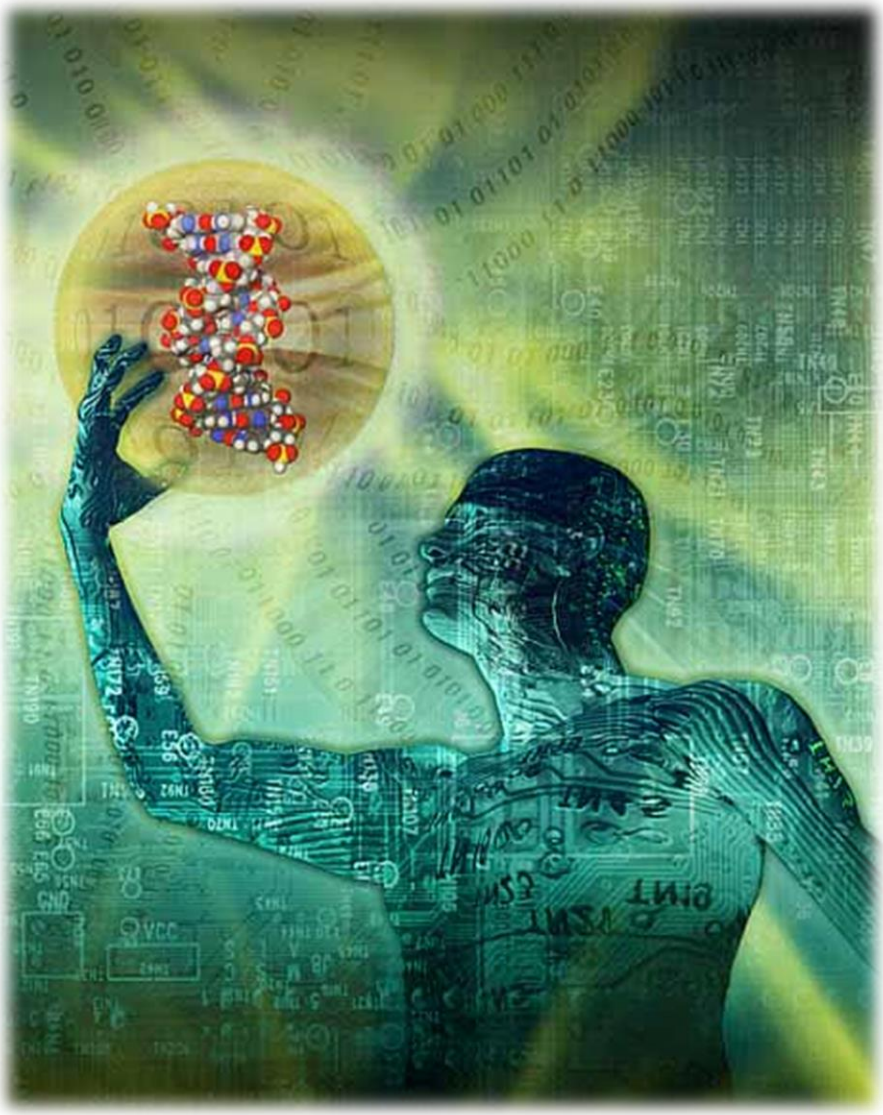
Topic 13
Genetics and Inheritance



Genetics and Inheritance

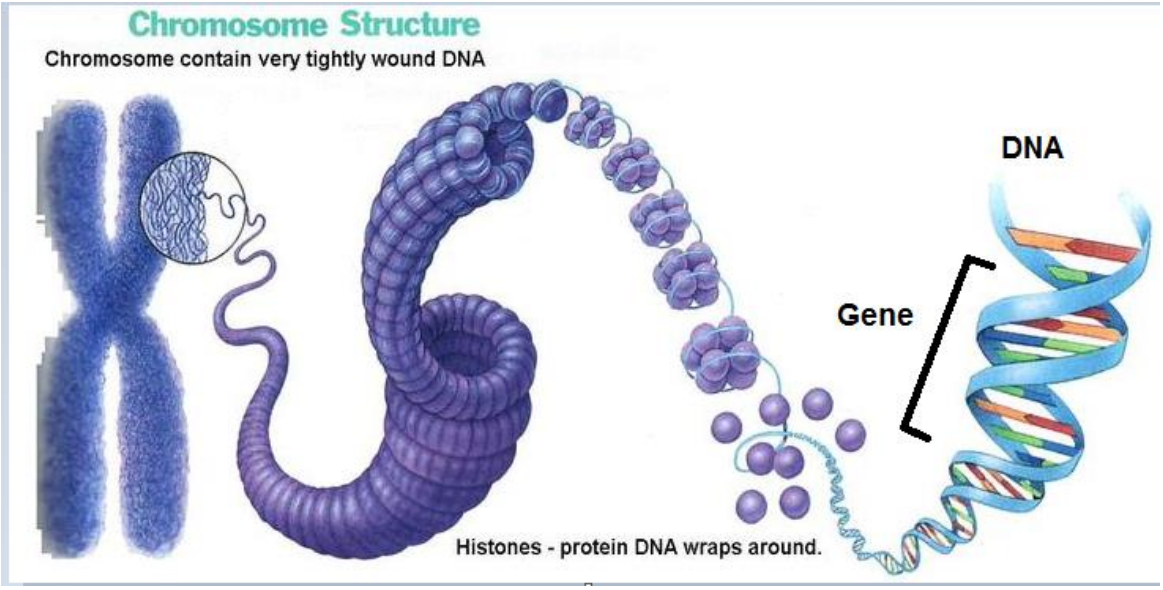
Inheritance is defined as the transmission of genetic information from generation to generation.

Genetics: is the science which deals with traits and characteristics that are transferred from the parents to their offspring

What does the nucleus of a cell contain?

Our Somatic cells contain 46 chromosomes (23 Pairs)

But what is a chromosome , DNA and Gene?



A Chromosome is a very long thread-like structures of DNA wrapped around proteins called **histones** , carrying genetic information in the form of genes Chromosomes are found in the nucleus of the cell carrying the genetic material of the cell.

DNA (deoxyribonucleic acid) a molecule that instructs the cell about which kinds of proteins should it makes.

A gene is part of DNA, part of a chromosome that codes for a specific protein. A gene may be copied and passed on to the next generation.

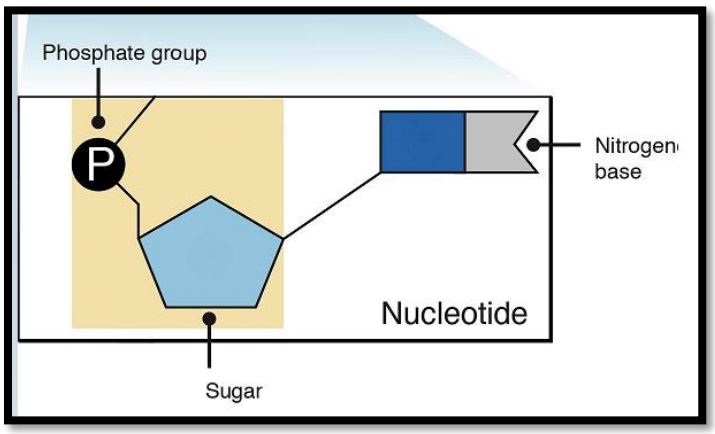
- It was found that the characteristics that a cell or an organism have (eye colour, skin colour, hair colour and texture, etc…) depend on the proteins that the cell

manufactures. **Each protein contributes towards a particular body feature.** Sometimes the feature is visible, such as eye colour or skin pigmentation. Sometimes the feature **is not visible** such as the type of haemoglobin in red blood cells .

DNA

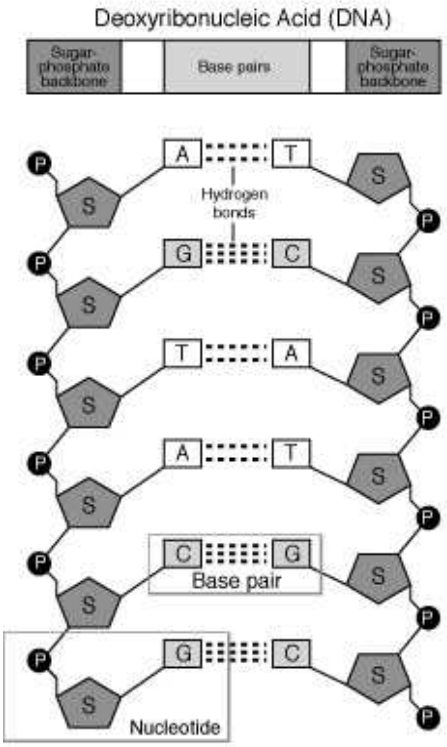
Chromosomes are made up of long coiled thread of DNA
What is Deoxyribonucleic acid (DNA) ?

Each DNA molecule consists of two long strands twisted together as a **double helix**. Each DNA strand is made up of many units called **Nucleotides making it a polynucleotide.**



☒ **Each Nucleotide is made up of :**

1. Sugar
2. Phosphate Group
3. Nitrogen base

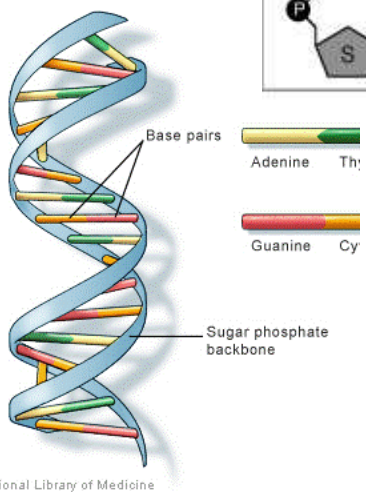


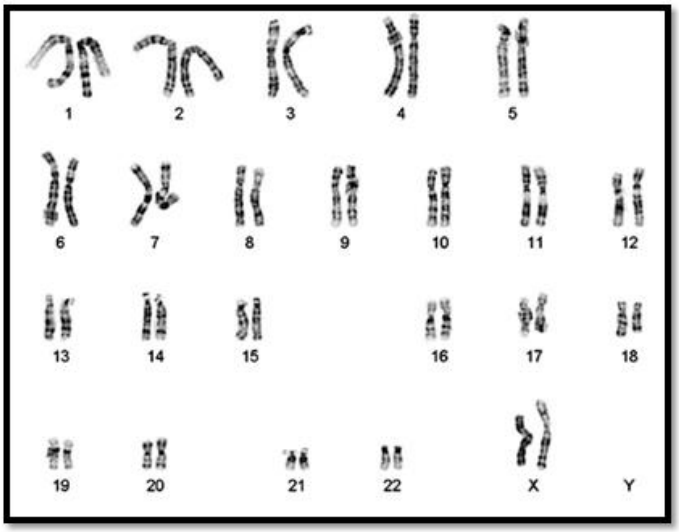
☒ **There are five different nitrogen bases :**

1. Adenine → A
2. Guanine → G
3. Thymine → T
4. Cytosine →C

- **<u>A always pairs with T</u>**
- **<u>C always pairs with G</u>** → they are called complementary bases, they always pair with each other and this is called the **base pairing rule**

☒ The Two strands are held together in the double helix by **hydrogen bonds** between the nitrogen bases





- Body cells are called **somatic cells**. All human somatic cells contain 46 chromosomes → these represent **the full set of chromosomes**. Each species of living organisms has a **different number of chromosomes** e.g. a mouse somatic cells have 40 chromosomes (full set of chromosomes)

Chromosomes are arranged in pairs, as you can see from the figure. In each pair, **one chromosome comes from the mother and the other comes from the father.** The chromosomes in a pair are called **Homologous Chromosomes:** which are two identical chromosomes that have the same shape and size, these carry **the same genes.**

A haploid nucleus is defined as a nucleus containing a single set of unpaired chromosomes, e.g. in gametes

A diploid nucleus is defined as a nucleus containing two sets of chromosomes, e.g. in body cells

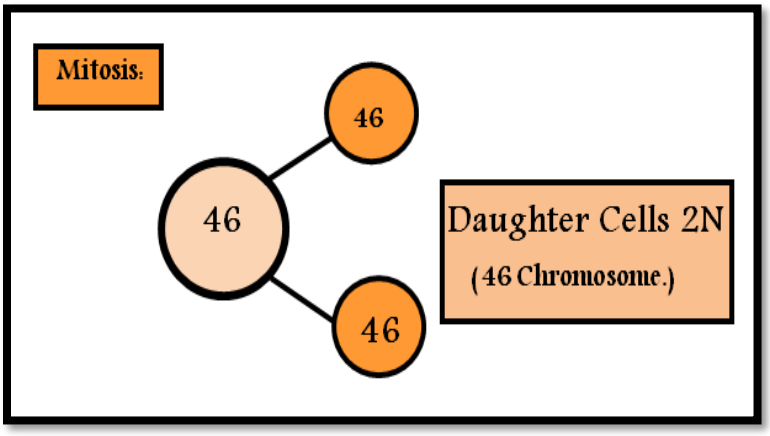
Haploid cell	Diploid cell
Human 23 Chromosomes	**Human 46 Chromosomes**
Mouse 20 Chromosomes	**Mouse 40 Chromosomes**

In a diploid cell, there is a pair of each type of chromosome and in a human diploid cell there are 23 pairs

- **Mitosis :** is defined as a nuclear division giving rise to genetically identical cells

<u>The Importance of Mitosis :</u>

1. For Growth (increasing the number of cells) Cell division in our body is a kind of asexual reproduction.
2. Repairing damaged cells
3. A method of asexual reproduction and the **production of clones** where cells divide like yeast and amoeba.



Notes:

- The produced cells are

genetically identical to the mother cell e.g. Muscle cells, Bones

- Cells divide because they reach a certain size in which they cannot grow anymore.

Stages of mitosis:

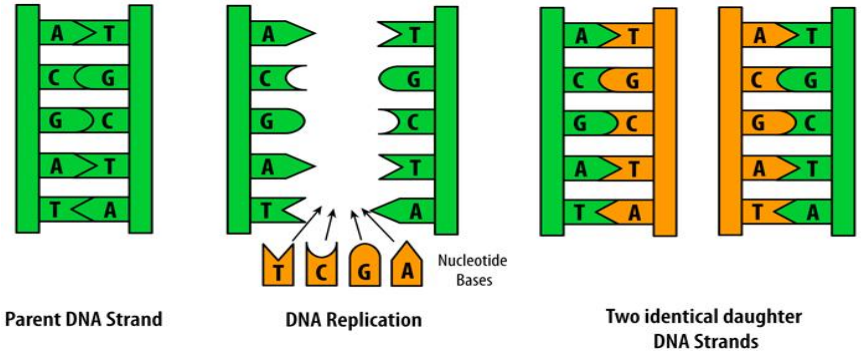
- Only four of the 46 chromosomes will be shown for simplicity

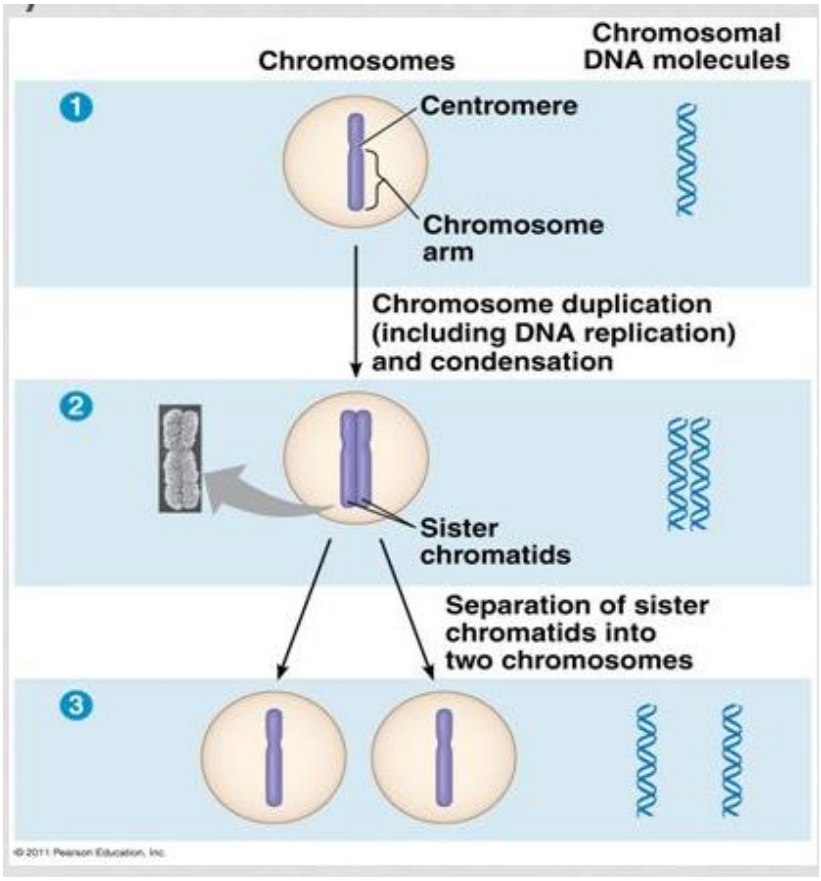
1) Before mitosis starts, **replication of the DNA** takes place so chromosomes are doubled.

- **Replication of DNA :**
  a. In DNA replication ,the double helix must first unwind and the hydrogen bonds holding the two strands must be broken and this is done by the action of certain enzymes
  b. When the hydrogen bonds between the two strands are broken , the two strands become separated and each strand is called a **Template strand**
  c. Each Template strand copies itself into another **complementary strand.**

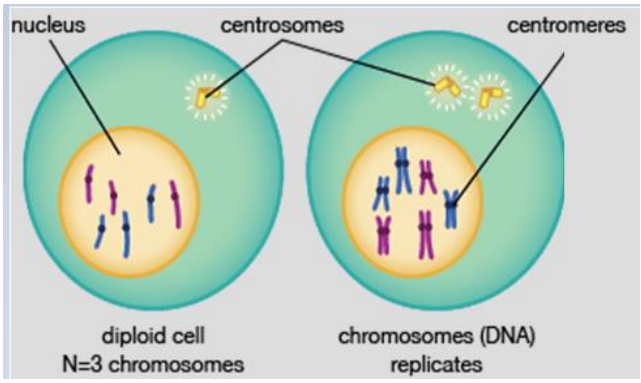
Adenine pairs with thymine and cytosine pairs with guanine .

  d. **DNA polymerase** assembles the nucleotides into two new strands according **to base pairing rule**





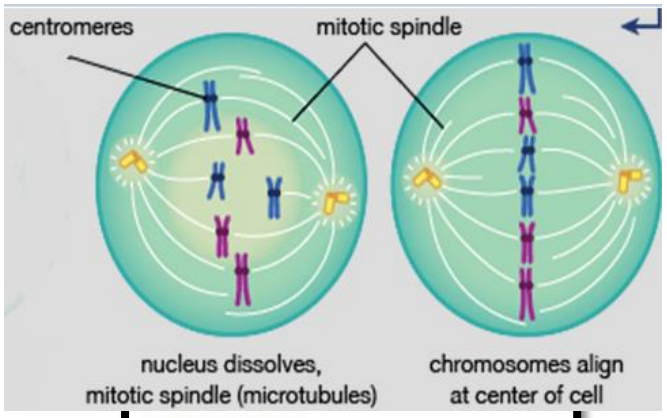
e. Hydrogen bonds then form between nitrogen bases of the two strands. The strands then wind up into a double helix.



- Chromosomes become **shorter and thicker (they become visible),** so

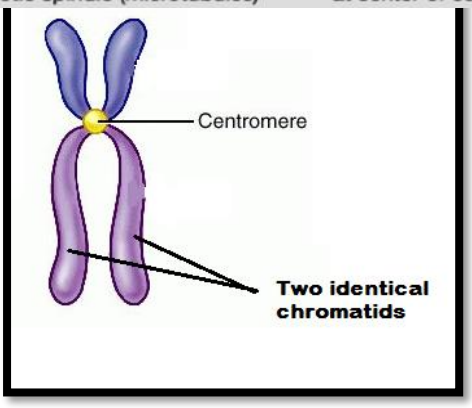
2) Each chromosome appears clearly to be made of **two chromatids** held together by a **centromere.**

- **Note : the chromosomes are only visible when chromosomes are dividing**



3) The nuclear membrane breaks down, the **centeriole** divides into two parts and each part goes to the opposite ends (Poles) of the cell and starts to produce **spindle Fibres**

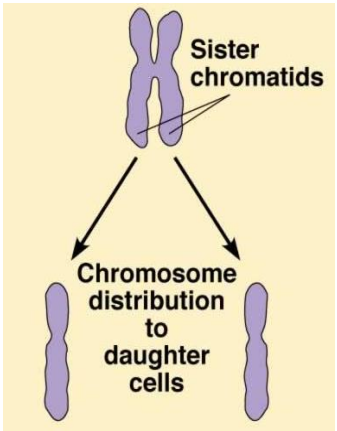
4) Chromosomes move to the equator of the cell and each chromosomes becomes attached by **two** spindle fibres from the centromere (From the two sides)



5) Spindle fibres **shorten** and pull the chromatids of each chromosome to the opposite poles of the cell. The centromere is divided and chromosome is divided into two chromatids .

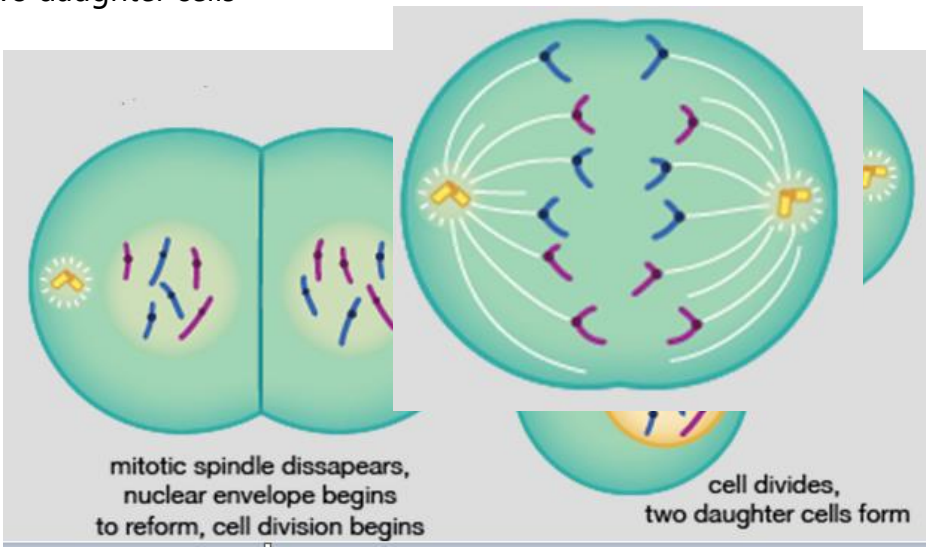
- **Note:** each chromatid is now considered as a daughter chromosome.

6) The cytoplasm starts to divide (pinching in the middle) and the **nuclear membrane reforms**. Each chromatid becomes a chromosome, chromosomes become thinner and longer.



7) They cytoplasm continues to divide till the two daughter cells are produced; the two cells are identical to the mother cell with the same number of chromosomes.

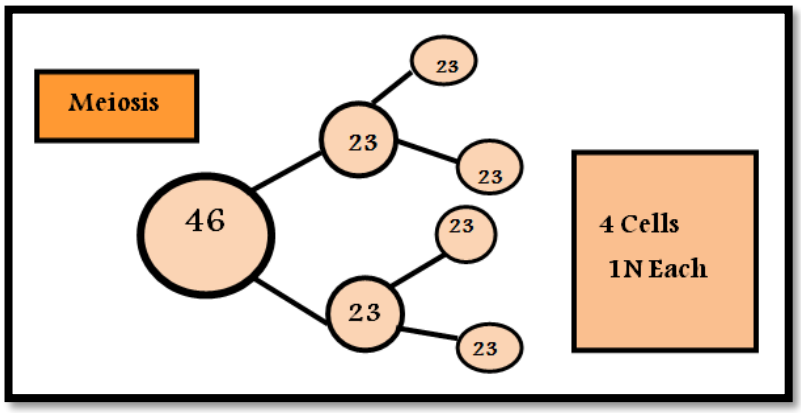
- **Note:** In mitosis, the two daughter cells produced have the **same genetic makeup**. This is because the DNA and the chromosomes were duplicated or replicated before the cell starts to divide.



Benefits of DNA replication :

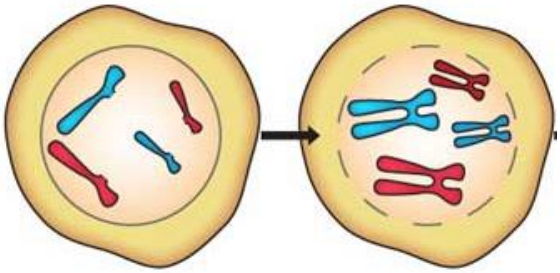
1. Ensuring that the genetic material is inherited from one living organism to another and from one cell to another
2. Ensuring that the new cells in formed by mitosis would have the same number of chromosomes as the mother cell

Meiosis : reduction division in which the chromosome number is halved from diploid to haploid resulting in genetically different cells



The Importance of meiosis:

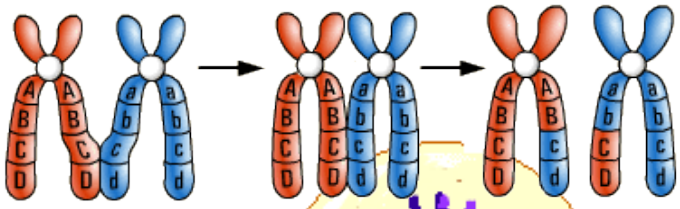
1. Production of sex cells
2. Halves the number of chromosomes , so that at fertilization the the diploid number of chromosomes is restored in zygote → the number of chromosomes remains constant in the next generation
3. Source of DNA Variation (through independent assortment and crossing over)

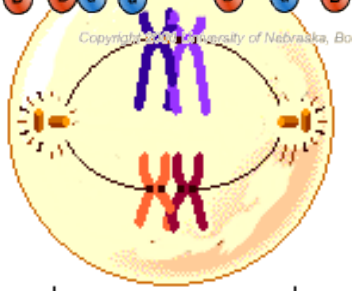


Stages of Meiosis:

1. Before Meiosis starts replication of the DNA takes place.

2. Chromosomes become shorter and thicker and pair together as **Homologous pairs.**

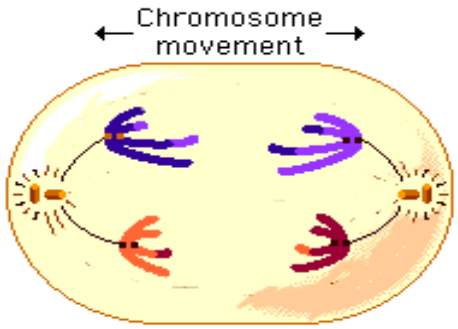




Note: when homologous chromosomes pair up, exchange of genetic material between the two chromosomes takes place → this is called **crossing over** and it is considered as a source of variation.

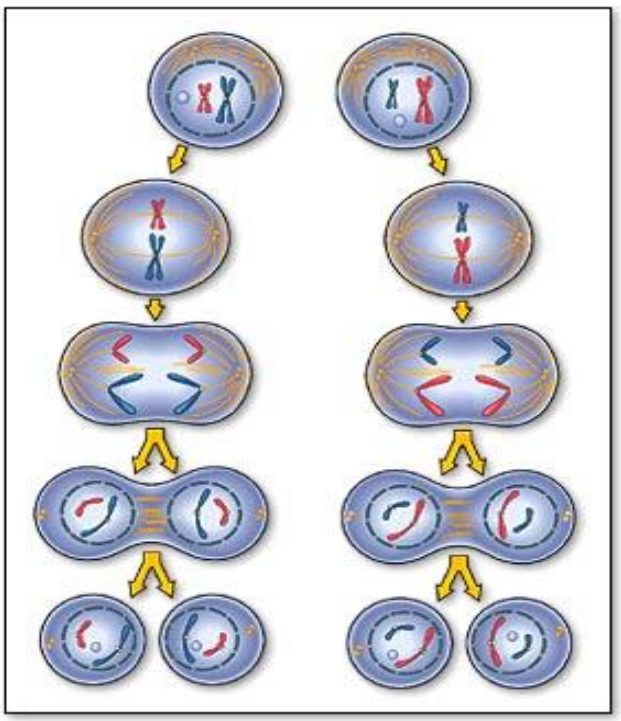
- **The nuclear membrane breaks down**
  Homologous chromosomes are arranged at the equator of the cell, and they are attached to the spindle fibres from the centromeres (Each Chromosome is attached by one spindle fibre)

3. Spindle Fibres shorten, pulling the chromosomes to the opposite poles of the cell.
4. **The nuclear membrane reforms**, the cytoplasm divides, and pinching happens starting to form the two cells.



5. **Cytoplasm continues to divide** till the two daughter cells are produced, each containing half the number of chromosomes.

1) Second Cell Division (Like Mitosis).



➔ Each cell enters a second cell division that resembles Mitosis producing four cells with **half the number of chromosomes of the original Mother cell.**

➔ The gametes produced will be genetically different from each other.

The four gametes produced will become sperms in males, whereas in females, one of the 4 gametes produced will become the ovum.

- **The Differences between Meiosis and Mitosis**

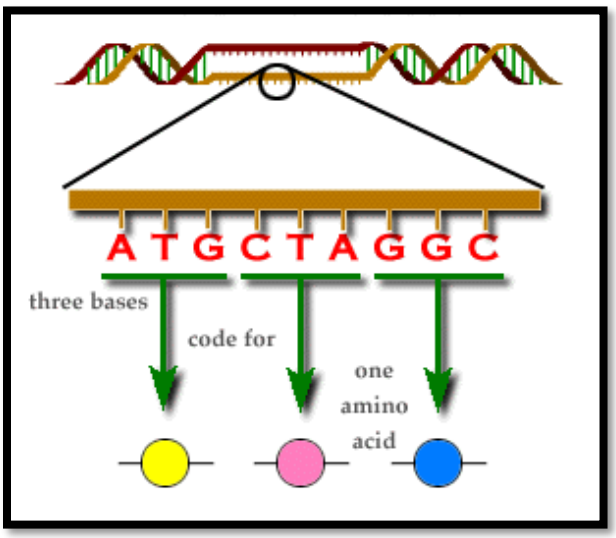
Meiosis	Mitosis
One cells gives four cells	One cell gives two Cells
Chromosomes are arranged in pairs	Chromosomes are arranged in a line
Each chromosome is attached to one spindle fibre	Each chromosome is attached by two spindle fibres
The two daughter cells contain half the number of original chromosomes {1N}	The two daughter cells contain the same number of chromosomes {2N }
Consists of Two stages	Consist of one stage only
During the shortening of spindle fibres the attached chromosomes don't split	During the shortening of spindle fibres , the attached chromosomes are divided into two chromatids and centromeres.
Happens in cells that form gametes In testes and ovaries	Happens in body cells for growth and repairing damaged cells.
Exchange of genetic material takes place	No exchange of genetic material takes place
The four daughter cells are not genetically identical to the mother cell	The two daughter cells are genetically identical to the mother cell.

The DNA code

- A GENE → Protein → Characteristic

Controls the production of **Which is responsible for**

Proteins are formed from chains of amino acids , the order of these amino acids in the chain determines which type of protein is produced.
A gene produces a certain protein because the sequence of the bases on the DNA strand controls the order of the amino acids that are joined together to form the protein.



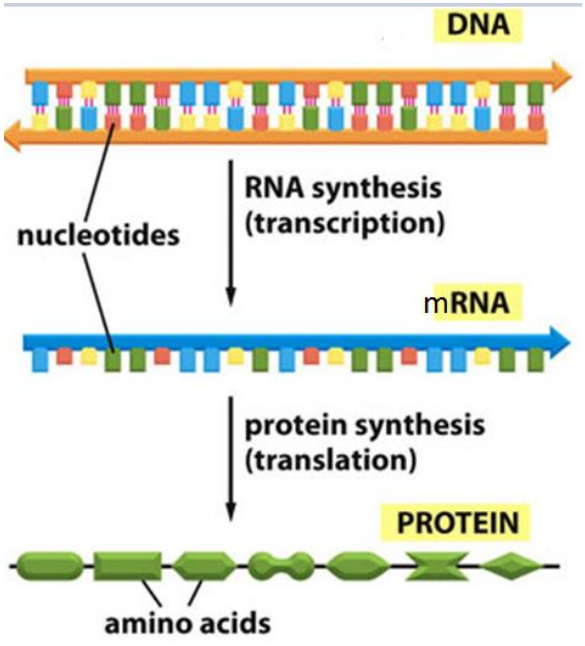
The sequence of bases in the DNA strand determines which type of amino acid is used and the sequence of amino acids.
Every three bases stand for one amino acid **(triplet code)** e.g. GGU→ codes for amino acid 1, ACT → Amino acid 2
The sequence of bases that code for all the amino acids in a protein is a gene

Protein synthesis takes place on the ribosomes (which are found in the cytoplasm), but the code lies in the DNA which always remains in the nucleus

So how does protein synthesis take place?

Steps of protein synthesis:

1) The two strands of the DNA separate and one of these strands is copied into strand of messenger RNA (mRNA) **this strand is called the template strand .** The sequence of bases on this DNA strand will determine the sequence of bases on the mRNA strand
2) When the forming of the mRNA is completed. It is released from the DNA and it escapes out of the nucleus through the nuclear pores into the cytoplasm
3) mRNA strand binds to a ribosome , the sequence of bases in the mRNA is used to build up a sequence of



amino acids into a protein in the ribosome

Because the base sequence on the mRNA is determined by the base sequence on the DNA → it's the DNA code that controls which proteins are formed in the cell.

All the cells of your body contain the same DNA . But not every body cell produces the same proteins. Specialized cells have different functions depending on which proteins they can produce.
e.g. muscle cells make special proteins for cell contraction , glands in the pancreas produce the protein hormone insulin.

Muscle cells do contain the gene for producing insulin and pancreatic gland cells also contain the genes for producing contractile proteins , however each cell only produces the particular proteins that it needs for its function.

- The DNA controls the cell function by controlling the production of proteins (some of which are enzymes) antibodies , receptors for neurotransmitters
- **Go back to pages 7-10 (Human Biology Book and study " The stages of protein synthesis "**

Mitosis in Stem cells :

What are stem cells ? They are **unspecialised** cells that divide by mitosis to produce daughter cells that can become specialised for specific functions

- Once the cells have differentiated (became specialized) , they lose the ability to divide and produce different kinds of cells .

There are Two types of stem cells :
Adult stem cells → undifferentiated cells that are found among differentiated cells in tissues and organs e.g. bone marrow . These cells will still have the ability to divide and produce some types of differentiated cells, usually the same as those in the tissue where they are found. The roles of these cells are to **replace damaged cells and produce new cells for growth** e.g. when muscles increase in size after puberty in boys and through exercise.

Embryonic stem cells: are stem cells in the early stages of embryo development, these cells can divide and produce daughter cells that can become any kind of body cell.

➔ These can be obtained from fertility clinics where parents choose to use their unused embryos (formed during IVF) for research.

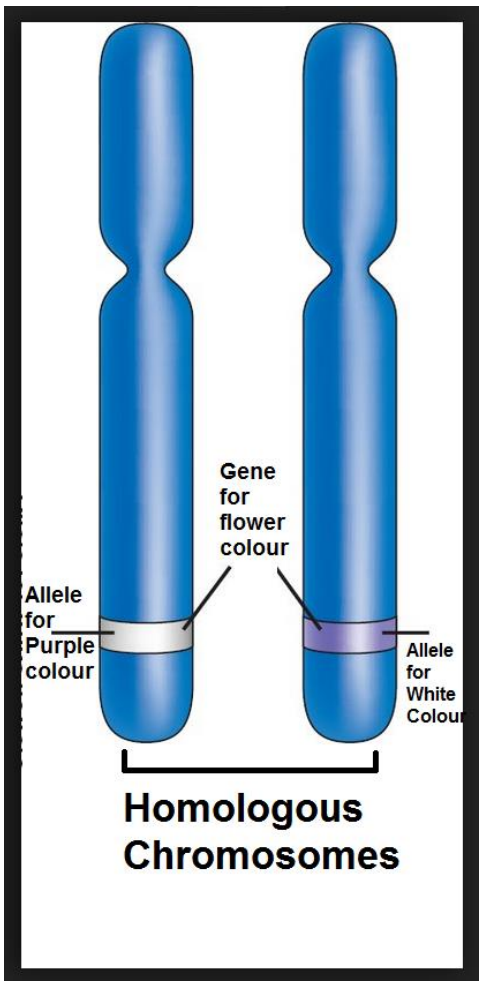
The use of stem cells to treat or prevent a disease, or to repair damaged tissues is called **stem cell therapy** ,

- Stem cells can be used in the treatment of certain disorders such as diabetes type 1 , bone marrow transplant for treating leukemia (a type of blood cancer) or after heart disease .
- Embryonic stem cells can be used more easily than adult stem cells to produce any kind of cells for these treatments.
- **Many people have moral or ethical objections** to using cells from embryos for medical purposes, even though they might one day be used to cure many diseases

Monohybrid inheritance

Each chromosome can carry alternative forms of the same gene, **these alternative forms are called alleles**, for example the gene for flower colour has two alleles that code for purple or white colour .

Alleles: two or more versions of genes that control a particular trait, these alleles occupy corresponding positions on the homologous chromosomes.
If the two alleles for a particular gene on the homologous chromosomes are the same then the organism is said to be **homozygous for that trait.**
If the two alleles for a particular gene are different then it is **heterozygous for that trait.**



<u>If a flower has an allele for purple colour and an allele for white colour what determines which colour this flower is going to have ?</u>
Alleles can be dominant or recessive :
If a dominant allele is present , **it is always expressed** even in the presence of a recessive allele, Which means whether the organism is **homozygous or heterozygous.**

The dominant allele switches off the recessive allele

A recessive allele is expressed only **if there is no dominant allele present (both are recessive),** this gene is only expressed in the homozygous recessive state.

We use letter to represent the alleles,
The **dominant allele** is represented by a **capital letter** and the **recessive allele** is represented by a **small letter** , for example, let's say that purple colour allele is dominant over white colour allele , then the allele for purple colour is represented by a **capital P** ,the allele for white colour is represented by **a small p**

- **Genotype :** is the genetic makeup of an organism in terms of the alleles present (combination of alleles for a particular trait) e.g. Skin colour
  B-Black b-White
  We might have the following characteristics of genotype:
  BB → the skin will be black because the dominant allele will be expressed
  Bb→ the skin will be black because the dominant allele will be expressed
  bb→ the skin will be white because the recessive will be expressed because the dominant allele is not present

Phenotype: the observable features of an organism which result from the interaction of genotype and the environment.

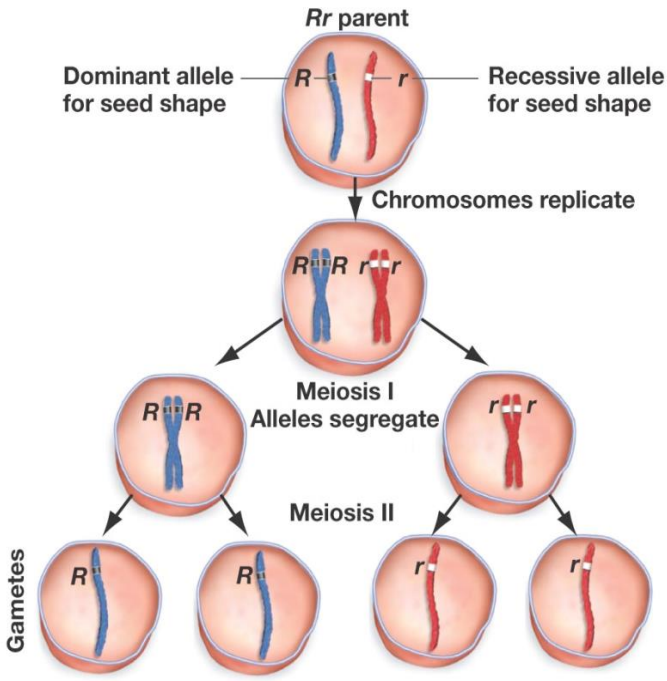
e.g. BB ⇒ Black Colour
bb ⇒ White Colour

Keep in mind that Dominant Gene (allele) → is the allele that shows itself in the phenotype even if there is a recessive allele,

E.g. Bb ⇒ Both genotypes show black trait because
BB B-black is dominant.

Note: the words homozygous and heterozygous can describe both the trait and (Or) the organism itself

❖ **<u>Gamete Formation :</u>**
Traits are passed from parents to the offspring by gametes; gametes only have 23 chromosomes, so they contain one chromosome from each homologous pair, that's why a gamete is



called a **haploid cell** , whereas any other cell in the body is called a **diploid cell** or a somatic cell → contains the full set of homologous chromosomes.

SOURCES OF VARIATION in SEXUAL REPRODUCTION :

- **Meiosis :**

In meiosis variation can occur due to

- **Crossing over** : chromosomes exchange genes
- the homologous pairs may line up in different ways producing different combination of genes in the gametes → **Independent assortment**

- **Random fusion of gametes** : any male gamete can combine with any female gamete

- **Mutation**

Let's consider this example

B- Black skin

b- White skin

Imagine a male mouse with the genotype Bb. When sperms are made inside the testes, each sperm will get only one of the two alleles. Each sperm either have the B-allele or the b-allele. There will be almost equal number of each kind of sperm.

- The same applies to a female mouse with the genotype bb. When the eggs are made in the ovaries, each egg with only get one of the b alleles.
- **So what do you think will happen when the two mice mate?**
  There are sperms with a B-allele and sperms with a b allele, so here if a sperm with a B-allele fertilizes the egg the genotype of the zygote will be Bb (black skin) but if a sperm with the b-allele fertilizes the egg, the genotype of the zygote will be bb (white skin)
- **Example :**
- **Assuming that black skin is dominant over white skin in mice**
  If a Homozygous black male has mated with a white female what would be the likely genotypes and phenotypes of the offspring
  To answer such a question, first start by
  - writing down the phenotypes and genotypes of the parents
  - then write down the different types of gametes they can make

	B- Black dominant	**b-White recessive**
Parents phenotype :	Black	White

Parents genotype BB bb
Gamete type B b

The next step is to write down what might happen when fertilization takes place

Offspring genotype **Bb , Bb , Bb, Bb**
Offspring phenotype : **100% Black skin**

- **Notice** that all the genotypes of the first generation is **Bb** ,this means that 100% of the expected individuals have the phenotype (black skin).
- two identical homozygous individuals that breed together will be **pure-breeding**
- Pure bred plants constantly have offspring with the same trait as the parents

Example 2
Suppose that a crossing was made between a tall plant and another plant , both are heterozygous.

Parent Phenotype Tall X Tall
Genotype of parents : **Tt** **X** **Tt**
Gamete formation : **T, t** **T,t**

offspring genotype : **TT, Tt, Tt,tt**

Note: This is called a **genetic diagram** for the first generation. The ratio of the phenotype is 75 % Tall, 25 % Short. Ratio (3:1)

Ratio 1:1 results in offspring when crossing or mating takes place between two mates one is **homozygous recessive** and the other is **heterozygous dominant**, the probability of the expected phenotypes would be:
50% Dominant Trait
50% Recessive Trait
This means that the resulting ratio would be **1:1**

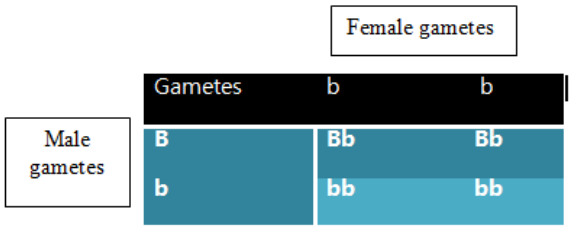
Note : heterozygous individual will not be pure-breeding

Example 3 .

Assuming that the black colour in rabbits is **dominant** while the white colour is **recessive.**
B- Black b-white

- **A heterozygous black male** was crossed with a white female. Construct a punnet square to show of the probabilities of the first generation :

Parents phenotype **Black male** **White female**
Parents genotype **Bb** **bb**
Gametes **B and b** **b and b**



Genotypes of first generation (f1) **1Bb: 1 bb**

Phenotypes of F1 : 1 Black : 1 White

- **Note:** This diagram is called **Punnet Square**
- **Result:** 50% Black, 50% white

Notice that the ratio is 1:1 the ratio only tells you the probability of the first generation,

There is one point that you should keep in mind , having a 50% black 50% white percentage **doesn't mean** that if the animal gave birth to two offspring one of them would be black and the other will be white ! It just says that the chances of getting a black or a white offspring are equal .

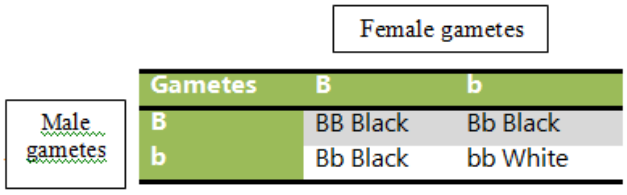
If both parents are heterozygous a 3:1 ratio results in offspring

Parent phenotype **Black** **X** **Black**

Parent genotype **Bb** **Bb**

Gametes **B or b** **B or b**

Offspring



Genotype : BB , Bb , Bb , bb
The ratio is 3:1
Offspring phenotype 75% Black , 25% white

The Ratio 3:1 results when a crossing takes place between two mates that are **dominant heterozygous**

- ## The inheritance of eye colour in humans:

The inheritance of eye colour is determined by two alleles

The dominant allele is for coloured eyes (Brown , black , green , etc)

The recessive allele is for uncoloured eyes (Blue only)

Example: If Mating took place between heterozygous brown eyed women and blue eyed man what would be the probable genotypes of their children?

These couple actually got four children with the following genotypes:
Parents genotype Male ee Female Ee
Actual children genotype Ee, Ee ,Ee, ee

- We notice from this example that the genotype of the actual children are from the probability of the genotype of male and female 1:1

1. How many phenotypes are formed in this family?

2. If the family gets another child with brown eyes what would be his genotype?

3. What is the percentage of getting a blue eyed child?

4. How many members of the family including parents are heterozygous?

Test cross\ back cross :

Remember that if the phenotype is the dominant characteristic there would be two probabilities for the genotype (homozygous or heterozygous). To find out

whether the dominant trait is homozygous or heterozygous we carry out a crossing with an organism that is known to have **a homozygous recessive genotype for the same gene.**

--> This is called a test cross

- If the ratio of the offspring produced is 1:1 this means that the unknown is **heterozygous dominant** or if there are two traits in the produced individual.
- If the offspring produced is 100% dominant, the unknown would be **homozygous dominant.**

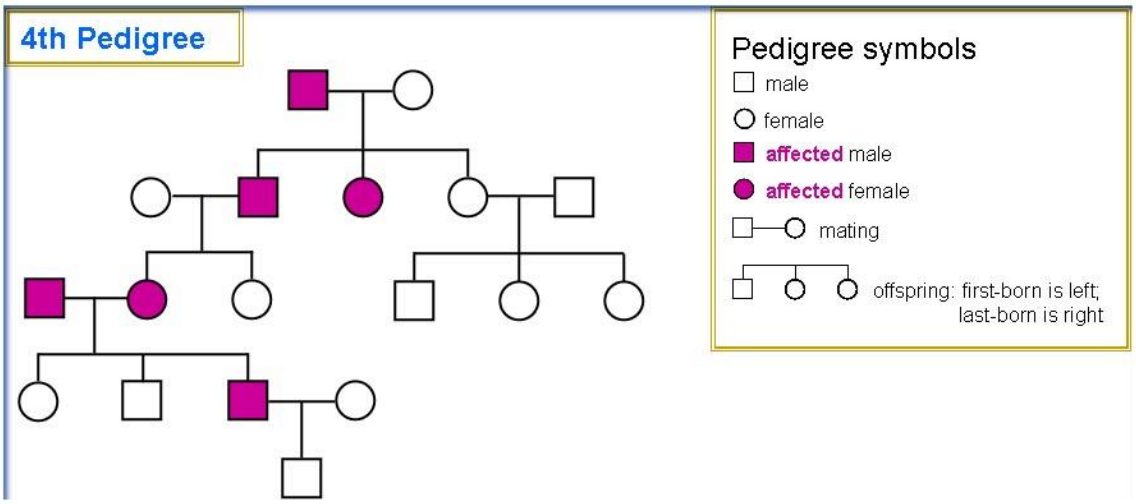
Example 5
B-Black b-White
Black dog  **BB or Bb (options)**

- **Cross breeding is carried out using homozygous recessive**
  F1 → BB X bb → Result: Bb 100% black none are white

This can also happen by chance: maybe none of the gametes carrying the recessive allele were successful in fertilization
That's why large numbers of offspring should be present

F1→ Bb X bb → Result : Bb, bb if any is white then it is heterozygous

Family pedigrees:



A Pedigree is the presentation of family information in the form of an easily readable chart to be able to trace the inheritance of a certain trait or disease in a certain family by drawing the family tree.

Co-Dominance:

All the previous examples were an example of **complete dominancy;** where an allele is dominant over a recessive allele and this dominant allele completely masks the effect of the recessive allele. **The recessive allele appears only if it was present with another recessive one.**
It is not true that always one allele would be dominant over the other, sometimes in some traits, if the two heterozygous alleles meet together no allele will be dominant over the other so they share their effects and give a new intermediate trait and this kind of inheritance is called **co-dominance**.

- **Example**: in some flowers there are two alleles for the flower's color.

  C^R → Red colour **C^W → White colour**
- No allele is dominant over the other in this case.
  → How?
  Suppose that a cross breeding happened between Red and White flowers.

  Genotype → Male C^RC^R Female C^WC^W
  Gametes → C^R, C^R X C^W, C^W
  F1 Genotype → C^RC^W, C^RC^W, C^RC^W, C^RC^W

- The colour produced would be pink which means that neither the white colour nor the red is dominant.

- Note: in this kind of genetics we don't use capital and small letters; instead we use the letter C for co-dominance with two different capital letters.

  Example 6:
  Suppose that a cross breeding took place between two pink flowers what would the phenotypes and genotypes produced be?

	Male	**Female**	
Genotypes of parents	C^RC^w	C^RC^w	25% Pink
Gametes :	C^R **,** C^W	C^R, C^W	25% Pink

  Genotype of Offspring. : C^RC^R, C^RC^W, C^RC^W, C^WC^W

- The ratio is 1:2:1
- Note that there are three phenotypes present → Indicates co- dominance

Another example of co-dominance is **Blood group inheritance**

- **<u>Inheritance of blood groups :</u>**
  Some genes have more than two alleles (multiple alleles) , for example the gene controlling the human blood groups has three alleles given the symbols I^A , I^B , I^O

  In humans, there are four different blood types: **A, B, AB, O**. The blood type is determined **by the presence or absence of antigens on the surface of the red blood cells.**

Genotype	Antigen produced	Blood group
I^AI^A , I^AI^O	A	A
I^BI^B , I^BI^O	B	B
I^AI^B	A and B	AB (codominance)
I^OI^O	Neither	O

  I^A → Determines the production of **A antigen** on the surface of red blood cells
  I^B -→ Determines the production **of B antigen** on the surface of red blood cells
  I^o → Determines that neither **A nor B is produced**

The blood type is determined by the three alleles I^A, I^B, I^O alleles I^A, I^B come together and both show themselves so they are **co-dominance**.

We need any 2 alleles and the presence of 3 of these gives us more variation and combinations between them.

Question:

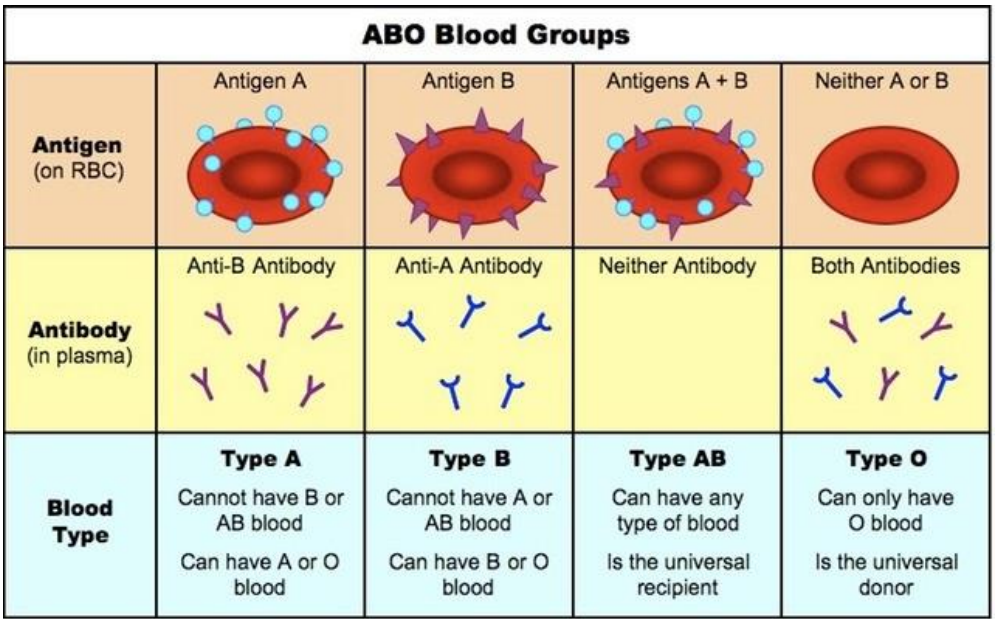
A man with blood group A married a female with blood group B, they had a child with blood type O. Draw a genetic diagram to show the possible genotype and phenotype of the children.

	Father	Mother
Genotype	I^AI^O	I^BI^O
Gamete Genotype	I^A, I^O	I^B, I^O

F1 Genotype : I^AI^B, I^AI^O, I^BI^O, I^OI^O

- **25% AB , 25% A , 25% B ,25% O**

Blood Transfusions:

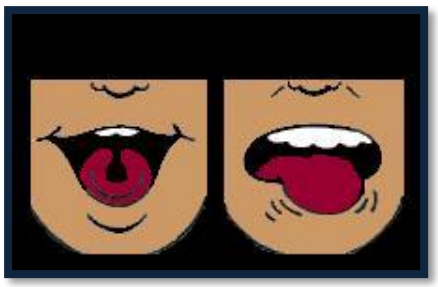


• Tongue rolling

Is a trait controlled by two alleles, **R-allele** for rolling and **r-allele** for not being able to roll.

Question:

A male got married to a female who couldn't roll her tongue. What would be the male's genotype if they gave birth to a girl that was able to roll and to a boy that couldn't.



Genotype Of parents:

Male Rr , ~~RR~~ , ~~rr~~ **female** rr

Because one child contain r

Because one child contain R

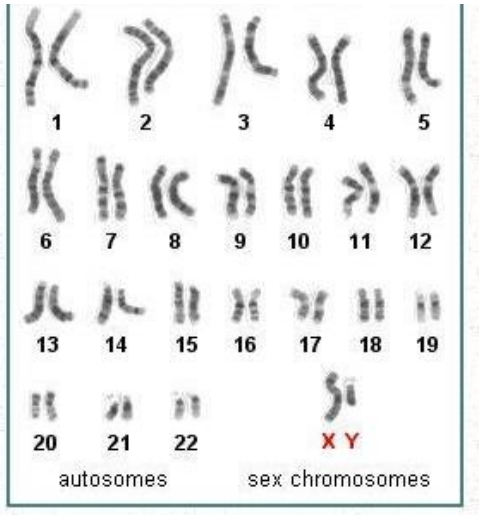
Gametes formed : R , r r,r

After Fertilization: Rr, Rr, rr, rr→ Girl

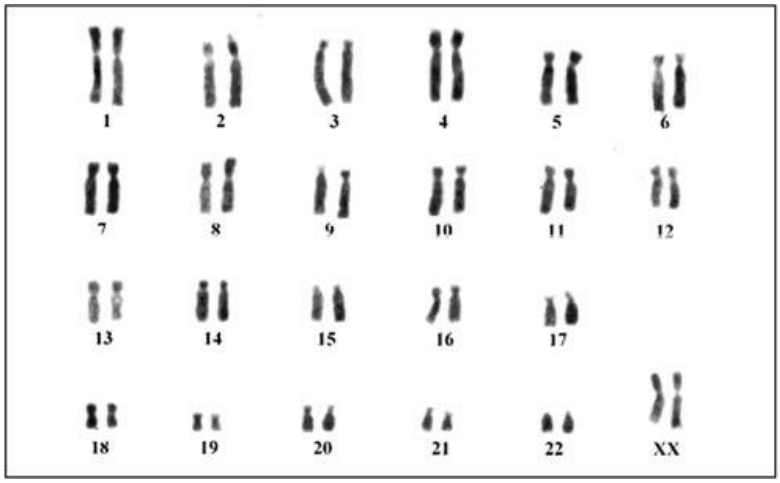
Sex Determination in humans

In somatic cells in humans , there are 23 pairs of chromosomes .22 pairs are called **somatic chromosomes (autosomes)** whereas the 23rd pair are called **sex chromosomes** .This sex pair is **different** in males than in females.

- In male the pair is different XY

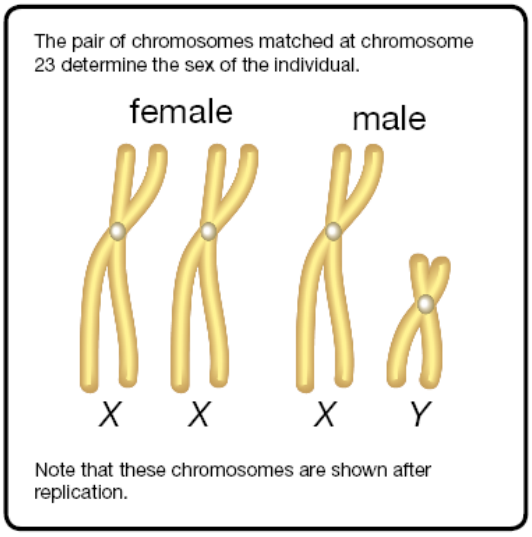


- In females both chromosomes are alike : XX



The Difference between male and female sex chromosomes :

Chromosome X	Chromosome Y
Longer	Shorter Contains fewer genes (because it is shorter)



- **How sex is determined in humans :**

Chromosomes in females that determine sex are **X,X** while in males they are **X,Y** . During gamete formation, the males will give two different gametes, one gamete might give X while the other might give Y. alternatively in females any ovum produced will **only contain X** . Hence the probability of getting a boy or a girl is:

50% Girl

50% Boy

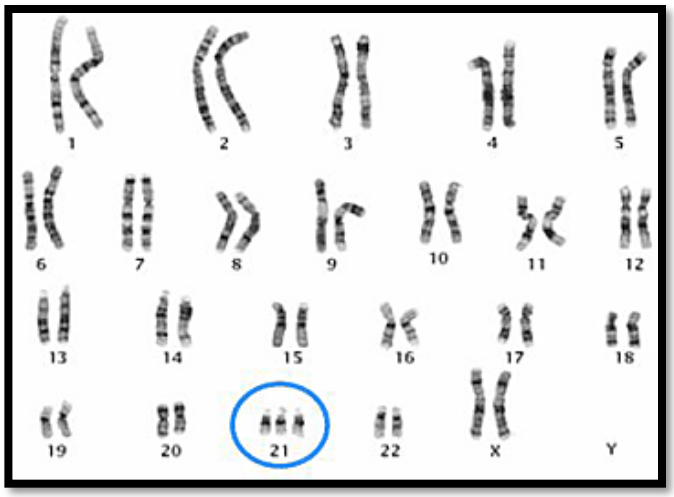
Parent phenotype	Male	Female
Parent genotype	XY	XX
Gametes	X or Y	X or X
Genotype of offspring	XX, XX, XY, XY	
Phenotype of offspring	**50% Girl 50% Boy**	

So each time a baby is conceived there is a 1:1 chance of getting a boy or girl

- The man's sperm determines the sex of his children because the woman can only produce gametes with an X chromosome .

Mutation:

An Unpredictable change that occurs in the DNA of the cell it can happen in **genes or in the whole chromosome.** This change leads to the appearance of different phenotypes.



• Chromosomal Mutation:

This type of mutation affects the whole chromosomes. For example when eggs are being made by meiosis in a woman's ovaries ,homolgous chromosomes number 21 in some cases , do not separate as a result both chromosomes end up in the ovum which will end up containing 24 chromosomes. If this ovum is fertilized, the zygote will contain 47 chromosomes (with three copies of chromosome 21 , one from the father and 2 from the mother) , A baby that contains 47 chromosomes has **Down's syndrome .**

A child with down's syndrome has different features from a normal one :

1. They have a shorter life expectancy, as heart and lung defects are common.
2. The baby is shorter with wide broad with some degree of mental disability.
3. Sexually immature

- **Note :** Older women are more likely to give birth to a child with down's syndrome (**trisomy of chromosome 21**) than younger ones.

Gene Mutation

It is defined as a change in the base sequence of DNA

- mutation is the way in which new alleles are formed
- Change in the sequence of bases in a gene → this leads to the gene coding for a wrong protein

Go back to pages 10-11 (Human Biology book) and study " gene Mutations- when DNA makes mistakes

Examples of Diseases caused Gene Mutation				
Cystic Fibrosis	Sickle Cell Anaemia	Albinism	Haemophilia (sex Linked)	Red-Green Colour Blindness (sex Linked)

Albinism:

It is caused by the inheritance of two recessive mutated alleles.

There is a gene that is responsible for the production of the brown pigment **"Melanin ".** Sometimes this gene mutates to produce an allele that does not code for the production of melanin at all. This gene is recessive so if two copies of this recessive gene were present no melanin will be produced and this leads to the production of very pale skin and hair with red eyes (albinism)

Cystic Fibrosis:

Results from recessive mutated alleles that result in the secretion of very thick mucus, the dominant allele controls the production of normal mucus.
To be infected → a person must inherit two recessive alleles (one from each parent) so each one of them must carry at least one recessive cystic fibrosis (heterozygous).

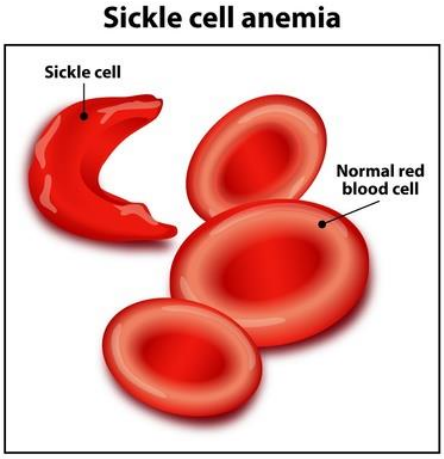
This has several effects on the body:

- This disease causes blockage in the pancreatic duct so that pancreatic enzymes cannot reach the small intestine, this affects the digestion of carbohydrates ,lipids and proteins which might lead to malnutrition .
- The thick mucus won't be easily moved out of the lungs by the action of cilia, so the mucus will collect in the lungs making gaseous exchange difficult and making infections more likely.
- Very thick cervical mucus which might affect fertility in females

- **Go back to page 223 (Human Bio Book) and study " using gene therapy to treat cystic fibrosis "**

Sickle cell anaemia :

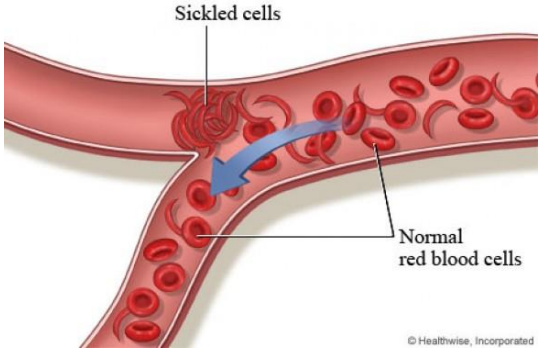
Some people have a mutation in the gene that codes for the production of haemoglobin . The normal allele HB^A codes for normal haemoglobin the mutant allele HB^S codes for an abnormal type of haemoglobin .



What are features sickle cell anaemia ?

- Fewer red blood cells (because they are destroyed in the liver at a higher rate than normal red blood cells because of their sickle shape)
- This abnormal haemoglobin can form fibers in the RBCs **at low oxygen concentrations** so the RBCs will develop a sickle shape.

- The Haemoglobin is less efficient at transporting oxygen → less respiration of cells → less energy is released
- The sickle –shaped red blood cells can get stuck in and cause capillary blockage → if the blood supply to the brain is affected it can cause a stroke.



Example :

HB ^{A}→ Dominant allele (normal haemoglobin)

HB ^{S} → recessive allele (abnormal Haemoglobin)

HB^{A} HB^{A} → Healthy

HB $^{A}HB^{S}$ → **Sickle cell trait :** mostly normal RBC's along with some sickle – shaped cells the person will be normal but with slight symptoms of anaemia (co-dominance)→ HB ^{S} HB^{S} → both are recessive, sickle cell anaemia with severe symptoms

A cross between heterozygous carriers of sickle cell anaemia

	Male	Female
Genotype of parents	$Hb^{A}Hb^{S}$	Hb^{A} Hb^{s}
Gametes	**HB^{A} and HB^{S}**	**HB^{A} and HB^{s}**

	Hb^{A}	Hb^{S}
Hb^{A}	**Hb^{A} Hb^{A}**	**Hb^{A} Hb^{S}**
Hb^{S}	**Hb^{A} Hb^{S}**	**$Hb^{S}Hb^{S}$**

Offspring genotype :

$Hb^{A}Hb^{S}$, $Hb^{A}Hb^{S}$, $Hb^{S}Hb^{S}$ $Hb^{A}Hb^{A}$,

Phenotypes of offspring 1 unaffected: 2 Sickle carrier : 1 affected

Sex Linked characteristics :

Sex chromosomes don't just determine sexual development (development of sex organs), they also carry genes for other characteristics. Some characteristics are carried on the sex chromosomes for that reason they are called **sex linked characteristics**. Since the Y chromosome in males is smaller than the X chromosomes, it will contain a fewer number of genes. This means that for some

genes, a male **will only have one allele** which is present on his X chromosome .

Sex-linked characteristic is defined *as* a characteristic in which the gene responsible is located on a sex chromosome and that this makes it more common in one sex than in the other.

An example of this is the gene that causes Haemophilia and red-green colour blindness:

What is haemophilia ?

When a healthy person's skin is cut, a clot forms, this prevent excess loss of blood and entry of bacteria. This process is controlled by many different plasma proteins some people have inherit a gene that affects the production of one of these plasma proteins, as a result, their blood fails to clot .

X^HX^H → Normal female

X^HY→ Normal male

X^HX^h→ Haemophilia carrier female (can pass to children)

X^hY → haemophiliac male

X^hX^h → haemophiliac female

Example :

A normal male married a normal female, but the female's father was affected with haemophilia . Construct a genetic diagram showing the children's phenotype and genotype.

- Answer

Red – Green colour Blindness :

- Red –green Colour blindness it is inherited in a similar way as haemophilia these diseases are more common is boys than girls.

This is not true blindness, but it is the inability to distinguish some colours from others, it occurs because some of the cone cells in the eye fail to produce the protein that responds to light. **The most common form is red-green colour blindness ,** but there are other types as well.

- The allele that causes red-green colour blindness is recessive .

as we can see , females need to **inherit two copies of the abnormal recessive allele to have the disease(one from each parent)** and if she has only one copy she won't have the disease because she has the a dominant allele on one of

her X chromosomes → but she will be considered a carrier (she can pass it on to her children)

- Males need to **inherit only one allele of the gene to have the disease.**

Go back to pages 215-217 (Human Biology Book) and study " • Pedigrees "

Causes of Mutation:

- Mutations that occur in body cells, or somatic cell often have no effects at all on the organism. Somatic mutations cannot be passed on to offspring by sexual reproduction. **However, mutations in cells in the ovaries or testes of an animal, or in the ovaries or anthers of a plant, may be inherited by offspring**. If a cell containing a mutation divides to form gametes, then the gametes may also contain the mutated gene. If such a gamete is one of the two which fuse to form a zygote, then the mutated gene will also be in the zygote. This single cell then divides repeatedly to form a new organism, in which all the cell will contain the mutated gene.

- It is important to realize that mutations take place randomly , however a number of factors can increase the rate at which mutations occur.
  These factors are called **mutagens** and they include :

- **Ionizing radiation** such as ultraviolet light (UV) , X-rays and gamma rays.
- **Many different chemicals** e.g. tar in tobacco smoke and high concentrations of preservatives.

Both of these can cause damage to the structure of DNA

Adaptive Features

An adaptive feature is defined as an inherited feature that helps an organism survive and reproduce in its habitat / environment , an adaptive feature increases organism's fitness;

. These features are inherited and passed from generation to generation in genes.

For example, fish have fins and stream-lined body that are adaptive features to allow them to move through water . Reptiles lay hard shelled eggs which is an adaptive feature for reproduction (so that the eggs won't dry out).

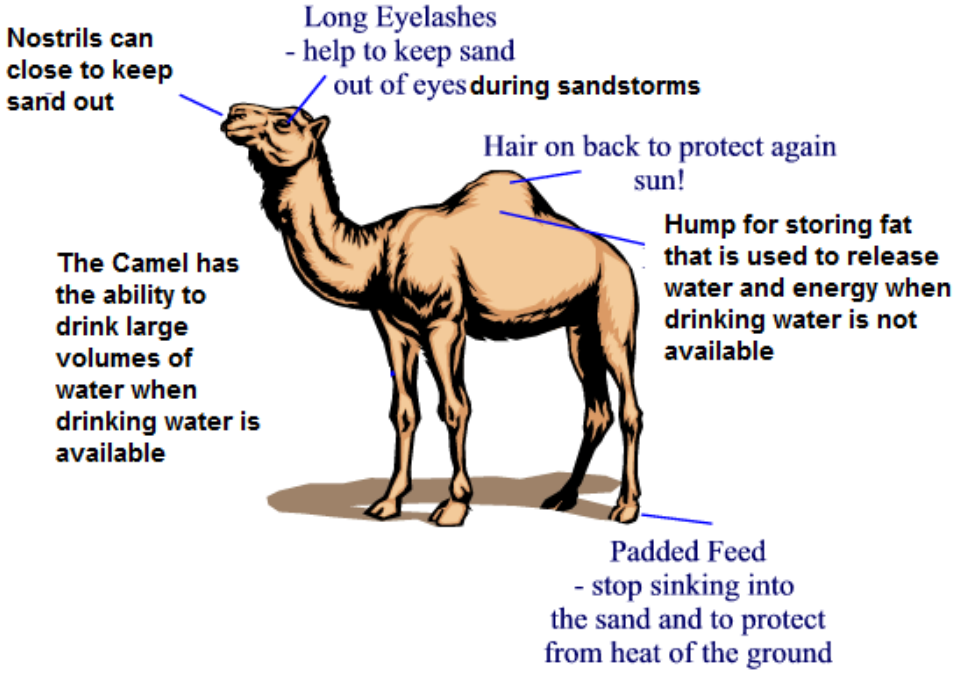
Different environments offer different challenges for survival and reproduction including :

1- Temperature : too hot like deserts or too cold like poles
2- Water : some living organisms can have different adaptive features in regions where lack of water is a problem
3- Lack of light: plants need light for photosynthesis and many animals also need light for vision although you won't find plants in the dark cave, you can find animals that are adapted to living without light.

Fitness : *is defined* as the probability of an organisms surviving and reproducing in the environment in which it is found.

An organism that are better adapted to the environment will have higher fitness.

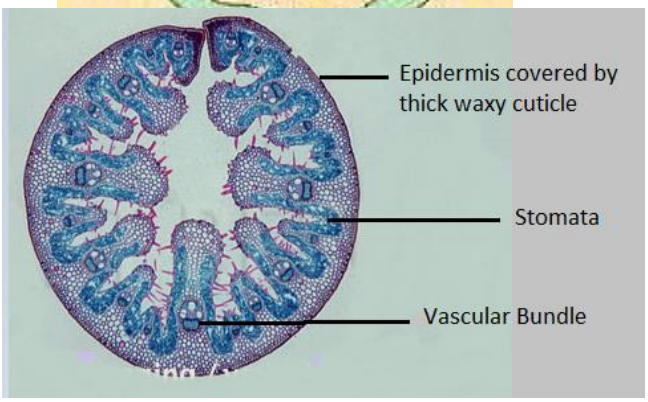
In spring season , mountain hare replaces the thick white winter fur with a thinner brown fur. The winter fur provides camouflage against snow and thick insulation against the cold , the brown fur provides camouflage against bushes and dead grass and is thinner to increase heat loss .
The large ears and all round vision help this animal see predators that are far. The large feet allow it to hold onto snow and rough mountain ground



Plants adapted to dry environments are called **Xerophytes** (from Greek *xero* dry, *phyte* plant) :

1) Leaves have **thick waxy cuticle** which reduces evaporation→ Longer distance for diffusion , not easy for water to pass through
2) Leaves with few stomata that are often **sunken (in pits or depressions)** below the level of the epidermis → this maintains pockets of local high humidity around stomata so they are protected from wind or moving air.
* Usually more stomata are found in the lower epidermis than on the upper because the lower is usually cooler than the upper one.

3) Hairy leaves or **rolled up leaves** to reduce the surface area available for transpiration → Air trapped inside rolled leaves has higher humidity → This lowers the rate of transpiration by reducing the diffusion gradient.



*** This reduces the surface area available for photosynthesis because less carbon dioxide and light can be absorbed.**

- These adaptations help to provide a static (Not moving) layer of air that is saturated with water vapour close to the leaf which will reduce diffusion gradient.
- Fleshy , succulent leaves → Stores water to be used later for photosynthesis
- They usually have either deep roots → To have access for water that drains deep , or they can have shallow roots that spread a long sideways from where the plant is growing → to absorb water immediately after rainfall before it drains

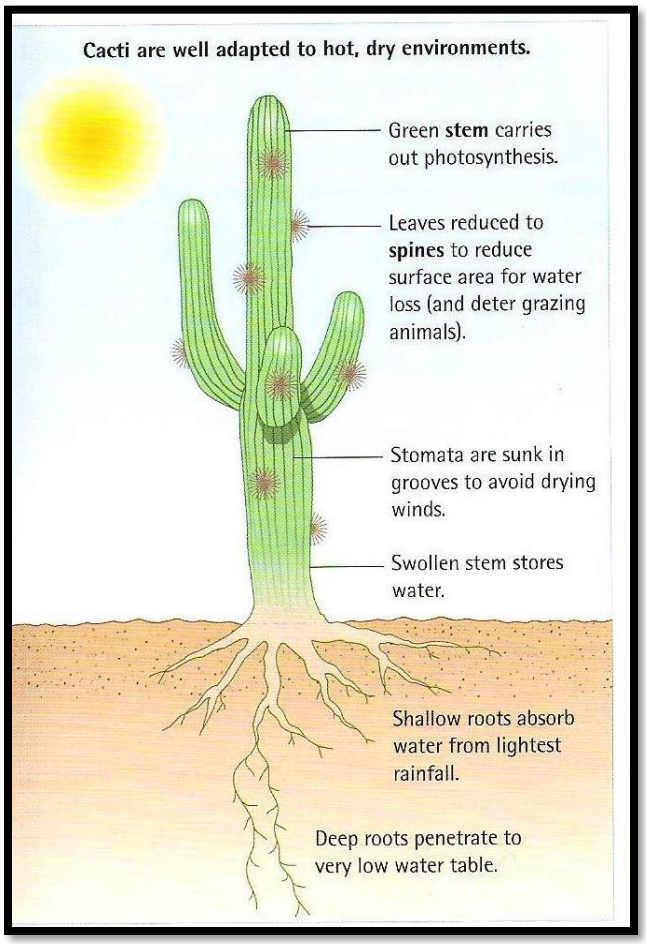
<u>Adaptations of Cacti:</u>

Cacti are adapted to hot, dry conditions in several ways:

1) They often have **needle-like leaves or no leaves at all** → to reduce the surface area for evaporation

* **This also protects them against herbivorous animals**

2) Photosynthesis is carried out by a **thick fleshy green stem that can perform photosynthesis and** can store water to be used later for photosynthesis



4) They have few sunken stomata which close during the day when the temperatures are high and open at night without much loss of water. At night carbon dioxide is absorbed and fixed with an organic acid which is broken down in day time and used for photosynthesis→ because stomata allow diffusion of water vapor so closure of stomata reduces transpiration. → Despite the fact that this reduces transpiration, this can also limit growth, because this prevents cactus from absorbing carbon dioxide **during the day** when light is available. Carbon dioxide is a raw material for photosynthesis which only happens when light is available → little food for growth.

5) **Deep roots**→ Absorbs water than drains deeply and shallow roots → absorb water immediately after rainfall

* Keep in mind that of the rate of photosynthesis = rate of respiration for a very long time , the plant won't be able to grow because all the glucose produced will be consumed in respiration so little will be available for building new cells and for growth .

Hydrophytes : are plants that live in water :

Although its roots are beneath water, its leaves lie at the top where there is enough light for photosynthesis as well as carbon dioxide from the air. Instead from having stomata on the underside of the leaf, most plants have

them on the upper surface because this is where they can be in contact with air
Their stomata is open all the time because there is no problem getting enough water for photosynthesis and transpiration because they live in water .
They have very small roots because they can also extract nutrients from the surrounding water through other tissues.
Water will also support the plant , so the roots will have very little role in keeping the plant in place. .
Their roots also need oxygen for growth that's why the stem **contains air spaces** so that oxygen can diffuse down from the leaves to the roots. The stems have large air spaces which allow the plant **to float on the surface of water** where they can get plenty of light for photosynthesis. There is no need for a thick waxy cuticle , because there is no need to prevent water loss from leaves .
*** If these plants grow excessively in water, they become a serious problem because they block out light , oxygen and carbon dioxide from plants growing in the water.**

Selection

There are two types of Selection	
Natural Selection	Artificial Selection

Mutations and natural selection

Many **gene mutations** are **harmful,** some mutations are considered **neutral** and if they occur in the gametes they might be passed to the offspring without affecting it. On the other hand, **few** mutations are considered **beneficial** to a living organism and are considered as a source of variation, that can increase the chances of survival of living organisms .
Remember that genetic variation arises from :
Meiosis
Fertilization
Mutation

Theory of evolution and how could it have happened?

Variation: most populations of living organisms have individuals that are slightly different from one another. Some of these variations may cause the organism to better adapt to the environment

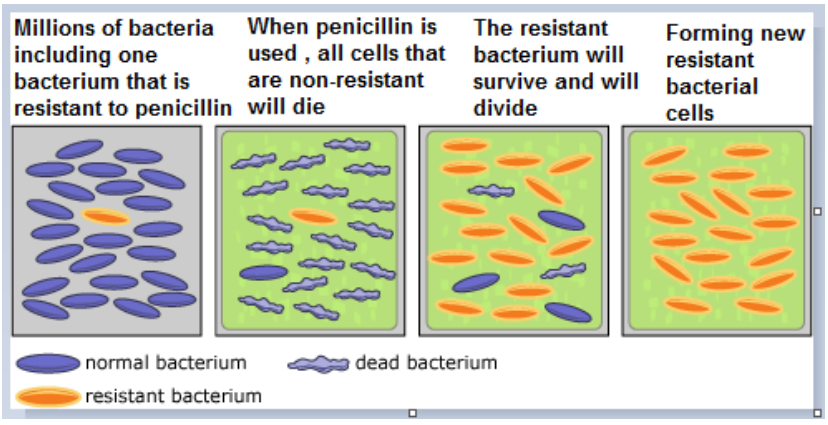
Over production:
Most organisms produce many offspring but many of them fail to survive to adulthood
Struggle for existence:
Organisms compete for the limited resources within the environment.
Survival of the fittest:
Only the organisms which are really well adapted are more likely to survive
And this is due to **selection pressure :** is any reason for organisms with certain characteristics to have either a survival benefit or disadvantage.
Advantageous characteristics are passed on to the offspring:
The well adapted individuals are more likely to breed than those that less-well adapted so they will pass on their genes to the next generation, with time the population gradually loses all the poorly adapted individuals and will become better adapted **and this changing through time is called evolution**

This theory **is called natural selection: the greater chance of passing on of genes by the best adapted organisms**

Evolution is described as the change in adaptive features of a population over time as the result of natural selection

Note**: if the environment does not change , selection does not change**. This will favour the individuals with the same characteristics as the parents. If the environment changes, **or a mutation produces a new allele**, the selection might now favour the individuals with the different characteristics or with the new allele (if they have the an advantage over the others) so they will survive and reproduce and pass on these different alleles to their offspring.

Bacterial resistance to antibiotics and how it takes place:



The process of adaptation is defined as a process, resulting from natural selection, by which populations become more suited to their environment over many

generations and example of this is the development of antibiotic resistance by bacteria.

Antibiotics are drugs that are produced by microorganisms and are used to kill \ stop the growth of bacterial cells without affecting our cells .
e.g. penicillin works by stopping the bacteria from forming cells walls , as a result the bacteria will burst open and die.
In a population of bacteria in an infected person's body, there will be millions of bacterial cells, there will be a chance that one of them has a mutated gene and this makes it resistant to the effect of penicillin this means that penicillin doesn't kill them as quickly as the other bacteria **(variation),** this is considered an **advantageous characteristic** that allows it to adapt more than the other non-resistant bacteria. when penicillin is used (acts as a selection pressure) , The resistant bacterium will survive **(survival of the fittest)** and will reproduce and will pass on the gene of antibiotic resistance for the next generations
(advantageous characteristics are passed on to offspring
So over time there will be an increase in the frequency of the resistance allele
➔ and this is how new resistant bacteria evolve.

Over time, the bacteria have developed resistance to a large number of antibiotics This is a reason why there are so many different antibiotics available so that if bacteria become resistant to one, they may be treated with another.
Many species show also resistance to many kinds of antibiotic (multiple resistance)

- An example of Antibiotic resistant bacteria is **MRSA**. MRSA stands for **Methicillin resistant *Staphylococcus aureus*** . It has been called a "super Bug" because it is resistant to many antibiotics including methicillin , a type of penicillin that is no longer used . It is a major problem in hospitals , where it is responsible for many infections that are difficult to treat .
- **Unless the full course of the antibiotic is taken the few bacteria that are more resistant will survive and reproduce.**

<u>Why antibiotics should not be used very often:</u>
The more we use antibiotics , the more we are introducing a strong selection pressure that allows the resistant bacteria to be the fitter to survive and reproduce more and eventually form populations of bacteria that are resistant to a certain antibiotic → antibiotic will no longer be effective

- Because the antibiotics offer strong selection pressures this can increase the rate at which the evolution of antibiotic resistance occurs.

Sickle cell anaemia and Natural selection

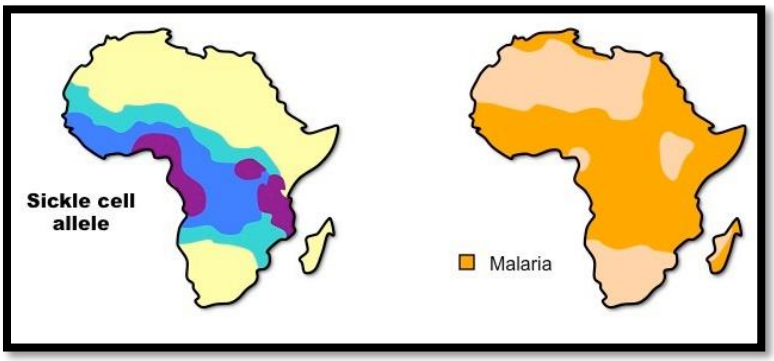
Natural selection doesn't only apply to bacteria, it applies to all living organisms including humans consider this example

As we previously mentioned, sickle cell anaemia reduces the person's lifespan because of its severe effects on the body.

So if sickle cell anaemia is a very serious disease why has natural selection not removed it from the environment?

Heterozygous carries of sickle cell anaemia(Hb^S Hb^A) usually show no or only slight symptoms to sickle cell anaemia regardless of the fact that 40% of HB in their red blood cells being abnormal .

In fact, they can benefit from this condition, that's because a carrier of sickle cell anaemia causes them to have increased resistance to malaria.



The distribution is similar to that for the infectious disease malaria

Explain why the distribution of sickle cell anaemia and malaria are similar.

malaria, is severe disease

→ it is the selective agent

people with sickle cell anaemia / HbS HbN are resistant to malaria;

HbNHbN → homozygous dominant, susceptible to malaria;

HbNHbN more likely to die of malaria before have children to pass on genes

HbN HbS→ sickle cell carriers, → are resistant to malaria (because sickle cells are less prone to infection) and also do not die from sickle cell anaemia → so they can survive and have children and pass on (HBS) Allele

That's why the distribution is similar because there is actually no advantage of HbS in areas where no malaria;

That's why you can notice from the map that in parts of the world where malaria is present people with **heterozygous genotype (** Hb^A Hb^S) are more likely to

survive and will reproduce passing on the sickle cell allele to their offspring, People who are **homozygous for the normal allele** (Hb^AHb^A) died early from malaria and people who are **homozygous for the sickle cell allele** (Hb^S Hb^S) died early from sickle cell anaemia.

That why over many generations, natural selection has maintained the sickle cell gene in the population because of the advantage of being a carrier .

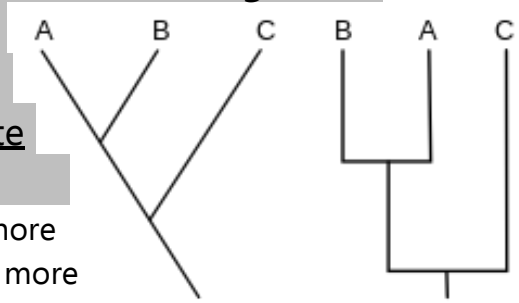
Advantages of Sexual Reproduction

(b)	**1**	maintain / increase, population ;
	2	allows variation ;
	3	(variation) caused by meiosis ;
	4	for example through crossing over / independent assortment ;
	5	random fusion of gametes ;
	6	ability to express recessive traits / AW ;
	7	adaptation to new / changed environments ;
	8	(allows) natural selection / evolution / formation of new species ;

- **Variation in Asexual Reproduction can only be due to mutations**

Classifying Living organisms according to their Evolutionary relationships

- **Scientist can use DNA sequences to determine if two species are closely related .**
- Traditionally , classification of living organisms was based on **morphology and anatomy (presence or absence of wings , Location of bones)** , but studying the Sequences of bases in DNA and of amino acids in proteins are used as a more accurate means of classifying living organisms



➔ Organisms which share a more recent ancestor (are more closely related) have base sequences in DNA that are more similar than those that share only a distant ancestor and have the shortest distance from a branching point on a classification tree e.g. A and B are more closely related to each other

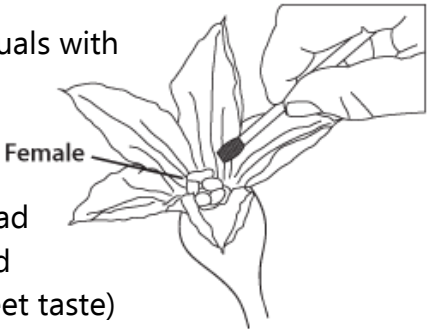
- # Artificial selection (selective breeding)

It is the way humans make use of variations between living organisms, this is done by selecting those individuals with the most useful characteristics and allowing only these individuals to breed → **this is called artificial selection.**

Steps:

– Selection by humans of individuals with desirable features
– crossing these individuals to produce the next generation
– selection of offspring showing the desirable features

Artificial selection in plants . Farmers choose two individuals with two different desirable characteristics and breed them together



For example ,a variety of tomatoes with sweet taste but deformed shape , and another variety with large size and bad taste, the farmer then grows large numbers of offspring and chooses the ones with the best combination (large and sweet taste) and breeds them together .

If he keeps doing so for many generations, eventually he will get a variety of tomatoes with good taste and large size .

- The bad genes will be lost over time

- **Disadvantages :**
  The incoming generations (hybrid generations) might inherit some new bad traits that are dominant and that might appear in the next generations **e.g.** Tomatoes → sweet taste but deformed shape.

The same process can also be done with domesticated animals

For example a farmer may have a variety of cows that have different qualities and quantities of produced milk. The farmer might choose **some cows that are good milk producers and breed them with a bull whose mother or sister are good milk producer by transferring sperm to the cow through artificial insemination , the offspring (cows) with the improved milk yield are chosen , these cows are then used to breed the next generation**

. The farmer can continue this selective breeding for many generations each time picking only the best milk producers to be the mothers of the next generation. The milk production of this herd will gradually increase.
Other applications

- It can also take place to develop the breed of dogs that make good pets
- Increase the size of chicken eggs.
- Increase the size of farm animals kept for meat

➔ This can take a longer than plants , as animals generally take longer to become mature enough for reproduction

Note: Artificial insemination can be used for this process

- Note : Plants of the same species but with different characteristics are called **varieties**
- Animals of the same species with different characteristics are called **breeds**

Differences between artificial and natural selection:

1. In artificial selection , humans are the agents of selection
2. Artificial selection is much quicker
3. Artificial selection offers no advantage to the animal or plant in its environment

Continuous and discontinuous Variation

Variation: it is the difference in the characteristics of members of the same species, so the offspring would have a variety of different phenotypes

Because variation exists between members of the same species we say that they show **phenotypic variation** because their features look different
Some of these differences can be caused by differences in genes which is **genetic variation**

There are two type of phenotypic variation :

1) Continuous variation
2) Discontinuous variation

1) Continuous variation :

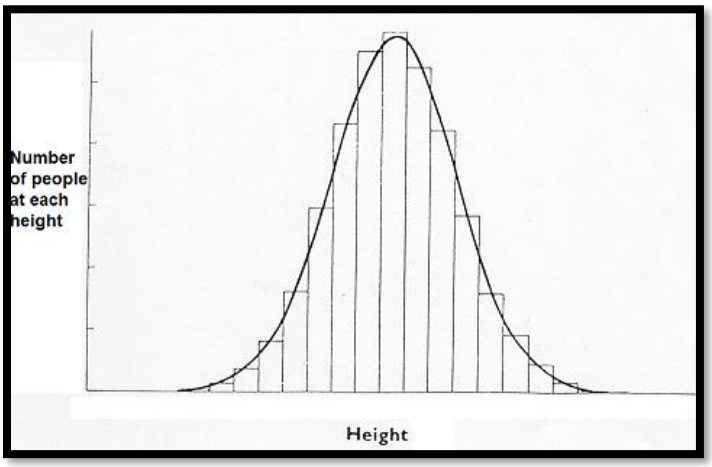
It is the type of variation, where there are many intermediate phenotypes for the trait between the upper and lower extremes of the phenotypes.

Examples include: Body weight, Body height, skin colour , length of hair …

- It is quantitative and it can be measured

- In these traits there is a continuation between each phenotype and the one after. Continuous variation is usually controlled by genes and the **environment** and can be represented by **histogram.**
- **These characteristics are usually quantitative – they can be measured.**

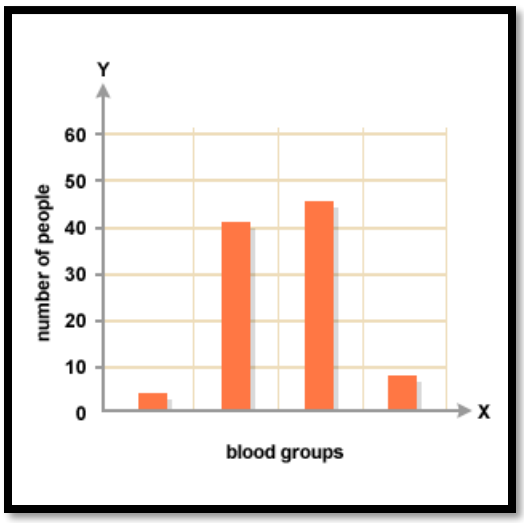
E.g. human growth is influenced by genes that influence protein synthesis in muscles, bone development and production of hormones. But growth will only take place if an individual is taking in a balanced diet.



If we take height as an example, the heights of people in a population will look similar to this
This is called **<u>normal distribution</u>** where most people come in the middle of the range and fewer people and the lower and upper end.

<u>Discontinuous Variation :</u>

This variation shows **no intermediate forms** between the two extremes of phenotypes, or sometimes it shows few intermediate traits.
E.g. Sex in humans (Male/Female), Blood Group (A, B, AB, O) , tongue rolling
* It is qualitative



- This Kind of variation is represented by **Bar charts**
- Discontinuous variation **is only affected** by genes and is **not affected** by environment.

- Other examples on discontinuous variation: haemophilia , sickle cell anaemia.

Phenotype = The observable characteristics of an organism	**Genotype + The full set of genes it possesses**	**Effects of the Environment**

- Characteristics caused by the organism's environment are called **acquired characteristics e.g. skin colour** , they are not caused by genes so they cannot be passed on to the next generation

Genetic Engineering

Genetic engineering as changing the genetic material of an organism by removing, changing or inserting individual genes.

- **Insulin Hormone:**

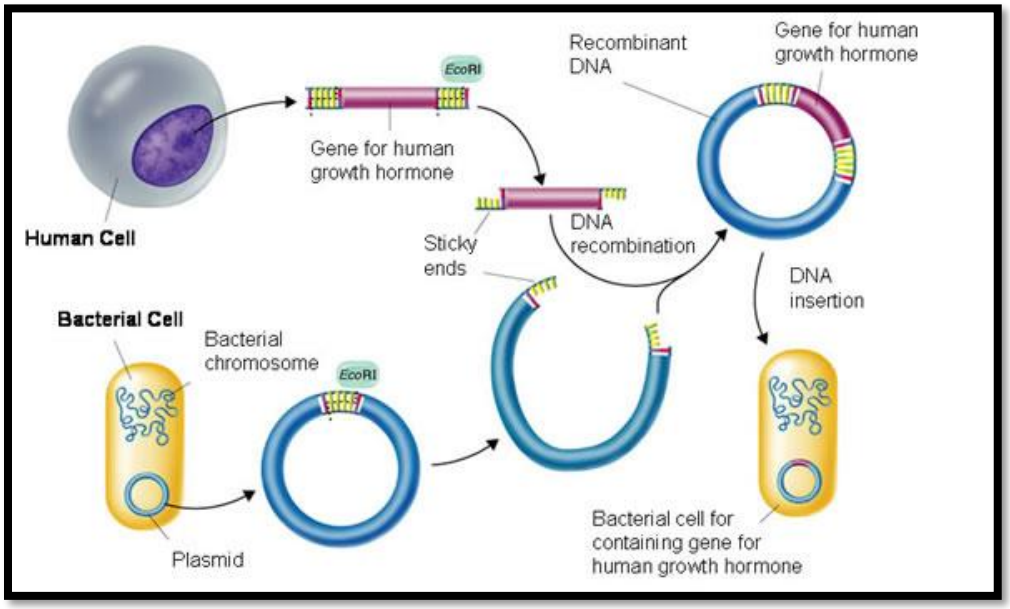
In the past, the only source of insulin was from animals which have been killed for food e.g. pigs but now, it can be produced by genetic engineering.

What are the advantages of using hormones (e.g. insulin) produced by genetic engineering rather than from dead animals?

1) Genetic engineering produces a protein hormone from a human gene; this hormone is likely to function more efficiently in a person.
2) Is less likely to be rejected by the body
3) Hormones e.g. insulin can be made in large amounts making it less expensive and more readily available
4) No risk of infection from animals to humans

Steps for Hormone (e.g.Insulin) production by genetic engineering

1. A part of DNA which represents the insulin hormone production gene (e.g. gene coding for insulin hormones in a pancreas cell) is cut off by **Restriction enzyme forming sticky ends .**



The plasmid which is a small circular DNA in bacteria is cut open at specific points using the same **restriction enzyme forming complementary sticky ends** .

2. The cut off gene will be joined to the plasmid by the enzyme **ligase** forming **recombinant DNA** (which is the human

gene plus the bacterial plasmid) Which is then inserted into the host bacterial cell → the bacteria is now called **genetically modified organism (GMO)/ transgenic bacterium**

3. The bacterial cell will now contain the new plasmid that contains **the insulin production gene**.
4. The bacteria containing the desirable gene is then cultured in a fermenter containing a nutrient medium under optimal pH and temperature to allow it to divide and produce large populations capable of producing the gene protein product (insulin)

This method can be used to produce different gene products like insulin , , sex hormones and human growth hormone (to restore normal growth) and hepatitis B vaccine

Hepatitis B vaccine : Yeast cells can be genetically modified to produce the surface proteins (antigens) of the hepatitis B virus, when the vaccine is injected into a patient , their body makes antibodies against the proteins , so the person becomes immune to the virus.

Go back to pages 19-21 (Human Biology Book) and study " producing genetically modified bacteria" .

Other examples of genetic modification

Crop plant such as maize or wheat, have been **genetically modified** to contain a gene from a bacterium that produces a poison that kills insects , so when the insects like caterpillars eat the plant these pests will be killed→ **Increased yield**

- Crop plants have been genetically modified to make them **resistant to herbicides,** this means that when the herbicide is sprayed over the growing crop it only kills weeds without harming the main crop
- Some crops have been **genetically modified to contain additional vitamins→ Increased Nutritional value**
- **→ Improved customer satisfaction / shelf life;**

Example : Golden rice have been genetically modified to contain genes that produce a chemical substance called Carotene that is turned into vitamin A in the human body

This type of plant can be grown in regions where people have diseases caused by the lack of vitamin A.

Genetically Modified Crops :

Advantages and Disadvantages of growing GM crops such as soya, maize and rice :

Advantages

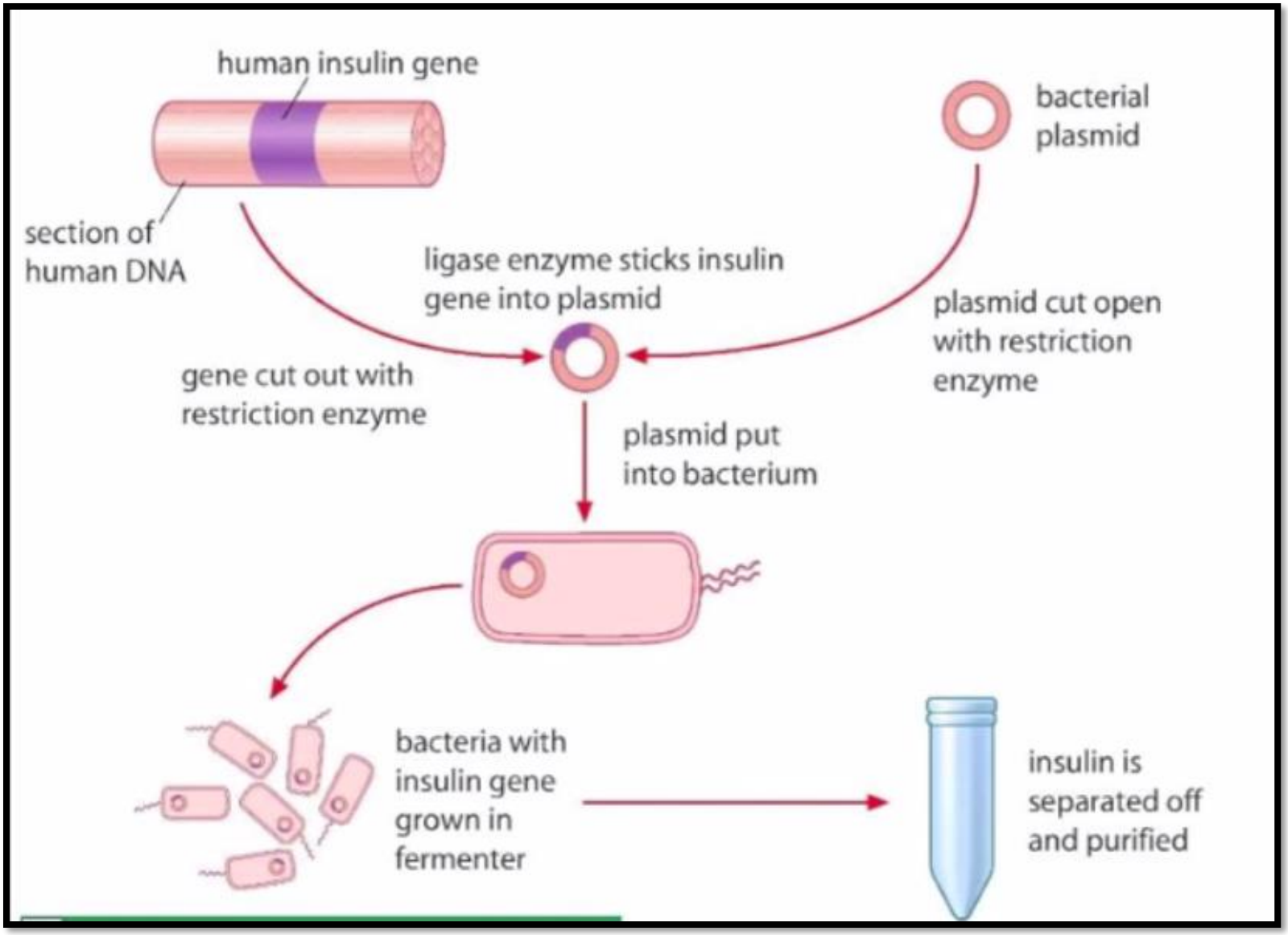
1) Less use of chemicals like herbicides and pesticides which is better for other living organisms in the environment and reduces time and cost
2) Increases the yield of crops because there will less damage from pests and less competition with weeds for water and nutrients

Disadvantages :

- Increases the cost of seeds for poor farmers
- The gene may be transferred to other plants by pollination When this pollen is around other plants, even things like grass or weeds, they cross pollinate. This could develop "superweeds" that won't be affected by herbicides
- Increased dependency on the use of herbicide that the GM plant is resistant to → so this leads to increased costs for farmers
- Many people argue that GM crops are unsafe for eating → but there is no evidence to support this

Bacteria are useful in biotechnology and genetic engineering due to their rapid reproduction rate and their ability to make complex molecules
Why bacteria are useful in biotechnology and genetic engineering:

- They have plasmids which are small and easy to extract
- The bacterial plasmid is made up of DNA – the same as the DNA found in the chromosomes of plants and animals this means that all organisms share the same genetic code → a gene that codes for a protein in one organism will code for the same protein in other organism lack of ethical concerns over their manipulation and growth


human insulin gene
section of human DNA
bacterial plasmid
ligase enzyme sticks insulin gene into plasmid
plasmid cut open with restriction enzyme
gene cut out with restriction enzyme
plasmid put into bacterium
bacteria with insulin gene grown in fermenter
insulin is separated off and purified

Topic 14

Human Diseases

Immunity and diseases

Immunity: The resistance of the body against pathogens either by phagocytes or by the production of antibodies.

There are two ways by which the body fights against pathogens

Natural Immunity	Acquired Immunity

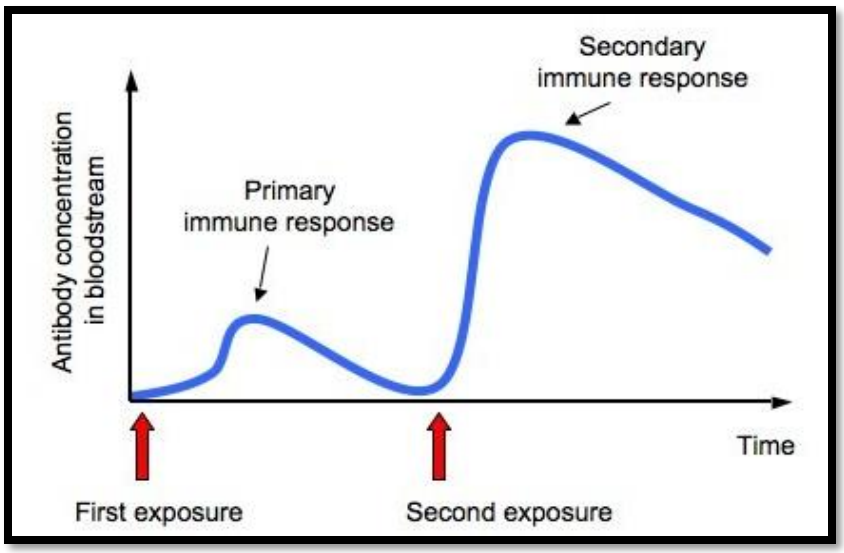
Natural immunity (innate immunity/ non-specific immunity)

a. **The first line of defense :**
   i. **Body Surfaces (skin)** which prevent the entry of pathogens
   ii. **Eyes :** Lysozymes in tears (Destroys bacteria)
   iii. HCL in stomach → Kills bacteria
   iv. **Cilia and mucus** in the nasal passages
b. **The Second line of defense :**
   Phagocytes: certain white blood cells, which engulf and digest bacteria by phagocytosis.

Acquired immunity (Specific immunity)

c. **The third of defense :**
   Antibodies produced by lymphocytes

- **Acquired Immunity: Acquired immunity:** Immunity acquired by infection or vaccination (active immunity) or by the transfer of antibody or an immune donor (passive immunity)
- **There are two types of Acquired immunity :**
  - **a.** Active immunity
  - **b.** Passive immunity



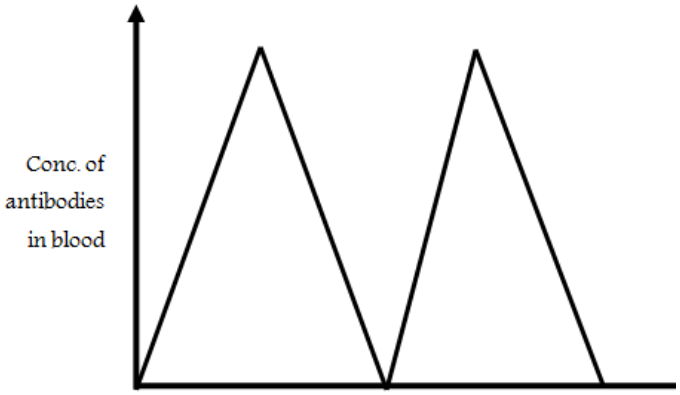
- **What are the differences between Active and passive immunity :**

Active Immunity

- It takes few weeks for the antibody level to build up
- Long lasting Immunity

- Booster Vaccines : A single **dose** of some **vaccines** provides lifelong immunity to most people, while other **vaccines** require additional **doses** (**boosters**) in order to increase immunity back to the protective levels . because the numbers of memory cells decrease over time

Passive Immunity



- Immediate high level but lasts for short time

Disease: it is a condition with a specific cause in which part or all of the body functions abnormally or less efficiently.
The cause can be **unhealthy activities** such as smoking or drinking alcohol, it can be **genetic,** such as the **mutated gene** responsible for sickle cell anaemia , or it can be due to a microorganism . Many

microorganisms cause **infectious diseases also called Communicable Diseases** (these can be transmitted from one person to another)
Pathogens : microorganisms that can cause disease

- **Non- Infectious Diseases: e.g.** lung cancer and COPD . They are not caused by pathogens. Inherited or genetic diseases . There are other categories of noninfectious disease, including deficiency diseases that are caused by malnutrition and mental diseases

Type of microorganism	How it can cause disease	Examples of diseases it can cause
Bacteria	Releasing toxins as they multiply, these can affect the region of infection and also in other body parts	Typhoid , Tuberculosis Gonorrhea
Viruses	They enter a living cells they affect the metabolic reactions of the cell , the genetic material of the virus instructs the cell to produce more viruses	Influenza , Poliomyelitis (polio) , Human immunodeficiency virus (HIV) , common cold
Fungi	Fungi are decomposers; The hyphae of fungi secrete digestive enzymes into tissues causing damage.	Thrush , Athletes foot
Protozoa	There is no one specific way of causing disease	Malaria

- ## The general course of a disease :

1. Infection :

Are the methods by which pathogens get into the human's body.

There are several ways by which pathogens enter the human body:

- **Droplet infection :** respiratory diseases such as common cold , influenza are usually carried in tiny droplets through the air (airborne) when a person sneezes or coughs and they are inhaled by others
- **Food borne:** by eating contaminated food, most food poisoning is caused by bacteria but some viruses can also be transmitted by this way e.g. polio
- **Waterborne:** when a person drinks contaminated water

- **Direct contact**: many skin infections are transmitted when a person makes direct contact with an infected person e.g. athlete's foot
- **Sexual intercourse:** organisms infecting the sex organs can be passed through sexual intercourse
- **Blood-to-blood contact**: like AIDS, Hepatitis B
- **Animal vectors:** for example some diseases are transmitted by the bites of insects, or when flies carry microorganisms from faeces to food

2. **<u>Incubation period :</u>**

It is the period of time between when a person is first infected with the pathogen and when they first show signs and symptoms of the disease.
During this period the person may not feel sick but can be infectious to others.
Incubation periods vary according to the disease ranging from hours to months.

The Duration of the incubation period varies according to :

- **Immunity of host**
- **Specific microorganism involved**
- **The number of infecting pathogens (infectious dose)**

<u>What happens in the body during the incubation period ?</u>

- Replication of pathogens rapidly in cells and body tissues.
- Production of toxins by pathogens which then circulate through the blood stream.
- Cell destruction: Pathogens may destroy body cells.

3. **<u>Signs and symptoms :</u>**

Signs are of a disease can be visible to people, it can be seen, heard or measured e.g. such as coughing, losing body weight, skin rash , blood pressure
Symptoms are the complaints made by a patient which cannot be seen by the medical observer, e.g. feeling of pain, nausea or dizziness.

<u>Distribution of diseases:</u>

- **<u>Endemic disease :</u>**

It is a disease, which is always present in a population of a particular area, giving rise to a steady flow of cases every year. For example, malaria is an endemic disease in Africa

- **Epidemic disease :**

It is a widespread outbreak of an infectious disease with a **large number of people becoming infected at the same time** over a large area.
For a disease to be considered an epidemic, **there must be an increase in the area affected not just the number of people affected.**

- **Pandemic :**

If the disease spreads across the world it is called a pandemic, just like swine flu that had spread worldwide in 2009.

Go back to grade 9 and study ; viruses and bacteria and fungi

Diseases caused by viruses (Ebola and AIDS):

1. Acquired Immunity deficiency Syndrome (AIDS):

- **Go back to Human Biology Book pages 234-235 and study HIV**

It is caused by HIV virus (**Human immune deficiency Virus**) which attacks the body's immune system, this virus **invades the lymphocytes** and in particular **helper T cells**. These cells required to activate other white blood cells (B lymphocytes and other T cells) to start fighting infections. After years of infection with the virus, the numbers of T-cells will be greatly reduced (the body will not be able to replace the lymphocytes at the same rate as they are being destroyed) as a result your body won't be able to fight against pathogens effectively.

Latency Period : the period during which the body replaces the lymphocytes as fast as they are destroyed . It can last for up to 20 years . The person shows no symptoms of AIDS during this period , but will be highly infective to others .

- **After about 10 years of HIV infection, a person is likely to develop AIDS if not given effective treatment.**
- The person would be exposed to infections and the body cannot defend these diseases. Some of these diseases cause death such as **Pneumonia and tuberculosis.** The person is also **more prone to cancer,** because one of the functions of the immune system is to destroy body cells that are becoming cancer cells.

Symptoms:

A. Fever
B. Diarrhoea
C. Weight loss
D. Weakness
E. Swelling of lymph nodes in the neck and spleen
F. Fungal infection in the mouth

- **Methods of transmission :**

a. During unprotected sexual intercourse
b. During blood transfusion using blood containing the virus.
c. A pregnant woman can pass it on to her baby through the placenta
d. By sharing needles and syringes (Drug misuse)
e. Using unsterilized tattooing and ear piercing machines
f. Sharing razors or toothbrushes (gums may bleed)

Note: HIV is not transmitted through kissing, or sharing cups

- **Methods of prevention**

- **Using condoms and limiting the number of sexual partners**

And Avoiding all the above

- **Treatment :**

No drug or cure is available has been found for the disease but scientists have found that some chemicals and medicines can slow down the reproduction of the virus inside the cell , No vaccine has been developed either.

2. EBOLA

Ebola is a severe, often fatal disease , caused by **a virus .**

The incubation period for Ebola is 2-21 days, and humans are not infectious until they develop the symptoms.

Symptoms

The first symptoms are fever , muscle pain , headache and a sore throat . These are followed by vomiting , diarrhoea , a rash and damaged liver and kidney function .

In some patients, there is internal and external bleeding (blood oozing from the gums and blood in the faeces). Lab tests show low white blood cell and platelet count.

It is believed that the natural hosts of the virus are the **African fruit bats.** Ebola was probably introduced to the human population through contact with the blood or tissues of the bats or other infected animals such as chimpanzees , monkeys .

Methods of transmission

- Direct contact with the body fluids (e.g. Blood , saliva , semen , faeces)
- Contact may occur through broken skin , or through mucus membranes such as the mouth and intestines .
- Infected materials such as bedding

- The disease is highly infectious and even well protected health workers who practice strict control precautions have become infected

Treatment :

At the moment , there is no proven drug treatment for Ebola , and no vaccine to prevent people becoming infected

Oral rehydration therapy, Patients can also be **given fluids intravenously** (Fluids transferred directly into a vein)

Prevention and control

Dealing with individual patients , checking their contacts , having laboratory testing facilities and organizing safe burial of people who have died from the disease . The key to success is engaging the help of people where the outbreaks happen and making them aware of the risk factors

The main preventive measures are :

1) **Reducing the risk of transmission of the virus from animals** – e.g. avoiding contact with fruit bats and monkeys , wearing gloves and thoroughly cooking meat to destroy the virus
2) **Reducing the risk of transmission from infected patients –** e.g. wearing gloves and protective clothing , regular hand washing and other hygiene measures
3) **Reducing the risk of transmission by sexual intercourse** – Survivors of the disease are advised to practice " safe sex" using condoms for 12 months after the disease , or until their semen sample twice tests negative for the Ebola virus
4) **Containing the outbreak** – Including safe burial of the dead , identifying and monitoring patient contacts , and isolating sick people to prevent the spread of the disease .

Diseases caused by bacteria :
Go back to grade 9 and study " the bacterial kingdom"

Bacterial Diseases: (Typhoid , Cholera and Gonorrhoea)

1. Typhoid or Typhoid fever:

It is caused by bacteria called ***Salmonella typhi*** .
Has an incubation period of 2 weeks

- **Symptoms :**

a. starts with flu-like symptoms ; fever , cough and general weakness
b. Later on, the person suffers from stomach ache , constipation or diarrhoea and vomiting → **diarrhoea can lead to severe dehydration**
c. The bacteria can attack the alimentary canal and cause damage and bleeding.
d. The bacteria can also release toxins that cause inflammation of the heart and can cause many organs to fail .

- **Methods of transmission**

a. Through contaminated water
b. Flies can carry bacteria from faeces to food
c. Drinking water that is contaminated by human faeces

That's why the disease is most spread in some developing countries where human faeces or urine come into contact with food or drinking water

d. Food that has been handled by carriers of the disease

Methods of prevention:

- **Washing hands before handling food ,** because hands may be covered by bacteria , washing removes bacteria so that it can't be transmitted to food
- **Keep the food covered** , to stop flies transferring bacteria
- Good hygiene and public sanitation are the best methods for preventing this disease (purification of water supplies / safe disposal of sewage / clean food handling
  a. By vaccination with dead microorganisms

- **Treatment :**
- by using antibiotics
- **Oral rehydration therapy** → to reduce the dehydration effects of diarrhoea

Housefly and typhoid Bacillus:

Houseflies are attracted to animal or human faeces and can transmit many viruses and bacteria on their body or in their saliva.
The housefly is the **vector** which is responsible for the transmission of the pathogens (bacteria) causing dysentery, Typhoid, cholera, food poisoning and other gastro-intestinal diseases.

What is a vector?

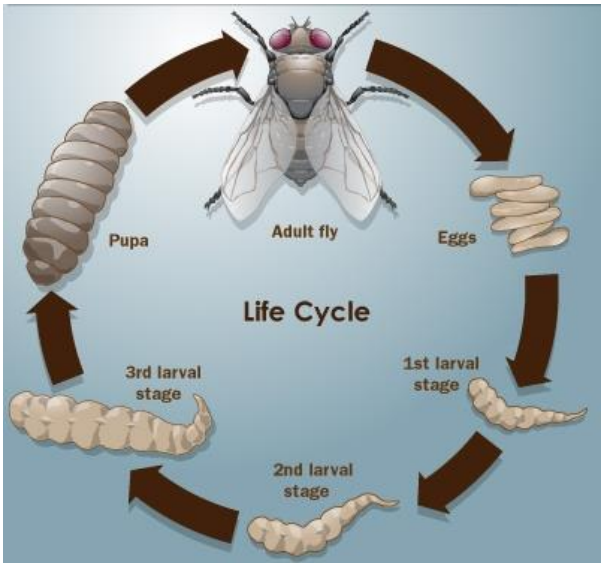
It's an organism that transfers the pathogen from one host to another without being harmed by the pathogen.

- **Methods of transmission of the disease by housefly :**

a. **Hairs on the body of the insect trap bacteria** which are deposited on food
b. Houseflies do not bite humans but they feed on human food if it is uncovered, as they feed , **they release saliva** onto food through their **proboscis** , the parasitic microorganisms can

be transmitted into humans when they eat the food

c. **Feet form a natural trap for bacteria** which are left on human food.
d. Faeces containing many bacteria may be left on food or be dropped onto food when the insect is flying over it.
e. They can fly long distances and spread the disease more widely



- **Methods of Control :**

a. Destroy all adult flies by **insecticide**
b. **Cover all food to prevent contamination by flies through proper food storage , screens**
c. **Eliminate breeding grounds** such as organic rubbish heaps ,use dustbin with lids
d. Washing hands before handling food; preparing and eating food.
e. Clean water supply (treatment of water supply)
f. Vaccination is essential for anyone visiting countries with ineffective water treatment facilities.
g. Safe disposal of sewage

3. Gonorrhoea:

It is one of the Sexual transmitted diseases (STDs)

It is caused by bacteria called ***Neisseria gonorrhoea*** which invades the urethra and results in inflammation, leading to pain on passing urine especially in males. In females the vagina is the most likely organ to be invaded. This disease is transmitted during sexual intercourse and can pass from an infected mother to her baby during birth through the mother's lower birth canal

- **Symptoms :**

a. In males: discharge of pus from the urethra leading to the blockage of the urethra which leads to infertility, in addition, the person may suffer from pain during urination and inflammation of urethra.

b. In females: discharge from the vagina or bleeding between the menstrual periods → If untreated the disease can spread to the uterus, oviducts and ovaries which leads to infertility

c. Inflammation of joints.
d. Blindness (affecting newborn children).

- **Methods of transmission :**

a. Sexual intercourse
b. Infected mother transmits disease to her child during birth
c. By direct contact with an infected person or using the underwear of an infected person.

- **Prevention :**

a. Men should use condoms during sexual intercourse
b. Avoid having sex or reduce the number of sexual partners

- **Treatment**

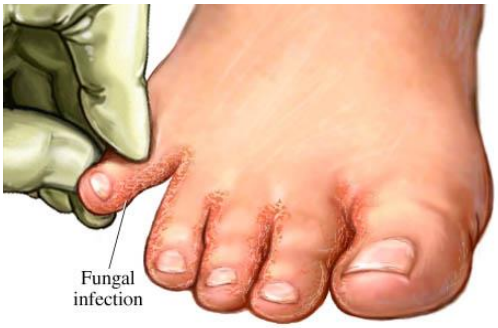
Taking antibiotics – though the bacteria have started to evolve resistance to antibiotics

Not all bacteria are harmful, some play very important beneficial roles e.g.:

1. **Recycling nutrients:** Decomposition of waste materials, In addition it adds nutrients to the soil making it more **fertile**
2. **Sewage treatment** :(both aerobic and anaerobic respiration) to get rid of harmful substances.
3. Some bacteria are used in food industries such as **yoghurt making** and **vinegar making**
4. Some bacteria are used in **genetic engineering** for example: insulin hormone can be made in certain bacteria and then extracted to be used in treatment of diabetes.
5. **Antibiotic manufacture**

2.Athlete's Foot:

It is caused by a **fungus** that attacks the warm, moist,soft skin between toes, causing irritation and inflammation. It is very common in adult males than females and children.



- **Symptoms :**
- The fungus feeds on the layers of the skin causing :
  a. Swollen ,red and sore pealing skin between the toes that can be subject to **secondary bacterial infection**
  b. Itching

- **Methods of transmission :**
  a. **Fungal spores may be transmitted to the skin from the air or floor (** it is commonly picked up in sports changing rooms and swimming pools)
  b. By contact with infected towels or clothing or by using things that belong to an infected person

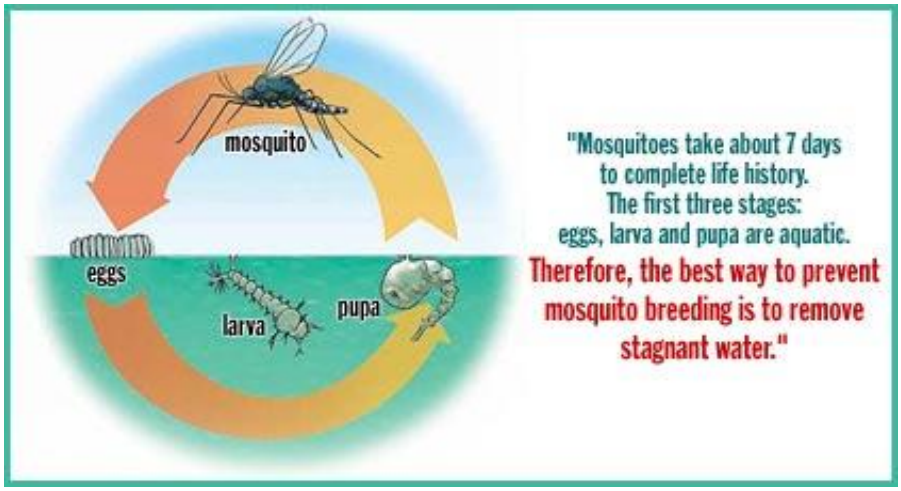
- **Methods of prevention :**
  a. Shoes should be light and ventilated
  b. Avoid sharing towels
  c. Avoid walking barefoot in public showers and swimming pools
  d. Regular washing of changing room's floors with disinfectants
  e. Keep toenails cut short
  f. Careful drying of the feet and between the toes.
- **Treatment :**

a. By using fungicidal drug
b. Using antibiotics (for secondary infection)

After an infection , it is virtually impossible to kill the fungal spores in shoes and socks , so to avoid re-infection , these should not be worn again.

Diseases caused by protozoa:

Protozoa are **single-celled organisms** that consist of animal-like cells and contain organelles that are found in animal cells. Protozoa are free-living organisms that live in any wet or moist



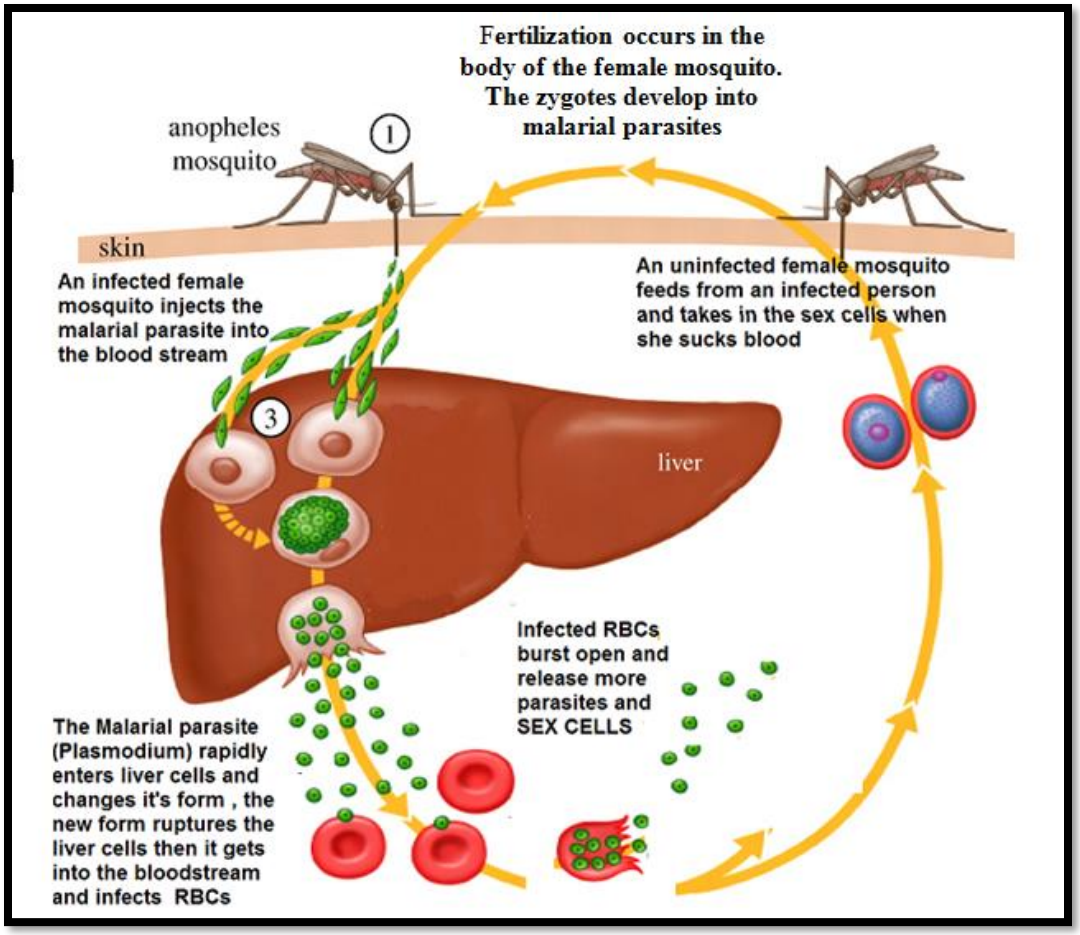
habitat like ponds, rivers and seas E.g. amoeba, and *Plasmodium*

- **Mosquito and malarial parasite :**
  Malaria is caused by a protozoan called ***Plasmodium***, and is transmitted by the female ***Anopheles*** **mosquito** .
  Malaria is very common in tropical areas, these areas offer **wet and warm** conditions that the female mosquito requires to lay eggs and breed.
- The malaria parasite spends from 2 weeks to several months in the liver, before infecting red blood cells. **The malaria symptoms then appear**
  a. Cold sweat and Fever
  b. Joint Pains
  c. vomiting
  d. Severe anaemia
  e. Severe malaria causes coma and even death if combined with effects of starvation

Lifecycle of malarial parasite

The parasite is found in the salivary glands of the female *Anopheles* mosquito. When the mosquito bites, it injects saliva containing an **anticoagulant** that prevents the blood from clotting, so facilitating feeding. At each bite ,hundreds of parasites may be injected to the bloodstream, the malarial parasites enter liver cells and change form, the new form ruptures the liver cells, escapes into the bloodstream and infects the red blood cells **where it feeds on haemoglobin** and **reproduces asexually**.

From time to time infected red blood cells burst open r**eleasing male and female sex cells (gametes)** and **more parasites which infect more red blood**



cells and toxins causing periodic fevers. When an uninfected mosquito sucks blood from a person with malaria , it ingests some of these **gametes**, Fertilization occurs in the body of the female mosquito → **zygotes are formed, the resulting zygotes develop into malarial parasites in the saliva of the mosquito** . The cycle is repeated when The Infected mosquito injects the malarial parasite as it feeds on the blood of another uninfected person.

- **Control and prevention :**

a. Controlling the numbers of mosquitoes →so that fewer insects will be able to transmit the pathogen by :

- Drain all stagnant water to eliminate breeding grounds
- Stocking ponds with a fish called *Tilapia* which feeds on the larvae of mosquito (biological control)
- **Draining swamps and speeding up the flow of streams** to wash away eggs, larvae and pupae.
- **Spray insecticide on water** and buildings to kill adult mosquitoes
- Prevent bites, by **putting insect repellents** on skin, by covering the skin with long-sleeved clothing, sleeping under mosquito nets and by providing screens and mesh on windows to prevent the entrance and bites of the adult mosquitoes.
- Spray oil on water to reduce surface tension so larvae and pupae of insects sink and suffocate because oil acts as an isolating layer for oxygen causing death of larvae.

b. Controlling the number of plasmodium parasite so there will be fewer opportunities for the mosquito to transmit it to humans

- Taking anti-malarial drugs in malarial area as a matter of routine and precaution, to kill any parasite as it enters the body.
- Treatment and isolation of carriers identified by microscopic examination of blood smear.

Note: it is very hard for our immune system to attack the malarial parasite because it stays much of the time hidden in the red blood cells and changes its form several times inside our bodies

- **Treatment :**

By means of anti-malarial drugs

Heterozygous for Sickle cell anaemia allele have malarial resistance why ?

The malarial parasite infects red blood cells and spends part of its lifecycle inside them. Being a carrier of the sickle cell anaemia allele causes 40% of the

haemoglobin in red blood cells to be abnormal, these red blood cells tend to be **slightly more fragile than normal cells,** that's why when the parasite enters the red blood cells of a carrier, the cells **usually burst open before the parasite has had enough time to develop** → the parasite dies → the lifecycle will be interrupted.

• Methods of food contamination

☒ **Methods of food contamination :**

- **Dirty hands:** micro-organisms can be transmitted from the urethra to the mouth. This can be prevented by washing hands after using the toilet.
- **Carriers of the pathogen (infected people):** They should be prevented from handling food.
- **Animal vectors (housefly, mosquito, rats, dogs, etc):** These should be controlled e.g. Biological control, pesticides.
- **Poor Food storage:** proper methods of food preservation should be used to avoid contamination of food.
- **Poor food preparation :** food should be cooked thoroughly
- **Unhygienic practices in kitchen , food stores and restaurants**
- **Poor sewage disposal:** this can also affect the clean water supply. It should be prevented by good sewage disposal, away from clean water.
- **Poor water purification**
- **Poor waste disposal :** good medium for reproduction of vectors

- **Methods of prevention of transmission of food-borne diseases :**
- Washing hands before handling food , because hands may be covered by bacteria , washing removes bacteria so that it can't be transmitted to food
- Keep the food covered , to stop flies transferring bacteria
- cook food thoroughly to kill bacteria
- Storing food in a refrigerator to **slow down** bacterial reproduction
- Wash working surfaces and utensil before use To remove bacteria
- Cooked and raw food should not be stored together (bacteria in uncooked food can be transferred to cooked food)

☒ Food preservation :

Prevents the activity of decomposers (bacteria and fungi) because it stops its growth and reproduction in food to halt (inhibit) the decomposition of food by micro-organisms' enzymes. If food isn't preserved it would have an **unpleasant smell**, **unpleasant taste** in addition to being **contaminated by the toxins** released which cause dangerous diseases that may kill.

- **Requirements of food microorganisms :**

1. Moisture
2. Warmth (temperature)
3. Oxygen and food

- **Food is preserved by storing it in conditions where moisture ,water and oxygen are absent**

Go back to page 72 (HUMAN BIO BOOK) and study methods of food preservation