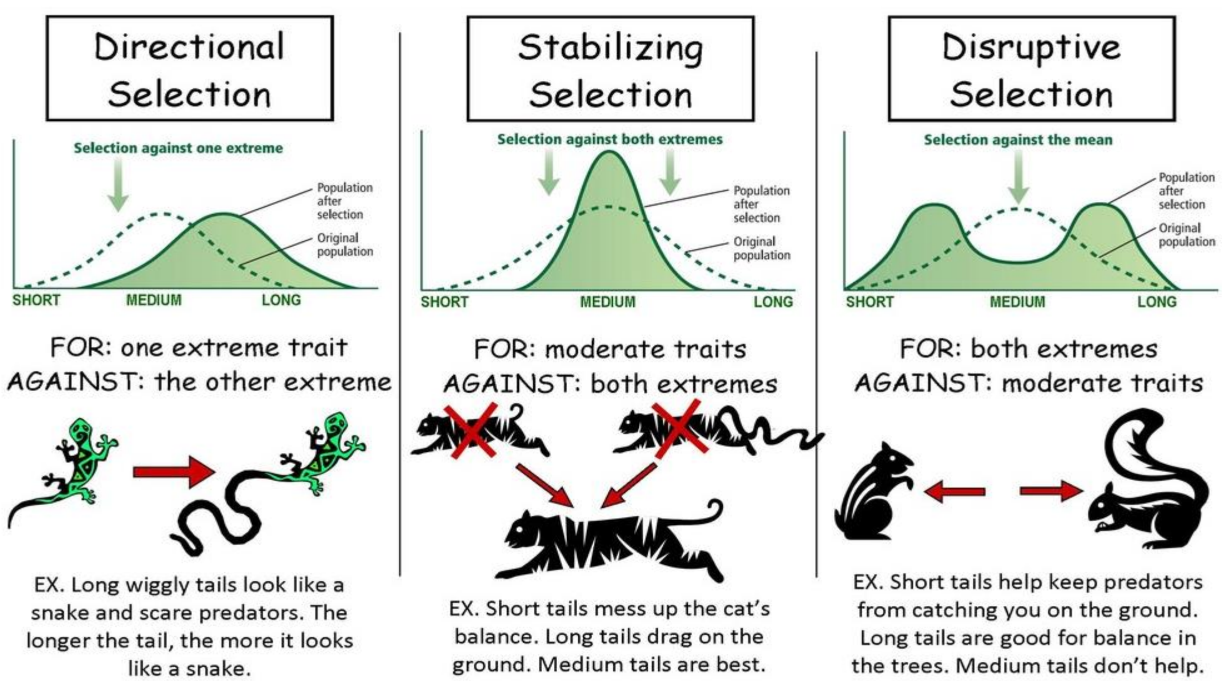
Group	Advancement	Adaptation Benefit
Protozoa	Diffusion	Sufficient for small organisms
Flatworms	Flame cells	Removes waste in simple

	(protonephridia)	multicellular organisms
Insects	Malpighian tubules	Conserves water in dry environments
Fish	Kidneys that excrete ammonia	Ammonia diluted in water directly
Mammals	Kidneys that excrete urea	Less toxic than ammonia, conserves water

Evolution occurs due to natural selection; in which organisms with traits that confer them a selective advantage survive and transfer their traits to the next generation on expense of less adapted organisms;

Evolution by natural selection occurs when heritable variation leads to differential success in survival and reproduction. If certain alleles are associated with the favored phenotypes, they increase in frequency while other alleles decrease in frequency. The result is evolution—a violation of the assumptions of the Hardy Weinberg model.

There three types of selection;

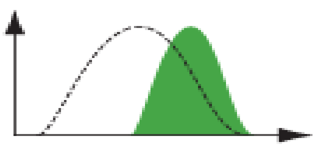
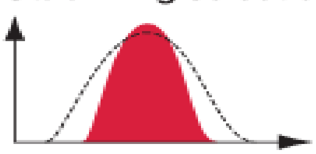
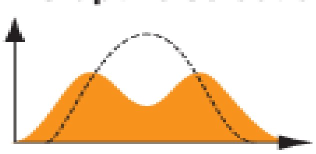
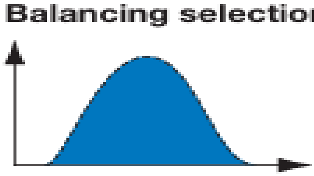


Key notes

Directional selection tends to reduce the genetic diversity of populations; also called purifying selection; as disadvantageous alleles decline in frequency

Disruptive selection is important because it sometimes plays a part in speciation, or the formation of new species; increased genetic variation.

Stabilizing selection reduces extent evolution, as it maintains the population's genetic stability - reducing genetic variation.

Mode of Selection	Effect on Phenotype	Effect on Genetic Variation
Directional selection 	Favors one extreme phenotype, causing the average phenotype in the population to change in one direction.	Genetic variation is reduced.
Stabilizing selection 	Favors phenotypes near the middle of the range of phenotypic variation, maintaining average phenotype.	Genetic variation is reduced.
Disruptive selection 	Favors extreme phenotypes at both ends of the range of phenotypic variation.	Genetic variation is increased.
Balancing selection 	No single phenotype is favored in all populations of a species at all times.	Genetic variation is maintained.

SPECIATION & RESISTANCE

e) Assess speciation and resistance, mechanisms driving them, and factors contributing to extinction events, through comparison of historical and contemporary examples.

Speciation is the process by which one or more species arise from previously existing species.

A single species may give rise different to new species (***intraspecific speciation***), or, as is common in many flowering plants, two species may give rise to a new species (***interspecific hybridisation***).

If intraspecific speciation occurs whilst the populations are separated it is termed ***allopatric speciation***. If the process occurs whilst the populations are occupying the same geographical area it is called ***sympatric speciation.***

Speciation will only occur as a result of the formation of barriers which lead to reproductive isolation between members of the population. Reproductive isolation is brought by some form of what the geneticist Theodosius Dobzhansky called ***isolating mechanisms.***

An isolating mechanism *is means of producing and maintaining reproductive isolation within a population. As shown below*

Pre-zygotic Isolating Mechanisms		Example
Temporal	Occurs when two species mate at different times of year	Frogs live in same pond but breed during different seasons (summer vs spring)
Ecological	Occurs when two species occupy different habitats	Lions and tigers can potentially interbreed, but usually occupy different habitats
Behavioural	Occurs when two species have different courtship behaviours	Certain groups of birds will only respond to species-specific mating calls
Mechanical	Occurs when physical differences prevent copulation / pollination	Certain breeds of dog are morphologically incapable of mating due to size
Post-zygotic Isolating Mechanisms		**Examples**
Hybrid Inviability	Hybrids are produced but fail to develop to reproductive maturity	Certain types of frogs form hybrid tadpoles that die before they can become a frog
Hybrid Infertility	Hybrids fail to produce functional gametes (sterility)	Mules are sterile hybrids resulting from mating between a horse and a donkey
Hybrid Breakdown	F_1 hybrids are fertile, but F_2 generation fails to develop properly	The offspring of hybrid copepods have less potential for survival or reproduction

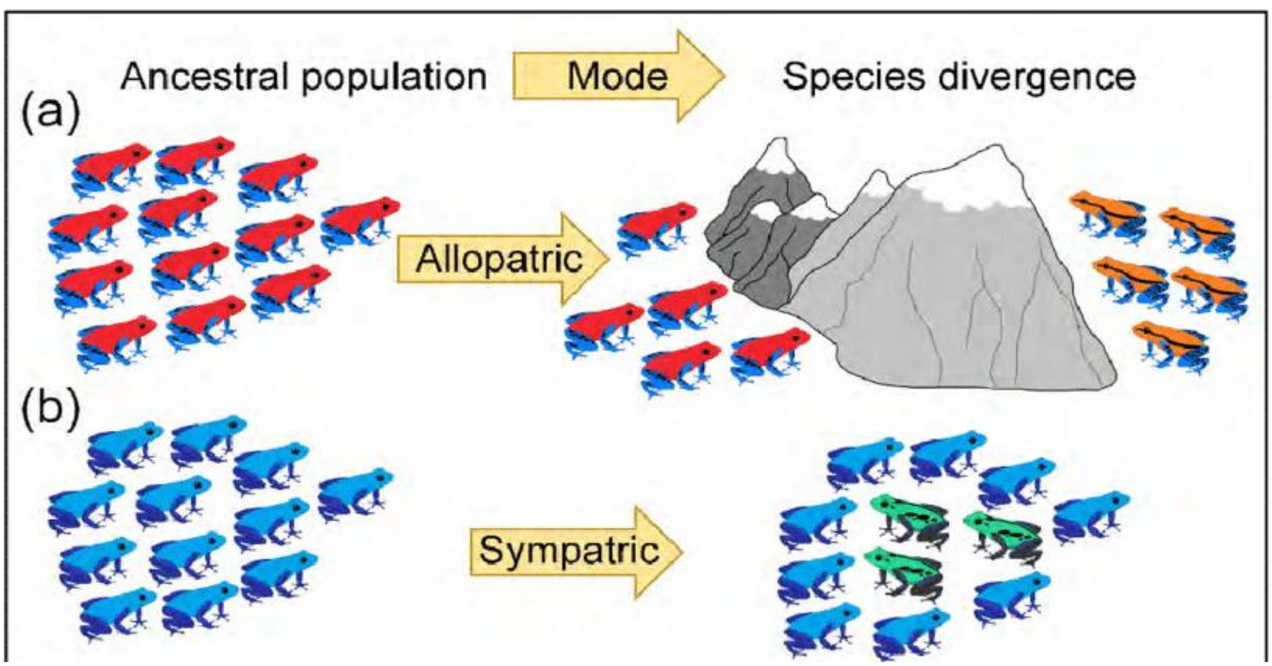
Types of speciation

Allopatric speciation involves **geographic isolation**, where a physical barrier such a mountain or river divides a population, leading to independent evolution and the formation of new species,

Sympatric speciation occurs within the same geographic area and involves a new species diverging due to ecological, behavioral, or genetic factors like polyploidy

Two main modes of speciation:

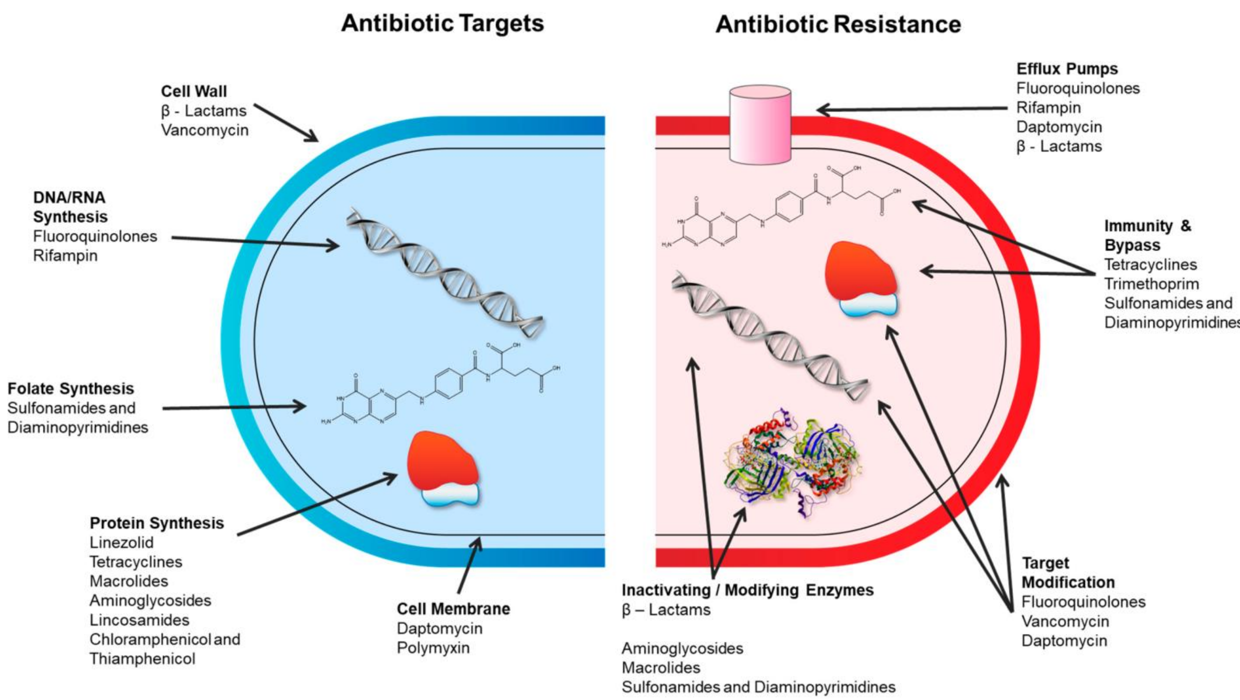
Allopatric Speciation *"other" "homeland"*	Sympatric Speciation *"together" "homeland"*
Geographically isolated populations	Overlapping populations within home range
• Caused by geologic events or processes • Evolves by natural selection & genetic drift	Gene flow between subpopulations blocked by: • polyploidy • sexual selection • habitat differentiation
Eg. Squirrels on N/S rims of Grand Canyon	Eg. polyploidy in crops (oats, cotton, potatoes, wheat)



Antimicrobial and Pesticide Resistance

Overview

- **Antimicrobial resistance (AMR)**: The ability of microorganisms (bacteria, fungi, viruses, and parasites) to resist the effects of drugs that once killed them or inhibited their growth.
- **Pesticide resistance**: The ability of pests (insects, weeds, fungi, etc.) to survive exposure to chemicals designed to kill them.
- Both are **evolutionary processes** driven by **natural selection** in response to environmental pressures (antibiotics or pesticides).
- These resistances can lead to **divergent evolution** and potentially to **speciation** under the right conditions.



Evolutionary Basis

1. Variation in Populations

- Populations of microorganisms or pests exhibit **genetic variation**.
- Mutations occur naturally and randomly in DNA.
- Some mutations may **confer resistance** to a particular drug or chemical.

2. Selection Pressure

- Exposure to antimicrobials or pesticides acts as a **strong selective pressure**.

- **Sensitive individuals** die, while **resistant individuals survive** and reproduce.
- Over time, the **frequency of resistant alleles increases** in the population.

3. Adaptation

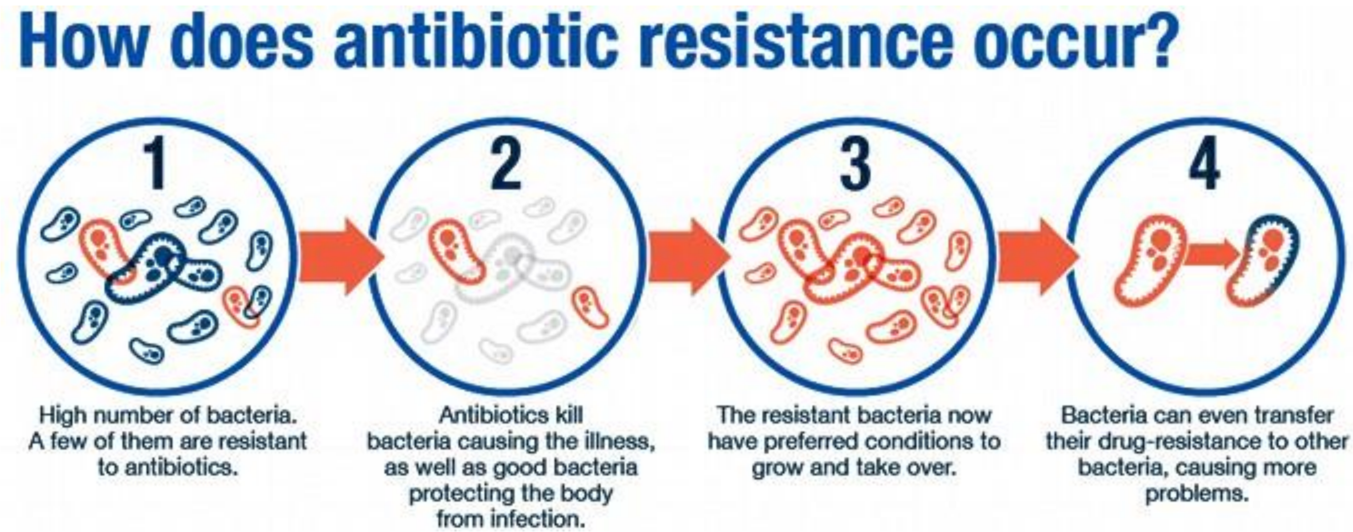
- The population becomes adapted to the environment where the antimicrobial or pesticide is present.
- This process is **directional selection**: selection favors one extreme phenotype (resistance).

4. Rapid Evolution

- Microbes reproduce rapidly (short generation times), allowing **quick evolution**.
- Resistance can evolve in a matter of **days to weeks** in microbes and **generations** in pests.

Mechanisms of Resistance

In Microorganisms (Antimicrobials)



- **Enzymatic degradation** of the drug (e.g., β-lactamase in bacteria).
- **Efflux pumps** to remove drugs from the cell.
- **Target modification** (mutation in ribosomes, enzymes, etc.).
- **Reduced permeability** (altered membrane proteins).
- **Horizontal gene transfer (HGT)**:
  - Conjugation (plasmids)
  - Transformation (uptake of DNA)
  - Transduction (via bacteriophages)

In Pests (Pesticides)

- **Metabolic resistance**: Detoxifying enzymes degrade the pesticide.
- **Target-site resistance**: Mutation in the protein targeted by the pesticide.
- **Behavioral resistance**: Avoidance of pesticide-treated areas.
- **Reduced penetration**: Thicker cuticle or altered entry points.

Resistance and Speciation

1. Reproductive Isolation

- Resistant populations may become **reproductively isolated** from the original population.
- Especially in microbes where **resistance genes** can cause **genetic incompatibility** over time.

2. Ecological Isolation

- Resistant organisms might inhabit **different ecological niches** (e.g., hospital vs. community strains of MRSA-**Methicillin-Resistant Staphylococcus Aureus**).
- This leads to **divergent evolution** and possibly **sympatric speciation**.

3. Genetic Divergence

- Accumulation of **resistance-associated mutations** can reduce gene flow between populations.
- In pests, pesticide-resistant populations may evolve distinct behaviors, mating times, or preferences.

4. Artificial Selection and Speciation

- Human use of antimicrobials and pesticides acts as **artificial selection**.
- Over long timescales, this can contribute to the emergence of **new strains or species**.

Examples

Antimicrobial Resistance

- **MRSA (Methicillin-resistant Staphylococcus aureus)**:
  - Resistance to β-lactam antibiotics.
  - Hospital-associated vs. community-associated strains → divergence.
- **Multi-drug resistant TB (MDR-TB)**:
  - Resistance to isoniazid and rifampicin.

- Often due to poor treatment compliance and rapid bacterial evolution.

 Pesticide Resistance

- **Mosquitoes (Anopheles)**:
  - Resistance to DDT and pyrethroids (used in malaria control).
- **Colorado potato beetle**:
  - Resistance to over 50 different insecticides.
- **Herbicide-resistant weeds** (e.g., glyphosate-resistant *Amaranthus* species):
  - A growing issue in agriculture due to over-reliance on single herbicides.

Implications

 Scientific

- Demonstrates **evolution in action**.
- Provides real-world evidence for **natural selection**.
- Can inform **evolutionary theory** and studies on **adaptive radiation** and **speciation**.

 Public Health

- AMR is a global health crisis.
- Leads to **longer hospital stays**, **higher medical costs**, and **increased mortality**.

 Agriculture

- Pesticide resistance reduces crop yields.
- Increases dependency on **new or stronger chemicals**, creating a cycle of resistance.

 Evolutionary Arms Race

- Continuous development of new drugs/pesticides → organisms evolve resistance → new development required.
- Ongoing **co-evolutionary dynamic**.

Mitigation Strategies

In Healthcare:

- Rational use of antibiotics (antibiotic stewardship- coordinated efforts by healthcare teams to ensure antibiotics are used appropriately, which means prescribing the correct drug, dose, route, and duration for the right patient and condition.).
- Development of new antimicrobial agents.

- Infection control measures.

In Agriculture:

- Integrated pest management (IPM).
- Crop rotation and biological control.
- Use of pesticide mixtures or alternation to prevent resistance buildup.

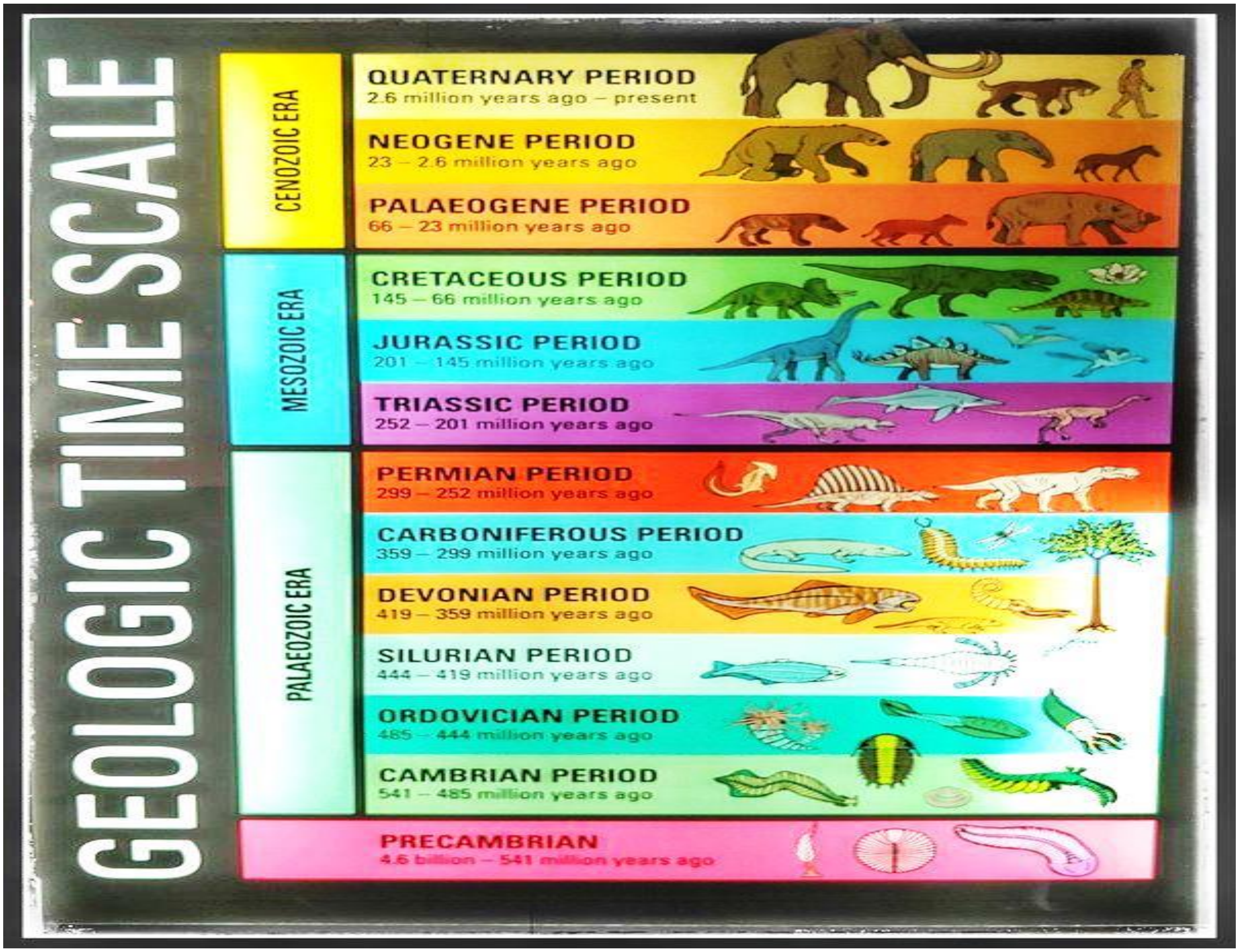
EXTINCTION AND EVOLUTION

During speciation some species get extinct as new ones arise;

Amass extinction refers to the rapid extinction of a large number of lineages scattered throughout the tree of life. More specifically, a mass extinction occurs when at **least 60 percent of the species present** are wiped out within 1 million years.

Background extinction refers to the lower, average rate of extinction observed when a mass extinction is not occurring.

Extinction marks the end of one geological period to another; ***Geological periods*** *are divisions* of the Earth's history, which is organized into a geologic time scale based on major **geological and biological events**. The largest units are **eons**, which are divided into eras, periods, and epochs. Major eras include the Precambrian (Hadean, Archean, Proterozoic eons), Paleozoic, Mesozoic, and Cenozoic, each subdivided into periods like the ***Cambrian, Jurassic, and Quaternary***



Factors leading to extinction

1. Climate Change

Historical Example:

- **Permian-Triassic Extinction (c. 252 million years ago)**

- The most severe extinction event in Earth's history.
- Likely caused by massive volcanic eruptions (Siberian Traps) that released enormous amounts of CO_2 and methane.
- Resulted in ***global warming***, ocean acidification, and anoxia (lack of oxygen in oceans).

Contemporary Example:

- **Current Biodiversity Crisis (Holocene/Anthropocene extinction)**
  - Driven in part by anthropogenic climate change due to greenhouse gas emissions from fossil fuel use, deforestation, and agriculture.
  - Affects species' habitats, migration patterns, and survival rates (e.g., coral bleaching, polar bear habitat loss).

Comparison:

- Both events involve rapid climate shifts, but modern changes are human-driven and occurring over decades, not millennia.

- Current extinction risks are amplified by additional human activities.

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2. Habitat Loss and Fragmentation

Historical Example:

- Not a primary cause in mass extinctions, but natural habitat changes due to continental drift, glaciation, or sea-level changes did contribute to localized extinctions.

Contemporary Example:

- **Amazon Rainforest Deforestation**
  - Large-scale land conversion for agriculture and logging.
  - Leads to the direct loss of biodiversity and disruption of ecosystems.

Comparison:

- Historically slower and more natural; today, habitat loss is rapid, extensive, and driven largely by human economic activities. **IDENTIFY THEM**

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3. Invasive Species

Historical Example:

- After the formation of land bridges (e.g., during the Great American Biotic Interchange), new species introductions disrupted ecosystems and led to extinctions.

Contemporary Example:

- **Brown tree snake in Guam**
- 
    - Accidentally introduced; caused the extinction of several native bird species.
- **Global trade and travel** allow species to be introduced far outside their native ranges.

Comparison:

- Invasions were previously linked to geological or climatic events; now, human transportation systems accelerate the spread of invasive species globally.- Introduction of Nile perch in Lake victoria;

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4. Overexploitation

Historical Example:

- **Late Pleistocene Extinctions (c. 50,000–10,000 years ago)**

- Megafaunal extinctions possibly linked to early human hunting (e.g., woolly mammoths, saber-toothed cats).

Contemporary Example:

- **Overfishing of marine species (e.g., Atlantic cod, bluefin tuna)**
  - Poaching and illegal wildlife trade (e.g., rhinoceroses, elephants for ivory and horns).
  - Unsustainable logging and plant harvesting.

Comparison:

- Overexploitation is one of the earliest human-driven extinction factors and remains highly relevant today, but it is now intensified by global demand and industrial-scale operations.

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5. Catastrophic Events

Historical Example:

- **Cretaceous-Paleogene (K-Pg) Extinction (c. 66 million years ago)**
  - Likely caused by a massive asteroid impact (Chicxulub crater) and subsequent global environmental disruptions (e.g., "nuclear winter" effect).

Contemporary Example:

- While natural catastrophes still occur (volcanic eruptions, tsunamis), human activities can cause "unnatural catastrophes":
  - **Oil spills**, **nuclear accidents** (e.g., Chernobyl, Fukushima, Hiroshima nuclear impact), and **chemical contamination**.

Comparison:

- Historic catastrophes were natural and rare; modern ones can be both natural and anthropogenic, and while often smaller in scale, their frequency and localized severity are increasing.

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6. Pollution

Historical Example:

- Limited relevance in prehistoric times; volcanic emissions may have caused atmospheric pollution in ancient extinctions (e.g., sulfur aerosols).

Contemporary Example:

- **Plastic pollution**, **pesticides (e.g., DDT)**, **heavy metals**, and **eutrophication** from agricultural runoff.
    - Cause direct harm to species and disrupt food chains (e.g., decline of pollinators and aquatic life).

Comparison:

- Pollution is a modern phenomenon, directly linked to industrialization and consumer culture, with no equivalent in past extinction events.