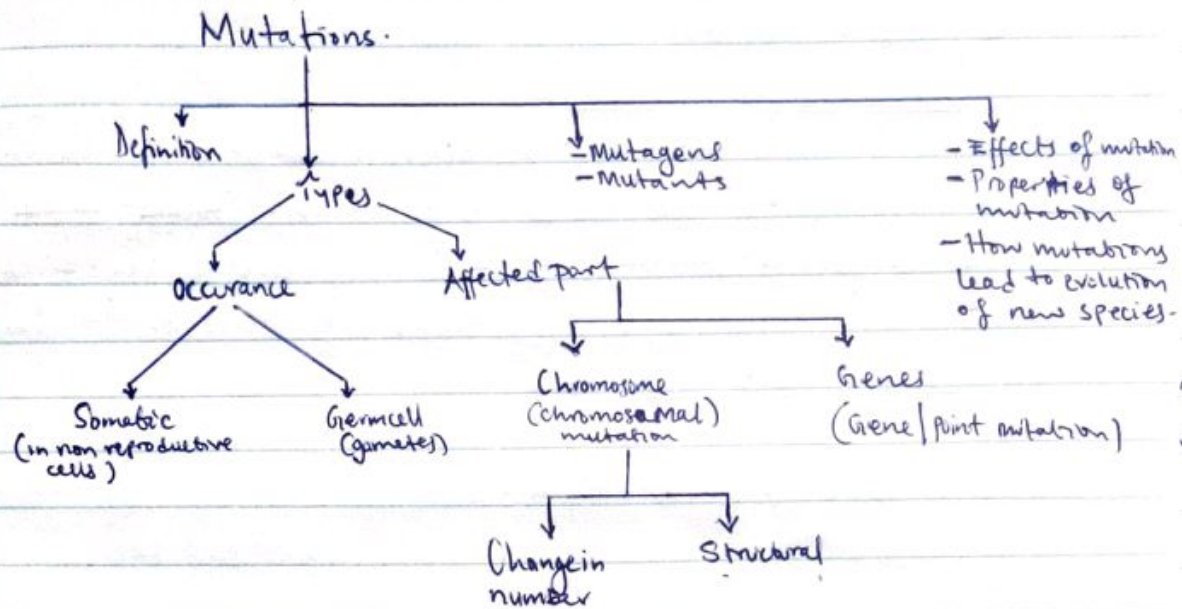


# MUTATIONS



**Defn:** Mutations are sudden changes in the DNA or genetic material of an organism.

**OR** Mutations are sudden changes in the amount and structure of chromosomes and genes of an organism.

The organism whose genetic material has mutated is referred to as a Mutant.

Substances which accelerate mutations are called Mutagens. They include  $\alpha$ -rays, Gamma rays, U.V radiations, Mustard gas, Tobacco, Pesticides, Caffeine, food preservatives, high temperature, formalin etc.

The human conditions resulting from mutations include:

- Haemophilia
- Colour blindness
- Sickle cell anaemia
- Down's syndrome
- Turner's syndrome
- Kline felter's syndrome

## PROPERTIES OF MUTATION

- They are spontaneous i.e. occur randomly without prior warning and pattern of occurrence.
- They are harmful
- They cause permanent changes
- They are rare
- They are abrupt



# TYPES OF MUTATIONS

## (a) Basing on Occurance

### (i) Somatic Mutations

Occur in non reproductive cells and hence can't be transmitted to the next generation. They often result in the different forms of ~~to~~ In organisms that reproduce asexually, somatic mutations are passed to daughter cells.

### (ii) Germ cell mutations

They occur in gametes and hence can be transmitted to the next generation.

## (b) Basing on the Part affected within the nucleus.

### (i) Chromosomal Mutations

• They occur within the chromosomes and can involve either change in their structure / change in their number.

- Chromosomal mutations involving change in number.  
There are two forms; i.e. Aneuploidy and Polyploidy.

#### \* Aneuploidy

It involves loss or gain of one or more chromosomes but not in multiples of the haploid chromosome number of normal being. E.g. In humans when having 45 or 47 chromosomes when 46 is expected in a human cell.

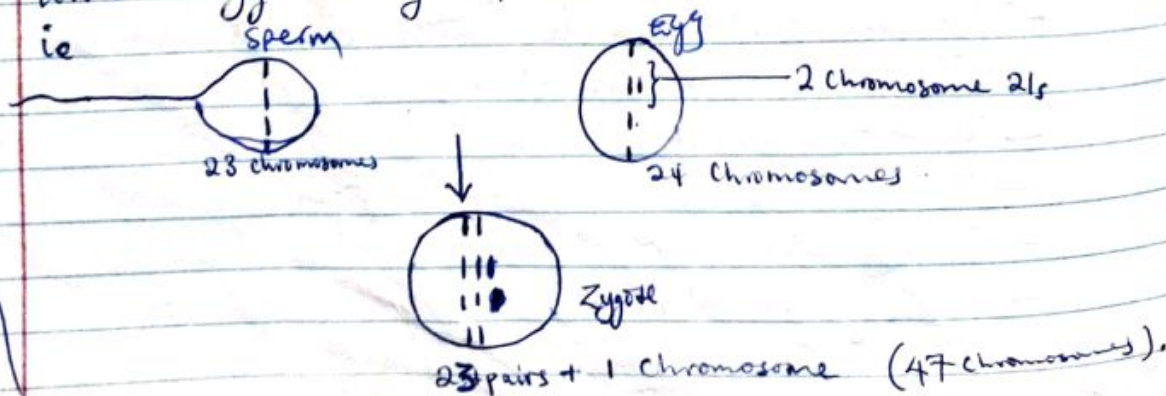
Examples of aneuploidy examples include;

- Down's Syndrome / mongolism
- Turner's syndrome
- Klinefelter's Syndrome

#### Down's syndrome / mongolism

It is a result of an individual having an extra chromosome 21 in each of his/her body cells.

It results when a sperm having 23 chromosomes fuses with an egg having 24 chromosomes i.e. with two chromosomes 21.





NB: Down's syndrome occurs due to non-disjunction in chromosome pair 21 leading to formation of  $\frac{1}{2}$  gametes with 22 chromosomes and another  $\frac{1}{2}$  with 24 chromosomes. ~~as shown above!~~

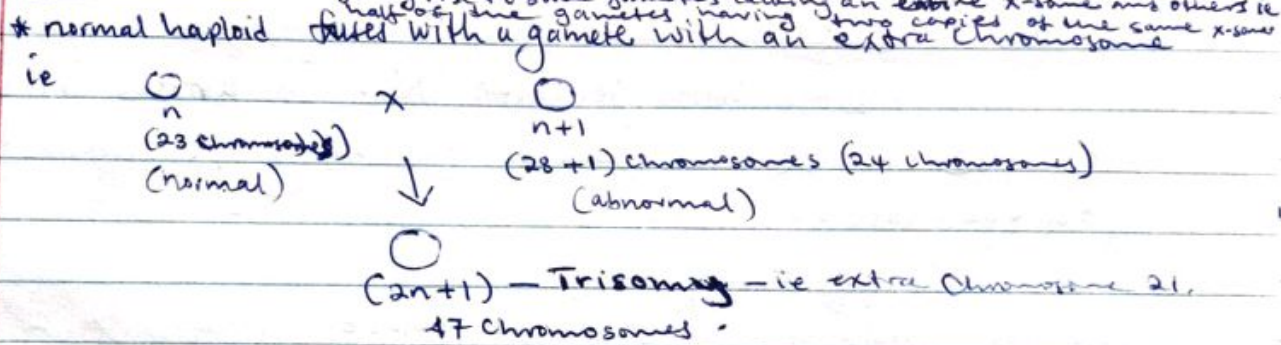
If these gametes fuse with normal gametes from another parent, some offspring will have 45 chromosomes and others ~~47~~ chromosomes.

Individuals with fewer chromosomes e.g. 45 fail to survive. This is because they may lack a number ~~number~~ of vital genes.

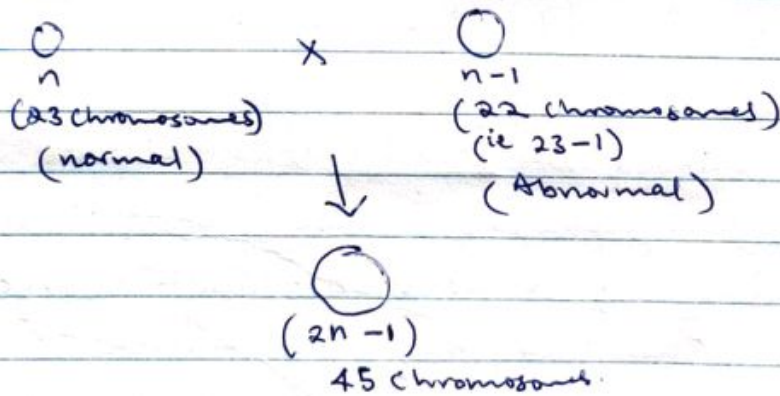
Individuals with extra chromosomes e.g. 47 survive with many abnormalities.

**NB: Non-disjunction** - Is a condition that occurs when a pair of homologous chromosomes fail to separate during Anaphase I of meiosis, giving rise to some gametes lacking an entire X-some and others having two copies of the same X-some.

Illustration



\* Normal haploid fuses with a gamete lacking or having fewer chromosomes.



The formed children are called Mongols (plural) or Mongoloids (plural) (Mongol or a mongoloid - singular) and have the following characteristics.

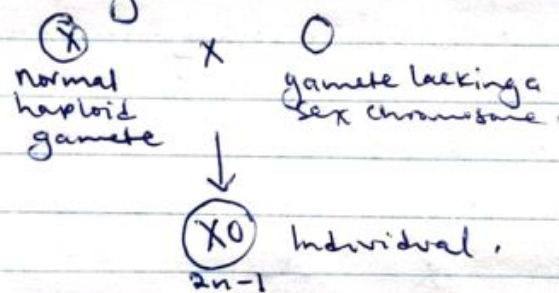
- Mental retardation
- Learning disabilities
- Very thick neck
- Very small head
- Very low disease resistance
- very thick tongue
- Congenital heart diseases
- Stocky body
- Round face with many skin folds
- Very friendly and extra love for music



- Slit-like eyes (mongolism)
- Flat face.
- Coarse, straight hair
- Short Stature and relatively small skull due to poor skeletal development.
- Heart defects occur in about one-quarter of Down's children
- They have increased risk of infection, particularly respiratory and ear infections. Intestinal problems and leukaemia are also slightly more common than normal.
- Squat hands with a characteristic crease which runs all the way.

### Turner's Syndrome

Individuals with genotype XO. It arises as a result of fusion of the sperm containing X chromosome with an egg lacking the X chromosome. i.e.



The individuals are females with the following characteristics.

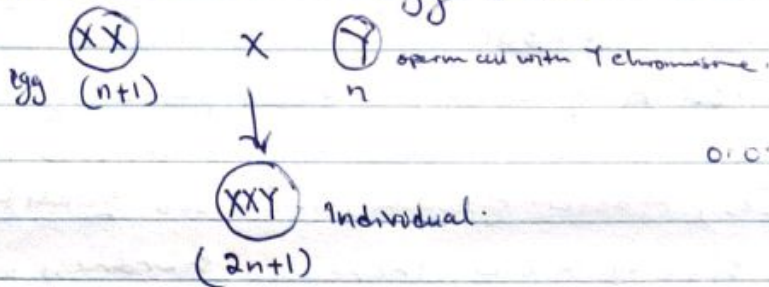
- They lack female secondary sexual characters e.g. no menstrual cycles; Their breasts don't enlarge and have no feature to show that they are female.
- They are infertile i.e. can not produce eggs (sterile)
- Ovaries are absent represented only as connective tissue.
- They are very short averaging 1.5m less than 5 feet.
- They have very close nipples.
- They have pimple like teats found in the center of their small chest.
- Have small uteruses.
- Webbed neck may occur
- Puffy (swollen) fingers with deep set finger nails which are more convex than normal.



- The hair line (line at which hair starts to grow) at the back of the head is lower than normal.

### Kline Felter's Syndrome

It arises as a result of fusion of a cell containing "Y-Chromosome" with an egg of XX. i.e.



Such Individuals are Males with feminine characteristics.

Other characteristics include:

- Having female secondary sexual characteristics.
- Having very tiny testis.
- Having very little facial hair.
- Having a very high or pitched voice higher than normal.
- Having very low IQ (Intelligent quotient).
- Having very big breasts.
- Infertility - Sperm are never produced, although erection and ejaculation are possible.
- Educational difficulties and behavioural problems are fairly common.
- They are usually taller than average.
- Trunk may show signs of obesity.
- Higher than usual FSH (follicle stimulating hormone) secretion for males.

### \*\* POLYPLOIDY

**Definition:** It is a situation where the offspring's chromosome number is a multiple of the haploid chromosome number of a normal being and not just a double of the haploid number. (3 or more times the haploid chromosome number).  
Chromosome  
^  
Mutation

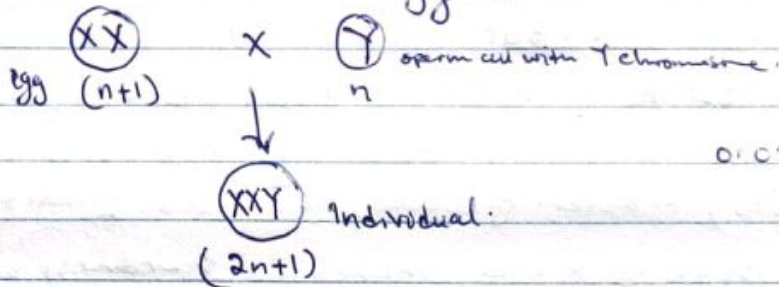
This type of  $n$  results in to increase in the entire haploid set of chromosomes. It leads to formation of triploids ( $3n$ ), tetraploids ( $4n$ ), Pentaploids ( $5n$ ) etc; So it is a condition where a cell contains three or more times the haploid chromosome number.



- The hair line (line at which hair starts to grow) at the back of the head is lower than normal.

### Kline Felter's Syndrome

It arises as a result of fusion of a cell containing "Y-Chromosome" with an egg of XX. i.e.



0.02% in western population.

Such individuals are Males with feminine characteristics.

Other characteristics include:

- Having female secondary sexual characteristics.
- Having very tiny testis.
- Having very little facial hair.
- Having a very high or pitched voice higher than normal.
- Having very low IQ (Intelligent quotient).
- Having very big breasts.
- Infertility - Sperm are never produced, although erection and ejaculation are possible.
- Educational difficulties and behavioural problems are fairly common.
- They are usually taller than average.
- Trunk may show signs of obesity.
- Higher than usual FSH (follicle stimulating hormone) secretion for males.

### \*\* POLYPLOIDY

**Definition:** It is a situation where the offspring's chromosome number is a multiple of the haploid chromosome number

of a normal being and not just a double of the haploid chromosome number. (3 or more times the haploid chromosome number).

This type of <sup>mutation</sup> results in to increase in the entire haploid set of chromosomes. It leads to formation of triploids (3n), tetraploids (4n), Pentaploids (5n) etc; So it is a condition where a cell contains three or more times the haploid chromosome number.



\* It occurs due to non-disjunction involving all  $\times$  chromosome pairs during meiosis leading to formation of gametes with extra sets of chromosomes.

• In plants, polyploidy can be induced by use of Colchicine which prevents spindle fibre formation and the cell division proceeds up to metaphase. Polyploidy in plants leads to hybrid vigour.

• Hybrid vigour in plants - Is a description of a plant with advantageous characteristics e.g. increased resistance to diseases, faster growth, increased quality of yield, increased size of yield, increased hardness of seed to reduce pest attack etc.

Question: State the significance of polyploidy in agriculture (5 marks).

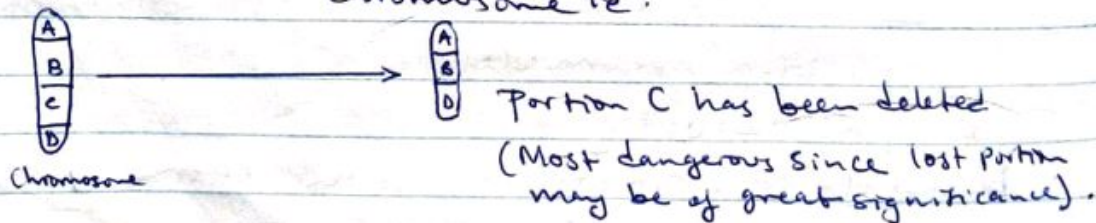
- ie \* Increasing - the size of yield
- Hardness of seeds to reduce pest attack
  - Quality of yield
  - Speed of growth / faster growth
  - resistance to diseases etc.

## • Chromosomal mutations Involving Change in Structure

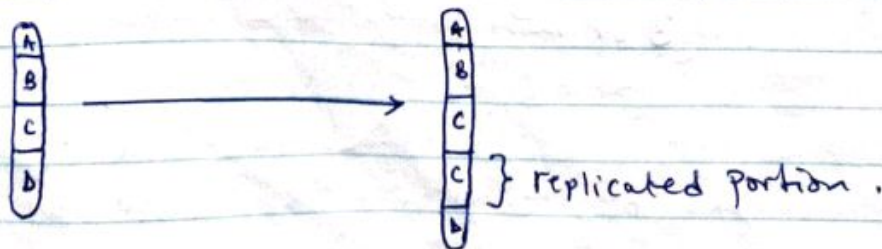
There are four types ie (D<sup>2</sup>IT) ie

- Deletion
- Duplication
- Inversion
- Translocation

(i) Deletion: - It involves the loss of a portion of a chromosome ie.

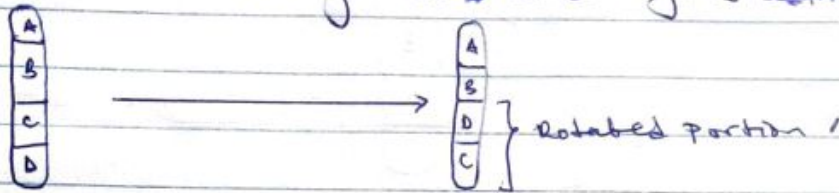


(ii) Duplication: - A chromosome portion replicates.

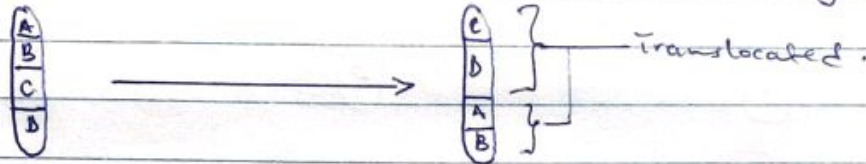




(iii) Inversion: A chromosome portion breaks off, rotates through  $180^\circ$  and rejoins ~~same~~ <sup>the same</sup> region.



(iv) Translocation: Chromosome portion breaks off and joins another area or homologous chromosome.



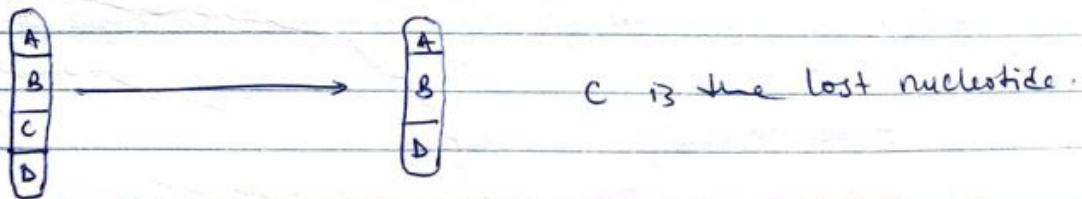
## (ii) Genetic Mutations.

They are also referred to as Point mutations and all involve change in the structure of genes.

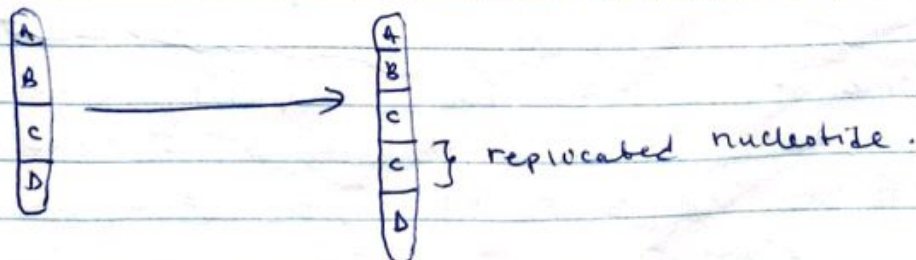
The building units of genes are nucleotides hence changes are in the nucleotides and take the following forms; i.e. D.I.S i.e.

- Deletion.
- Duplication
- Inversion
- Insertion
- Substitution

(i) Deletion: This involves loss of a nucleotide.

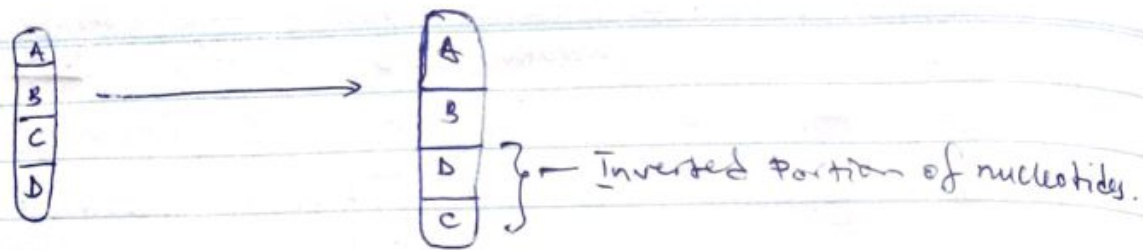


(ii) Duplication: Nucleotides section replicates.

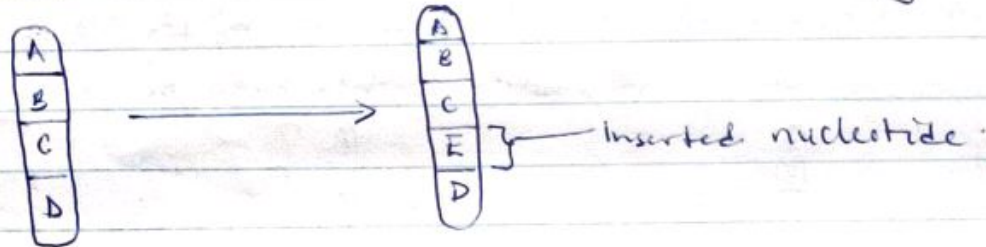


(iii) Inversion: Nucleotide portion breaks off, rotates through  $180^\circ$  and rejoins the same region.

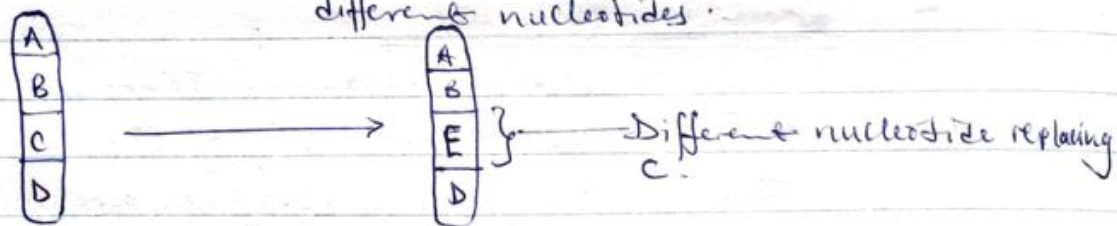




(iv) Insertion: New nucleotides added to gene.



(v) Substitution: Nucleotides removed and replaced with different nucleotides.



### EFFECTS OF MUTATIONS.

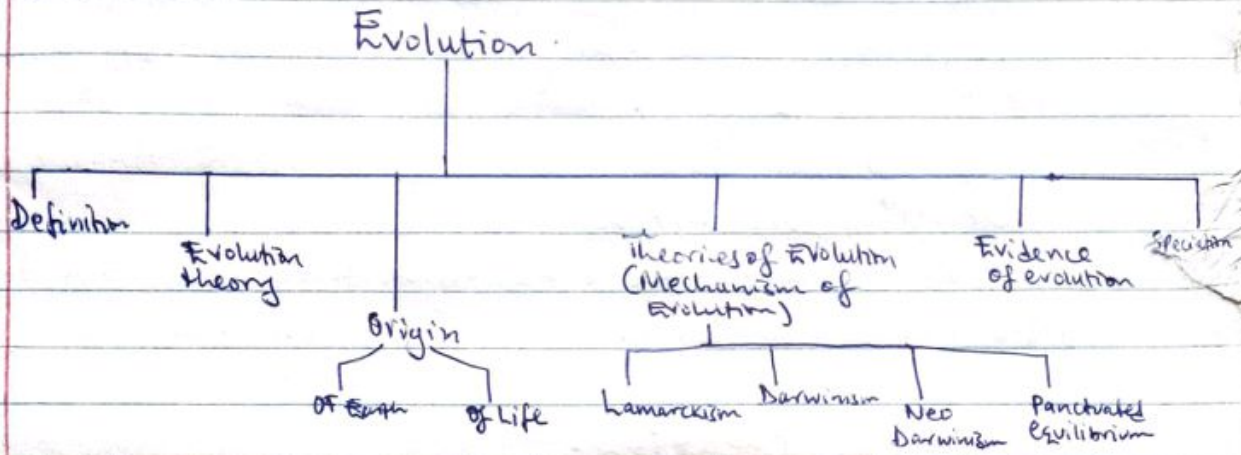
- They cause sterility
- They cause cancer
- In plants, Polyploidy leads to hybrid vigour
- Leads to mental retardation.
- Leads to death.
- leads to anatomical (structural) abnormalities.
- leads to reduced resistance to diseases eg mongolism - etc.

### HOW MUTATIONS LEAD TO EVOLUTION OF SPECIES (NEW SPECIES)

Mutations result into formation of new genotype. These determine a range of phenotypic characters, such as characters are exposed to prevailing environmental conditions. ~~Selective pressure acts on them~~ Some are favoured by the prevailing conditions reproduce and multiply/increase in number while others that are not favoured fail to reproduce, die and decrease in number and become extinct in the long run. Hence this leads to evolution of new species.



# EVOLUTION



## EVOLUTION

**Definition** It is a process by which more adapted complex organisms are gradually formed from the pre-existing organisms by natural selection. eg formation of more resistant mosquitoes to pesticides; more resistant Plasmodium to chloroquine; more resistant bacteria to penicillin.

## EVOLUTIONARY THEORY

It explains how great diversity of living organisms arose.

## ORIGIN OF EARTH

Three (3) theories have been put forward to explain origin of the earth i.e

### (i) Big bang theory

It suggests that a neutron's ball exploded to form the planets which occupied different orbits around the reaction center i.e the sun. The assertion is supported by the fact that the center of all these planets is extremely hot implying that they were once hot balls whose surface got cooled and hardened.

### (ii) Steady state theory

It suggests that the earth has no origin and has always been there to support life. It further proposes that species vary and become extinct but their changes are very minor in case they occur. It was (the theory) developed due to contradictions regarding the age of the earth.



### ii) Special creation.

This suggests that the earth was created by a supreme natural being in its present form and it has not changed.

## ORIGIN OF LIFE

Different theories have also been put forward to explain the origin of life on earth and these include;

### i) Special creation.

This suggests that all living organisms were created by a super natural being (God) in their present form and they have not changed.

This is supported by religious books e.g. Genesis 1:1-26 explains how God made the earth and Hebrews 11:1 and 3 re-emphasises the role of God.

Major critics of the theory suggest that it can not be scientifically proven in the lab.

In 1650 AD, Archbishop Usser of Amagh used Biblical geneologies [Geneology - is the study of families and tracing of their lineages and history] from Adam to Jesus and concluded that God made the earth in 4004 BC and concluded the process with man in the morning at exactly 9:00 am on 23<sup>rd</sup> October.

### iii) Spontaneous generation.

It suggests that living organisms arise from non-living organisms so long as there is an active principle. The theory was proposed by Aristotle. According to him, non-living matter contains an active principle that gives rise to life.

It was supported by Van-Helmont who even designed an experiment to make life. All one needed was a dirty shirt and some grains of wheat and a drop of sweat and rats would be found.



He suggested that insects came from Plant juice, houseflies from faeces and Micro organisms from air and water.

This theory of abiogenesis (ie life came from non living things) was disapproved by Louis Pasteur in 1862 who boiled two nutrient solutions, left one closed and the other uncovered. Maggots developed in only the uncovered one suggesting that living organisms can only be formed by visiting living organisms ie flies; Hence theory of biogenesis (ie life can arise only from pre-existing life).

### (iii) Steady State theory

It suggests that life has always been on earth and living organisms slightly vary.

### (iv) Cosmozoan theory or Theory of Panspermia

It suggests that life has an extra terrestrial origin and after being made was just brought on earth. ie life arose from different parts of the universe and it was introduced to the earth probably by meteorites.

NB A meteorite - is a solid piece of debris from a source such as an asteroid or a comet, that originates in outer space and survives its impact with the earth's surface.

An asteroid - is a small rocky body orbiting the sun. (minor planets especially those of the inner solar system).

A comet - is an icy small solar system body that when passing close to the sun, heats up and begins to outgas, displaying a visible atmosphere.

A meteorite's size can range from small to extremely large. They have many organic molecules such as cyanogen and hydrocyanic acid which may have acted as "seeds" falling on a barren earth.

### (v) Biochemical Evolution

It suggests that due to radiation from the sun, lightening and other energy sources, complex



Chemical reactions occurred in the atmosphere forming specialised droplets of chemicals which upon landing in oceans started growing, dividing and replicating to form the first cell. This was formed by reactions that occurred on the primitive earth. Gases and other elements reacted due to change in temperature and dissolved in water to form "Organic soup". In the organic soup, biochemical compounds such as proteins, lipids, nucleic acids, carbohydrates were formed. This led to the formation of the first cell that was prokaryotic in nature. Multicellular organisms were formed by Endosymbiosis.

## MECHANISMS OF EVOLUTION (THEORIES)

**NB** They explain how the complex more adapted organisms were formed from the preexisting organisms.

These include:

- (i) Lamarckism
- (ii) Darwinism
- (iii) Neodarwinism
- (iv) Punctuated equilibrium.

### (i) LAMARCKISM

It was proposed by Lamarck and he suggested that more complex organisms are formed as a result of cumulative inheritance of acquired characters.

He suggested that use of a given body part makes it <sup>to</sup> properly grow and disuse of a given body part makes it to get reduced hence Law of Use and Disuse.

The law of use and disuse states that use of a structure would lead to increased size and/or efficiency while disuse of a structure would lead to degeneracy and atrophy (ie waste away, become vestigial or decline in efficiency).



eg weight lifting makes one develop muscles.  
He explained that the long necked giraffes were formed from short necked giraffes as a result of cumulative inheritance of the acquired increase in length of the neck of giraffes as they stretched their necks to feed on leaves since the grass had been overgrazed.

He was disapproved by August Weismann who cut off tails of rats, copulated the rats and over generations they continued producing tailed rats. This was evidence that acquired characters are never inherited.

#### Highlights in Lamarckism

- Change in env't
- Use or disuse of structure
- Acquisition of x-ters
- Inheritance of x-ters.

### (iii) DARWINISM

It was proposed by Charles Darwin who was an English Naturalist.

He proposed that evolution occurs by natural selection. In any given population, there are variants (Individuals) expressing differences. In case of environmental change, some individuals with adaptive characteristics are selected for (favoured by <sup>the</sup> changes) while other variants are selected against (not favoured). Over generations of natural selection, the selected for organisms become predominant and even accumulate the adaptive characteristics while the selected against organisms gradually become eliminated.

#### Evolution of the Giraffe according to Darwin.

Long ago, there were two varieties of giraffes, i.e. long necked giraffes and short necked giraffes.

The change in env't where food became scarce resulted into survival of the long necked giraffes b/c they could reach the leaves on <sup>the</sup> tall trees. This made them to reproduce more and the short necked giraffes to reproduce less because of the survival disadvantage. Over generations of drought, the long necked giraffes were selected for and even



Concentrated the characters of being longnecked while the short necked giraffes were selected against and even got extinct hence a generation of only long necked giraffes.

### Conclusions from Darwinism.

- There must be a struggle for existence.
- Organisms possess variations.
- Some variations better suit organisms to prevailing environmental conditions and survive and pass it on to the next generation.

dichlorodiphenyltrichloroethane. ( $C_{14}H_9Cl_5$ )

### (iii) NEO DARWINISM

Neo means New

It proposes that evolution occurs by Natural Selection of genetically determined characters.

This implies that it is like Darwinism except that it emphasizes that the character selected for is genetic. It is the scientifically agreed upon mechanism of evolution.

Fig. 2

Evolution of Resistant mosquitoes to DDT.

There were two varieties of mosquitoes which could have <sup>arise</sup> arisen as a result of a mutation. Use of DDT killed non resistant mosquitoes and <sup>did</sup> not affect the resistant strains. As a result, the resistant strains had a reproductive advantage and since this character was genetically determined, it concentrated in the offspring making them even more resistant.

The non resistant strains were selected against and over generations their population was reduced till they became extinct leaving only a generation of resistant mosquitoes to the DDT.

### (iv) PUNCTUATED EQUILIBRIUM

It proposes that evolution occurred by rapid new creation of living organisms by God.



# EVIDENCE OF EVOLUTION (Proof of Evolution)

1. Development of resistant forms of pests to pesticides. It shows selection for most adapted varieties.
2. Resistance increase of Plasmodium to antimalarials. It shows Nature selecting for resistant forms.
3. Emergence of resistant forms of bacteria to antibiotics shows selection for the most adapted bacteria.
4. Increased resistance of weeds to herbicides.

5. Industrial melanism - This is a classical example of natural selection; where the black moth were selected for while the white <sup>peppered</sup> moth were selected against ~~during~~ during the industrial revolution.

6. Evidence from Palaeontology. Palaeontology is the study of fossils.

When fossils in the recent earth stratum are compared with fossils in the old earth stratum, it is evident that fossils in the new earth stratum are more complex in structure than those in the old earth stratum. This suggests that organisms have become more complex with time.

## 7. Comparative anatomy

Is the study of different structure of different organisms with an aim of relating them.

It reveals three (3) forms of structures i.e.

- Homologous structures
- Analogous structures
- Vestigial structures; which all show proof of evolution.

### • Homologous structures -

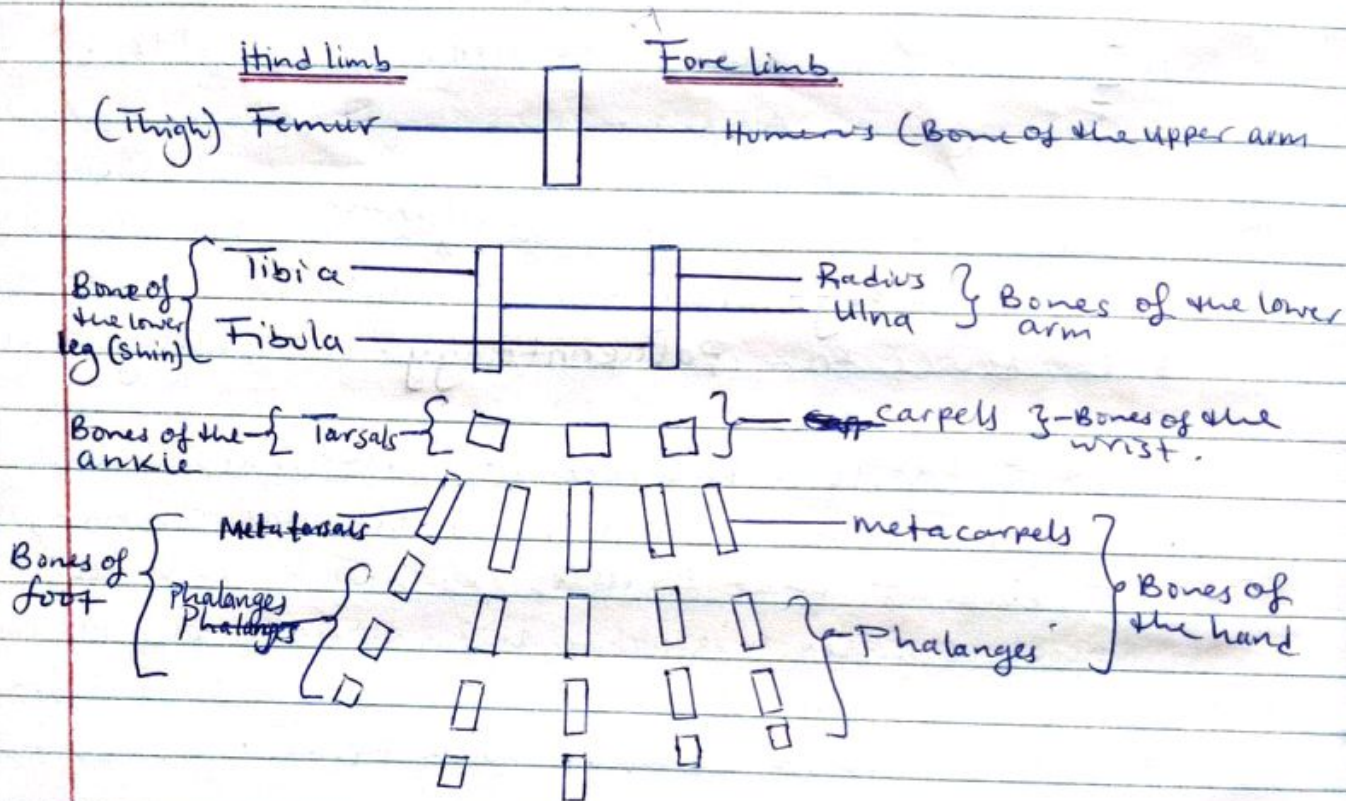
Structures with the same basic plan which have undergone modifications in order to adapt to doing particular function e.g. the arm of humans, the wings of a bird, flipper of a whale, ~~arm~~ arm of chimp etc have the same basic form of a pentadactyle limb but have undergone modifications for manipulation of materials, flight, swimming, increased grip respectively.

The similarities suggest a common ancestry and



The differences amongst the limbs are attributed to evolution. Homologous structures indicate divergent evolution.

The pentadactyl limb - in all vertebrates except fish. A pentadactyl limb is a limb plan of five digits found in all vertebrates.



Same function  $\rightarrow$  Different modifications for different functions  
hence ~~if~~ divergent evolution.

### • Analogous Structures

These are structures that have undergone evolution to perform similar functions when their basic structure is different. E.g. Wings of birds and Insects. It indicates Convergent evolution.

Same function  $\leftarrow$  Different basic structure.

### • Vestigial Structures.

These are structures that have reduced in size and have lost their function in present day organisms but functional in ancestors. E.g. Human tail is reduced to coccyx, Limb girdles in snakes.



- Human appendix although not functional ~~in~~ in humans is homologous with functional appendix of herbivorous mammals.

These structures show common ancestry to the organisms which are having them and they ~~have~~ <sup>are</sup> been modified because of evolution.

### 8. Comparative Embryology

Is the study of developmental stages undergone by embryos of different organisms.

It reveals a number of similarities among vertebrate embryos e.g.

- Possession of a tail,
- a single circulatory system which include a two chambered heart with no separation into right and left halves.
- They all live in a fluid medium
- External branchial groove (visceral clefts) in the pharyngeal region and a series of internal paired gill pouches.

All these similarities suggest common ancestry and attributes differences amongst adults ~~to~~ to evolution.

### 9. Comparative Biochemistry.

When chemicals in different living organisms are compared, a number of similarities are revealed e.g. in their DNA, hormones, enzymes etc which suggests common ancestry, and attributes differences amongst organisms to evolution.

### 10. Comparative Physiology.

When processes occurring in bodies of living organisms are compared, they are significantly similar e.g. digestion, respiration, gaseous exchange etc.

Similarities suggest common ancestry and differences amongst organisms are attributed to evolution.



## Classification:

In nature, organisms are classified depending on what they share in common to come up with Kingdom, phylum, <sup>organism</sup> class, order etc. However, organisms in each grouping exhibit a number of differences which could be attributed to evolution.

## 12 Geographical distribution:

The geographical isolation of the earth into continents enabled organisms to evolve so as to exploit resources in the localities. Eg. llamas of South Africa resemble the African Camels in a number of ways and also exhibit a lot of differences as a result of evolution and that isolation also leads to evolution.

## SPECIATION

Defn: This is the process by which one or more species arise from previously existing species.

A single species may give rise to new species and this is called Intraspecific Speciation.

Or as is common in many flowering plants, two different species may give rise to a new species and this is called Interspecific Speciation.

If Intraspecific speciation occurs whilst the populations are separated, it is termed as allopatric speciation.

If the process occurs whilst the populations are occupying the same geographical area, it is called Sympatric speciation.