

GENETICS & HEREDITY

Genetics: This is the study of the mechanism by which characteristics (traits) are transmitted from parents to the offsprings. This transmission occurs via gametes during fertilization in sexually reproducing organisms. Therefore genetics can also be referred to as the study of inheritance characteristics of the parents by the offsprings. The characteristics of organisms are controlled by internal factors called **genes** located on chromosomes.

A GENE is a section of DNA that determines a particular characteristic in an organization **or** a section of DNA that controls the production of a polypeptide chain in an organism.

HEREDITY is the transmission of characteristics from parents to offsprings (**progeny**). Such characters are also called **traits**. In humans, characters like hair texture, height; skin colour, body build and hair colour are inheritable characters.

THE IMPORTANCE OF GENETICS

- It is used in genetic engineering where better breeds and varieties of plants and animals are produced. This is intended to increase production and improve resistance of diseases and pests. This can be done locally through cross breeding.
- It is used in the legal profession to determine the paternity of the child i.e. genetics is used to settle paternal disputes by confirming who the father of the child is. This can be proved through use of blood groups as these groups are genetically inherited and can therefore be used to prove the rightful father of the child. If the blood groups fail to prove then DNA analysis can be used.
- They are used in blood transfusion. Genetic principals are used during blood transfusion so that blood being transfused is compatible to avoid blood clotting (Agglutination) in the recipient.
- It is used in the control of the transmission of genetic diseases. These diseases are genetically engineered e.g. hemophilia, colorblindness, acandropiasia etc. can be eliminated from the human population by following the principles of genetics as these diseases are genetically inherited.
- It can be used in crime investigation i.e. use of the DNA finger prints to identify criminals
- It is used in molecular biology to manufacture artificial enzymes, hormones and vaccines.
- It enables humans to choose the right partners during marriage by choosing those with characteristics for reproduction.

TERMINOLOGIES INVOLVED IN GENETICS

Alleles. These are alternative forms in which the gene can exist but control contrasting features of characteristics. Alleles exist in pairs e.g. consider a gene for height. This gene can be expressed inform of allele as T (for tallness) and t (for shortness). Therefore, these two alleles can exist as TT and tt.

Locus (plural loci). This is the position on the chromosome where the genes are located.

Homozygous. This is a condition where an individual possess identical alleles for a particular gene e.g. TT, tt, AA.

Heterozygous. This is a condition where an individual possess non-identical alleles for a particular gene e.g. Tt, Bb

Pure breeding (breeding true). This is where the individuals crossed are homozygous and therefore produce consistently the same characteristic, generation after generation. A pure breed should therefore be a homozygous individual when considered for a particular characteristic.

Hybrid. This is heterozygous individual obtained from crossing two parents with contrasting characteristics but when these parents are pure breeding e.g. **tt X TT**

Dominant allele. A dominant allele is the one that can express its self phenotypically in both homozygous and heterozygous forms.

Recessive allele. This is an allele that can only express itself phenotypically in the homozygous form as it is suppressed by the dominant allele in the heterozygous form.

Note: Recessive alleles are presented by small letters (lower case) while dominant alleles are represented by capital letters (upper case)

Phenotype. This is the physical or outward appearance of an organism.

Heterozygous individuals are genetically called carriers of the recessive characteristic. Recessive characteristics can only be expressed when two carriers make an organism which is phenotypically recessive appears e.g. the sickle cell anemia individuals, albinos, hemophiliac e.t.c.

Crossing (X). This refers to the mating of the male and female organisms under a consideration.

First filial generation (F₁). This refers to the set of offsprings obtained from crossing two pure breeding parents with contrasting characteristics. These individuals are therefore heterozygous or hybrids.

Second filial generation (F₂). This refers to the set of offsprings that are obtained from crossing mature hybrid of parents of the first filial generation.

Selfing. This refers to the crossing of offsprings of the same parents.

Test cross. This is the mating of a phenotypically dominant individual with a recessive individual so as to determine the genotype of the phenotypically dominant individual. This is due to the fact that a phenotypically dominant individual can either be heterozygous or homozygous. If the homozygous offsprings resemble the dominant parent then the dominant parent is said to be homozygous and if the offsprings formed from the test cross shows a phenotypic ratio of 1:1, then the parent with an unknown genotype is heterozygous.

Back cross. This is the mating of an offspring with one of its parent so as to prove the genotype of the parents.

Reciprocal cross. This is a cross in which the phenotypes of the same characteristics are interchanged among the parents during a genetic experiment.

MENDEL'S GENETIC EXPERIMENTS AND MONOHYBRID INHERITANCE

This is the inheritance of a single pair of characteristics from the parent to offsprings.

Examples include, height, blood groups, albinism, sickle cell anemia, and sex linked characteristics e.t.c.

This mechanism of inheritance was discovered by a scientist called **Gregory Mendel** who carried out a number of genetic experiments using the garden pea plants. He also observed many sexually reproducing organisms and found out that they had variations among themselves despite being of the same species.

In these experiments, Mendel carried out cross pollination between tall pea plants and short pea plants he had grown in his garden. In order to carry out a proper cross, Mendel covered the stigma of all flowers of one group of pea plants in order to have male pea plants.

He also removed all the anthers from the flowers of another group of pea plants in order to have female pea plants. Using a brush he transferred pollen to tall pea plants from short pea plants. He observed the F₁ offsprings were all tall. He then selfed the F₁ pea plants to get F₂ which was found to be a mixture of tall pea plants and short pea plants.

In order to perform good genetic experiments, Mendel used a garden pea plant because such plants have good characteristics for genetic experimentation which included the following;

- They have many contrasting characteristics without any intermediates such as tall and short stems, smooth and wrinkled seeds, yellow and white flowers i.e. a good genetic organism must show many discontinuous variation characteristics
- They produce large numbers of offsprings which provide a large sample for experimentation so as to get reliable results.
- It is possible for them to undergo controlled pollination.
- They are so small that they can be conveniently handled.
- They have a short life span and they can be reproduced very quickly before the end of the life span.

Mendel was successful in his genetic experiment because;

- He had a systematic approach to his work. This is because he dealt with a single characteristic (monohybrid inheritance) and then later on he considered two characteristics simultaneously (dihybrid inheritance).
- He was very patient during his experiments so that he was able to reproduce the garden peas for several generations.
- He used a very good experimental organism, the garden peas.

Conclusions from Mendel's experiments

About the actual mechanism of inheritance

1. The phenotypic characteristics are under the control of internal factors (these factors were later named genes).
2. It is these factors that are transmitted from the parents to the offsprings i.e. (from one generation to the next).
3. For each character, an organism inherits from the parents two alleles (internal factors), one from each parent and it is these factors which account for variations in inherited characters.
4. The factor which phenotypically appears in F1 generation is dominant to the one which fails to phenotypically in F1 but instead appears in the F2 generation.
5. During sexual reproduction the egg cell and the sperm make equal contribution to each of the characteristics of the offspring such that the offspring has both male and female parental characteristics. This is because the two alleles for a heritable character separate or segregate during formation of gametes in meiosis and end up in different gametes.
6. Always in F2 generation, the dominant and recessive offsprings appear in a phenotypic ratio of 3:1. The results, using proportions only, are summarised in the table below;

Character	Type of cross	F1 generation	F2 generation	Ratio
Stem length	Tall X Short	All tall	787tall, 277short	2.84:1
Seed colour	Green X Yellow	All yellow	6022yellow, 2001green	3.01:1
Seed shape	Round X Wrinkled	All round	5474 round, 1850wrinkled	2.96:1
Seed coat	Coloured X White	All coloured	705 coloured, 224 white	3.15:1
Pod colour	Green X Yellow	All green	428 green, 152 yellow	2.82:1
Pod shape	Inflated X	All inflated	882 inflated,	3:1

	Constricted		299 constricted	
Flower position	Terminal X Axial	All axial	651 axial, 207 terminal	3.14:1
Flower colour	Purple X white	All purple	705 purple, 224 white	3:1

MENDEL'S FIRST LAW OF INHERITANCE

From this experiment about monohybrid inheritance he suggests the law of genetics which is known as the **law of segregation**.

This law states **that in diploid organisms each characteristic is controlled by a pair of alleles but during gamete formation the alleles separate so that each gamete possesses a single allele.**

Explanation of Mendel's first law of inheritance

This law is explained by meiosis which halves the chromosome number in that each characteristic of an organism is determined by a pair of alleles located on the pair of homologous chromosomes in the nucleus of the cell of an organism. Each allele of the pair for a characteristic is therefore carried by a single chromosome of the homologous pair when homologous chromosomes segregate and move towards opposite pole of the cell during anaphase I of meiosis. This results into each gamete carrying one allele of the gene pair due to the separation of a pair of chromatids during anaphase I.

Diagram

COMPLETE DOMINANCE; Is a condition where one allele suppresses the effect of another allele in heterozygous condition

E.g. Inheritance of Height, appearance of seed Testa, colour of unripe pod and colour of cotyledon in Garden pea plant.

WORKED EXAMPLES

1. In a garden pea plant there are two forms of heights i.e. tall and short. When a pure breeding tall pea plant was crossed with a short pea plant all the offsprings obtained were tall when the offsprings were selfed a phenotypic ratio was obtained in F₂.

- Using suitable genetic symbols, work out the genotypes and phenotypes of the F₂ generation
- What are the phenotypic and genotypic ratios of the F₂ generation
- Explain how you would determine the genotype of F₁ tall pea plants formed
- Suppose 300 pea plants were produced in the F₂ generation
 - How many were tall?
 - How many were short?

Solution

(a)

Let **T** represent the allele for tallness

Let **t** represent the allele for shortness

Parental phenotype (2n): tall pea plant X short pea plant

Parental genotype;

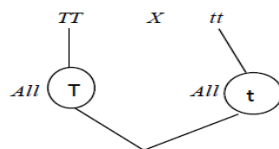
Meiosis

Gametes (n):

Fertilization:

F₁ genotypes (2n):

F₁ genotypes (2n) are all tall pea plants.



Selfed:

Parental phenotypes (2n): tall pea plant X tall pea plant

Parental genotype:

Meiosis

Gametes (n)

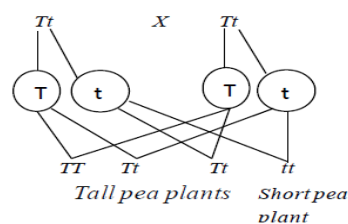
Fertilization

F₂ genotype (2n)

F₂ phenotype

F₂ genotypic ratio 1TT:2Tt:1tt

F₂ phenotypic ratio: 3tall:1short

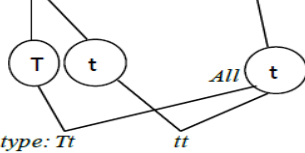


- (c) In order to determine the genotype of F₁ pea plants, carry out a test cross, by crossing the F₁ tall pea plants with short pea plants.

Parental phenotype: tall pea plant X short pea plant
 Parental genotype (2n): Tt X tt

Meiosis

Gametes (n)
 Fertilization



Test cross offspring genotype: Tt

Test cross phenotypic ratio 1 tall:1 short

This shows that F₁ tall pea plants were heterozygous

- (d)
- $$= \frac{\text{homozygous tall}}{\text{total}} \times 700 = \frac{1}{4} \times 700 = 175 \text{ plants}$$

Homozygous tall
 - $$\text{short} = \frac{\text{homozygous short}}{\text{total}} \times 700 = \frac{1}{4} \times 700 = 175 \text{ plants}$$

Homozygous
 - $$\frac{\text{heterozygous}}{\text{total}} \times 700 = \frac{1}{2} \times 700 = 350 \text{ plants}$$

Heterozygous =

Question 2

Suppose a man who is a tongue roller marries a woman who is a non-tongue roller and all the children obtained in F₁ are tongue rollers.

- Work out the phenotypic and genotypic ratio as obtained in F₂ generation.
- What is the probability that the 4th born is a non-tongue roller?

Question 3

A man with normal skin marries a carrier for albino skin. What is the probability that some of their children will be albinos?

Note: albinism is a monohybrid condition due to lack of melanin pigment in the skin. It arises due to a mutation which alters the gene responsible for the synthesis of melanin. This makes an albino to have white hair, very light coloured skin and pink eyes.

FACTORS WHICH MODIFY OR AFFECT MENDEL'S MONOHYBRID 3:1 AND 1:2:1 RATIOS (GENE INTERACTIONS EXAMPLES)

1. LETHAL GENES:

These are genes that lead to the death of the bearer lethal genes are divided into 3 major categories;

- Gametic lethal genes.** These are genes which kill the gametes and therefore prevent fertilization.
- Zygotic lethal genes.** These are genes which kill the zygotes and embryos before birth e.g. the gene that determine coat color in mice.
- Infantic lethal genes.** These are genes which kill individuals between birth and reproductive stages e.g. the gene that determines chlorophyll formation in maize, sickle cell anemia in man e.t.c.

Lethal alleles: This is a combination of alleles which result into death of offspring before birth

Lethal genes in mice

The gene that determines coat color in mice is a zygotic lethal gene. In mice, there are two colours determined by these genes i.e. yellow and grey (agouti). If two yellow mice are crossed they produce both yellow and grey offspring however these offspring appear in a phenotypic ratio of two yellow; 1 grey instead of **3:1**.

This is because the homozygous dominant yellow mice die in the uterus which reduces the phenotypic ratio.

The yellow mice produced are always heterozygous and this changes the monohybrid genotypic ratio from

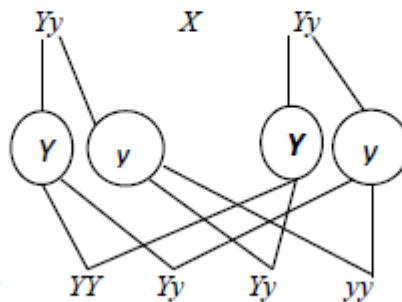
1:2:1 to 2:1

Let Y represent the allele for yellow coat color

Let y represent the allele for grey coat color.

Parental phenotype (2n): yellow mouse x yellow mouse

Parental genotype (2n):



YY dies before birth and therefore the offspring phenotype will be 2 yellow: 1 grey

Some genes are lethal when both alleles are present. Lethality can occur before or after birth

- Huntington's disease in humans is caused by a lethal allele, death occurs later in life
- Other examples: Mouse **coat color** (yellow), **Crepper legs** in chickens, **Manx Cats** (no tails)

When two Manx cats are mated, the ratio of the offspring is 2 manx cats to 1 long tailed cat

An example is the "creeper" allele in chickens, which causes the legs to be short and stunted.

- Creeper is a dominant gene, heterozygous chickens display the creeper phenotype
- If two creeper chickens are crossed, one would expect to have (from Mendelian genetics) 3/4 of the offspring to be creeper and 1/4 to be normal
- Instead the ratio obtained is 2/3 creeper and 1/3 normal.
- This occurs because homozygous creeper chickens die.



	P	p
P	PP lethal	Pp creeper
p	Pp creeper	pp normal

Manx Cats

Cats possess a gene for producing a tail. The tailless Manx phenotype in cats is produced by an allele that is lethal in its homozygous state. The allele interferes with normal spinal development, in heterozygous cats this results in lack of a tail.

Show the cross between a Manx cat and a normal cat.

There is a phenotype in chickens called the creeper condition. This phenotype is caused by an autosomal mutation and displays as a chicken that has short and stunted legs. Chicken embryos that are homozygous for the creeper condition have severe developmental problems and never hatch from their eggs. What is the inheritance pattern? What are the different phenotypes and the associated phenotypic and genotypic ratios from a cross of two chickens with the creeper condition? Show your work with a Punnett square.

Note:

- Dominant lethal genes are very rare in a population because they are usually manifested easily in growth and development of the offspring at an early age and hence easily eliminated.
- A **pleiotropic gene** is the one which controls more than one aspect or characteristic in the metabolism of an organism e.g. the **Y** gene in mice is controlling both viability and coat colour, for viability the **Y** gene acts as a recessive gene since homozygous **YY** mice dies in the uterus and since **Yy** mice are yellow this phenomenon is called **pleiotropy**.

2. CO-DOMINANCE

CO-DOMINANT: This is a condition whereby when two genes co-exist in heterozygous state both equally express themselves to produce a phenotype which is intermediate of the two parents.

This is a phenomenon whereby the alleles controlling a particular characteristic have equal powers of expressing themselves in the phenotype in the heterozygote. Therefore the offsprings produced will have a mixture of the two parental characteristics in the phenotype.

Co-dominance is taken to be a form of incomplete dominance since no allele suppresses the phenotypic expression of another. In co-dominance which use capital letter to represent all the two alleles each letter corresponding to each of the two characteristics.

Examples of co-dominance include the following;

- The gene that determines coat color in cattle
- Inheritance of blood group AB in man
- Inheritance of sickle cell trait

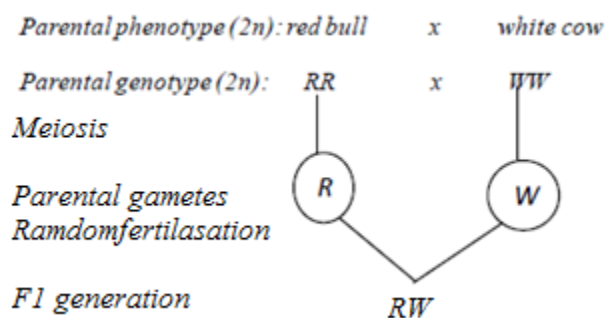
Inheritance of coat colour in cattle.

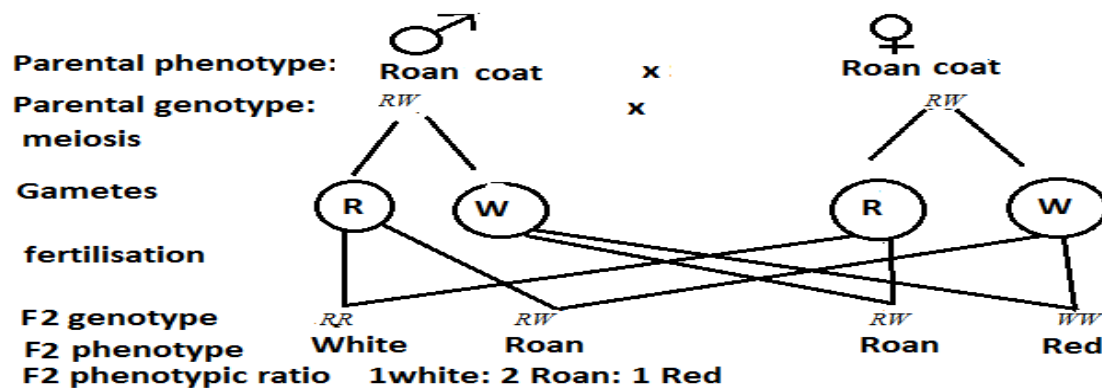
Consider a cross between a red bull and a white cow whose **F₁** offsprings are selfed. Work out the genotypes and phenotypes in **F₁** and **F₂** generation stating in each case the ratios

When the **F₁** roan offspring are selfed, the **F₂** offspring will be red, roan and white in the ratio 1 red: 2 roan: 1 white as shown below

Let *R* represent the allele for red coat color

Let *W* represent the allele for white coat color.





Inheritance of sickle cell anaemia

This is an abnormal condition in which the red blood cells collapse into a sickle shape under low oxygen concentration due to the presence of abnormal haemoglobin (Hbs) in the red blood cells. The normal haemoglobin is found in red blood cells with a bi concave disc shape

Genetic crossing illustration

Sickle cell anaemia is caused by a substitution mutation, in which an amino acid known as **glutamic acid**, responsible for the formation of normal haemoglobin is replaced by another amino acid called **valine** which leads to the formation of **HbS** in the red blood cells.

Haemoglobin is made of four polypeptide chains, two α -chains which are 141 amino acids long and two β -chains which 146 amino acids long. The substitution mutation occurs at the sixth amino acid in the β -chain, this results in wrong amino acid, valine, being incorporated into two of the β -polypeptide chains. **Valine** is non-polar and hydrophobic which makes its presence in the haemoglobin (**HbS**) less soluble when deoxygenated. Therefore when HbS loses its oxygen, the molecules come out of solution and crystallise (solidify) into rigid rod-like fibres. These change the shape of the red blood into a sickle shape.

Effects of sickling red blood cells

a. Anaemia this is occurs because the sickle cells are destroyed which lowers the amount of oxygen to be carried leading to acute anemia. This leads to;

- Fatigue (weakness)
- Poor physical development
- Dilation of the heart which may lead to heart failure

- Infections which lead to frequent illness
- b. Interference with circulation of blood because the cells get jammed in capillaries and small arteries. This leads to;
- Heart damage which leads to heart failure
 - Lung damage which leads to pneumonia
 - Muscle and joint damage which leads to rheumatism and pain
 - Gut damage which leads to abdominal pain
 - Kidney damage which leads to kidney failure
 - Liver damage

c. Enlargement of the spleen because the sickle cells collect in the spleen for destruction

The effects above make the homozygous sufferers to often die before reproductive age.

Note: The allele that erythrocytes to have a distorted form in sickle-cell anaemia also causes these blood cells to rupture easily, thereby inducing anaemia. This gene can also be described as pleiotropic since it has more than one effect in an organism.

In heterozygous individuals, half the molecules made are HbS and HbA i.e. the alleles HbA and HbS are co-dominant and the faulty gene is not recessive. Heterozygous people are not affected except at unusually low oxygen concentrations, such as when flying in an unpressurised aircraft or climbing at high altitude. Then some of the cells sickle. The **heterozygous** condition is known as **sickle cell trait**.

These individuals have a **selective advantage** over non carriers because they are far less susceptible to malaria (the malaria parasite multiplies inside normal red blood cells) so are more likely to survive in malaria infested areas, and pass on their genes to the next generation. A single copy of the sickle-cell allele increases resistance to malaria. The final frequency of the gene in the population varies according to the amount of malaria.

Consider a normal man marrying a sickle cell anaemia woman. Consider another man who is a carrier of sickle cell anaemia of the same disease. Work out the phenotypic and genotypic ratios arising from these two marriages.

Example

Consider a normal man mating with a woman with sickle cell anemia to obtain F1 offsprings which will be phenotypically normal but carriers, if the two carriers mate to form F2 the phenotypic ratio will **be 1:2:1**

Solution

Let Hb^A represent the allele for normal red blood cells

Let Hb^S represent the allele for sickle celled red blood cells.

Parental phenotypes: normal man x sickler woman

Parental genotypes (2n): $Hb^A Hb^A$ x $Hb^S Hb^S$

Meiosis

Gametes (n): all Hb^A all Hb^S

Fertilization:

F₁ genotype (2n): $Hb^A Hb^S$

F₁ phenotype: All are carriers

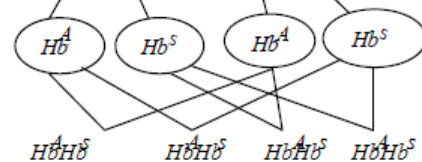
Parental phenotype: sickler man x sickler woman

Parental genotype (2n): $Hb^A Hb^S$ x $Hb^A Hb^S$

Meiosis

Gametes (n)

Fertilization



F₂ genotypic

ratio: **1 $Hb^A Hb^S$: 2 $Hb^A Hb^S$: 1 $Hb^S Hb^S$**

F₂ phenotypic ratio: 1 normal: 2 carrier for the sickle cell trait: 1 sickle cell anaemia

Carriers (heterozygotes) of sickle cell anemia show the sickle cell trait, a co-dominant condition, in which most of the red blood cells have normal hemoglobin and only about 40% of the red blood cells have abnormal haemoglobin **S**. This produces mild anemia and prevents carriers of the sickle cell trait from contracting malaria. This is because when the plasmodium that causes malaria enters a red blood cell with haemoglobin **S**, it causes extremely low oxygen tension in the cell which leads to the cell sickling in heterozygotes. These sickled cells are quickly filtered out of the blood stream by the spleen, thus eliminating the parasites.

3. INCOMPLETE DOMINANCE; Is a type of inheritance where two alleles forming a gene pair, neither of them dominate the other, but contribute equally to determine a particular character.

This is a condition whereby the characteristics of the alleles blend together to form an F1 Offspring phenotype which is intermediate between the two parental phenotypes. Therefore the F1 individuals do not resemble any of the parents.

It can also be defined as a situation where the heterozygote shows a phenotype intermediate between the parental phenotypes.

In incomplete dominance no gene dominates the other in the phenotype but instead forms intermediate phenotypes and are therefore represented using capital letters.

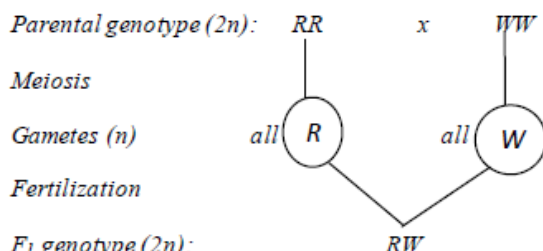
Example

In a snap dragon plant, when a red flowered is crossed with a white flowered plant, all the F1 plants obtained are pink flowered. When the F1 are selfed, the F2 phenotypic ratio is 1:2:1 instead of 3:1. Using suitable genetic diagrams, explain the above results.

Let R represent the allele for red flower colour

Let W represent the allele for white flower colour.

Parental phenotype (2n): red flowered x white flowered



F1 genotypic ratio: all RW

F1 phenotypic ratio: all pink flowered plants

When selfed

F1 x F1

Parental phenotype: pink flowered plant x pink flowered plant

Parental genotype (2n): RW x RW

Meiosis

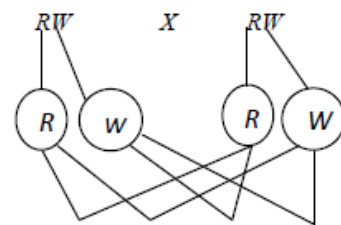
Gametes (n)

Fertilization

Offspring genotype (2n): RR RW WR WW

F2 genotypic ratio: 1RR: 2RW: 1WW

F2 phenotypic ratio: 1Red: 2pink: 1White



DIHYBRID INHERITANCE

Is the inheritance whereby two characteristics, each controlled by a different gene at a different locus.

When Mendel considered the inheritance of two characteristics simultaneously, he concluded that these characteristics are inherited independently and each pair of alleles separates during meiosis and during fertilization each of the alleles combines randomly with either alleles of another pair. From this conclusion Mendel made his **second law of inheritance** which states that; “each characteristic in diploid organisms is controlled by a pair of alleles which separate so that each allele randomly combines with any other allele of another pair.”

From this law Mendel also described it as the **law of independent assortment**.

This law is explained by meiosis as follows.

During gamete formation, during meiosis, the distribution of each allele from a pair of homologous chromosome is entirely independent of the distribution of alleles of other pairs. During metaphase I of meiosis homologous chromosomes lineup on the equator of the spindle and subsequently separate (segregate) independently during metaphase I and move to opposite poles independently during anaphase I which leads to a variety of allele recombination in the gametes formed, as long as each gamete has one allele for each gene.

Example

In the garden pea plant, the gene controlling flower color is located on the same chromosome with that controlling height. Suppose a pure breeding tall red flowered plant is crossed with a white short flowered plant, the F₁ offsprings obtained are tall red flowered plants. If the F₁ offsprings are selfed,

- What would be the phenotypic ratio in the F₂ generation
- If 700 pea plants are formed in F₂ generation, what would be number of pea plants in each phenotypic class
- How would you experimentally determine the genotypes of the F₁ plants

Solution

Let **T** represent allele for tallness

Let **t** represent allele for shortness

Let **R** represents allele for red flower color

Let **r** represents allele for white flower color

Parental phenotype: **tall red flowered pea plant x short flowered**

Parental genotype (2n):

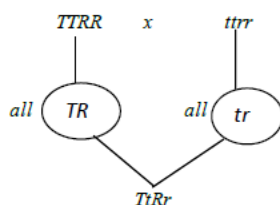
Meiosis

Gametes (n)

Fertilization

Offspring genotype (2n):

F₁ phenotype: all tall red flowered plants

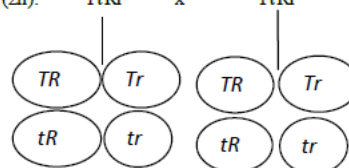


F₁ parental phenotype: tall red flowered pea plant x tall red flowered pea plant

F₁ parental genotype (2n):

Meiosis

Gametes (n)



	TR	Tr	tR	tr
TR	TTRR Tall & red	TTRr Tall & red	TtRR Tall & red	TtRr Tall and red
Tr	TTRr Tall & red	TTrr Tall & white	TtRr Tall & red	Ttrr Tall & white
tR	TtRR Tall & red	TtRr Tall & red	ttRR short & red	ttRr short & red
tr	TtRr Tall and red	Ttrr Tall & white	ttRr short & red	ttrr short & white

F₂ genotypic ratio: 9T_R_ : 3T_rr : 3ttR_ : 1ttrr

F₂ phenotypic ratio: 9 tall red flowered: 3 tall white flowered: 3 short red flowered: 1 short white flowered

- $$= \frac{\text{tall red flowered}}{\text{total}} \times 700 = \frac{9}{16} \times 700 = 393.75 \text{ plants}$$

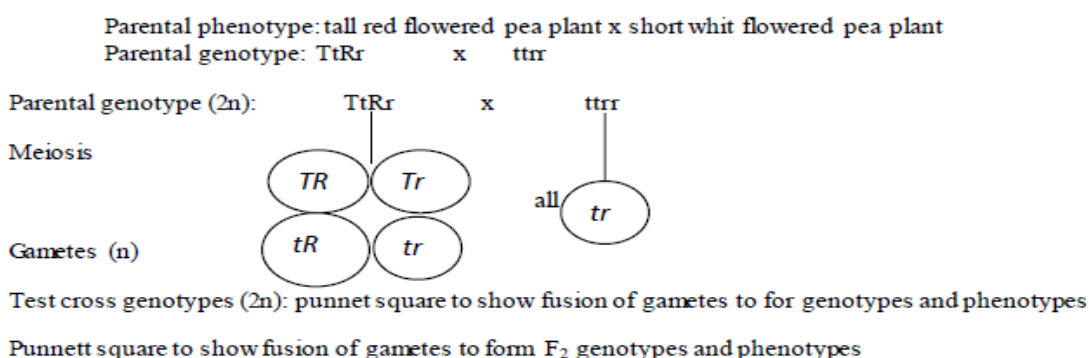
Tall red flowered
- $$\text{flowered} = \frac{\text{tall white flowered}}{\text{total}} \times 700 = \frac{3}{16} \times 700 = 131.25$$

Tall white
- $$\text{flowered} = \frac{\text{short red flowered}}{\text{total}} \times 700 = \frac{3}{16} \times 700 = 131.25$$

Short red
- $$\text{flowered} = \frac{\text{short white flowered}}{\text{total}} \times 700 = \frac{1}{16} \times 700 = 43.75$$

Short white

(c) By carrying out a test cross between F₁ tall red flowered pea plant with short flowered pea plants



	TR	Tr	tR	tr
tr	$TtRr$ Tall & red	$Tttr$ Tall & white	$ttRr$ short & red	$tttr$ short and white

Test cross phenotypic ratio: 1 tall red flowered: 1 tall white flowered: 1 short red flowered: 1 short white flowered.
 This ratio proves that the F_1 plants were heterozygous ($TtRr$).

Test cross phenotypic ratio: **1 tall red flowered: 1 tall white flowered: 1 short red flowered: 1 short white flowered.** This ratio proves that the F_1 plants were heterozygous ($TtRr$).

Note: in dihybrid inheritance, some of the offsprings formed in the F_2 have a mixture of the two parental phenotypes that gave rise to F_1 and such offsprings are known as **recombinants** while other offsprings in F_2 resemble one of the two parental phenotypes that gave rise to F_1 and such offsprings are known as **parentals**.

9 tall red flowered: 3 tall white flowered : 3 short red flowered: 1 short white flowered
(parental offsprings) (recombinant offsprings): (parental offsprings)

Recombinants arise when **crossing over** takes place during the formation of gametes in meiosis which leads to the mixing of the two parental characteristics. The number of recombinants in F_2 is usually smaller than that of the offsprings which resembles the parental phenotypes (parental offsprings). This is because crossing over occurs by chance which reduces the number of recombinants formed.

Example 2

In *Drosophila melanogaster* flies, the gene determining the size of the abdomen occurs on the same chromosome with that determining the length of the wings. When a pure breeding broad and long winged female fly was crossed with a narrow and vestigial winged male fly all the F_1 offsprings obtained head broad abdomen and long wings. If the F_1 offsprings were selfed to obtain F_2 .

- Using suitable genetic symbols work out the phenotypes and genotypes that were obtained in F_2 generation.
- Suppose 480 flies were obtained in F_2 work out the numbers of the flies for each phenotype class.
- How many of these flies were recombinants.

Let B represent allele for broadness
 Let b represent allele for narrow
 Let N represent allele for long
 Let n represent allele for vestigial winged

Parental phenotype: broad and long winged x narrow and vestigial winged

Parental genotype (2n):

Meiosis

Gametes (n)

Fertilization

Offspring genotype (2n):

F₁ phenotype: all broad and long winged flies

F₁ parental phenotype: broad and long winged x broad and long winged

F₁ parental genotype (2n):

Meiosis

Gametes (n)

Punnett square to show fusion of gametes to form F₂ genotypes and phenotypes

O ⁺ O	BN	Bn	bN	bn
BN	BBNN Broad and long	BBNn Broad and long	BbNN Broad and long	BbNn Broad and long
Bn	BBNn Broad and long	BBnn Broad and vestigial	BbNn Broad and long	Bbnn Broad and vestigial
bN	BbNN Broad and long	BbNn Broad and long	bbNN narrow and long	bbNn narrow and long
Bn	BbNn Broad and long	Bbnn Broad and vestigial	bbNn narrow and long	bbnn narrow

F₂ genotypic ratio: 9B_N_:3B_nn: 3bbN_:1bbnn

F₂ phenotypic ratio: 9 broad long winged: 3 broad vestigial winged: 3 narrow vestigial winged: 1 narrow short winged

(d)

i.
$$= \frac{\text{broad and long winged}}{\text{total}} \times 480 = \frac{9}{16} \times 480 = 270 \text{ flies}$$
 Tall red flowered

ii.
$$\text{flowered} = \frac{\text{broad and vestigial winged}}{\text{total}} \times 480 = \frac{3}{16} \times 480 = 90 \text{ flies}$$
 Tall white

iii.
$$\text{flowered} = \frac{\text{narrow and long winged}}{\text{total}} \times 480 = \frac{3}{16} \times 480 = 90 \text{ flies}$$
 Short red

iv.
$$\text{flowered} = \frac{\text{narrow and vestigial winged}}{\text{total}} \times 480 = \frac{1}{16} \times 480 = 30 \text{ flies}$$
 Short white

MODIFICATION OF 9:3:3:1 PHENOTYPIC RATIO

This ratio is mainly modified by the inheritance of lethal genes, linkage of genes and epistasis.

1. EPISTASIS:

This is a form of gene interaction in which one gene suppresses the effect of another gene that is situated at a different locus on a chromosome. **Or**

This refers to a condition in which non-allelic genes interact during which the epistatic allele of the epistatic gene on one locus suppresses the phenotypic expression of the hypostatic allele of the hypostatic gene on another locus.

An **epistatic allele** is the one which suppresses the action of another allele in the phenotype though they are not located on the same locus and the suppressed gene or allele at another locus is the **hypostatic one**.

There are different types of epistasis which include the following;

i. **Dominant epistasis**

ii. **Recessive epistasis**

iii. Duplicate dominant genes

(a) **Dominant epistasis (12: 3: 1):**

This is the type of epistasis where the epistatic allele is dominant such that its presence suppresses the phenotypic expression of the recessive allele on another locus. This type of epistasis changes the phenotypic ratio from 9:3:3:1 to 12:3:1.

When dominant allele 'A' masks the expression of 'B' 'A' is epistatic gene of 'B'. A can express itself only in the presence of 'B' or b allele. Therefore it is called dominant epistasis. B expresses only when 'aa' is present. Therefore, in 9: 3: 3: 1 ratio both 9 and 3 are expressing the 'A' gene the ratio is now 12: 3: 1.

Example:

Complete dominance at both gene pairs, but one gene, when dominant, epistatic to the other.

Fruit colour in summer squash:

Gene pair A: White dominant to colour 12/16 white

Gene pair 'B' – Yellow dominant to green 3/16 yellow

Green – 1/16 both recessive aabb

Dominant white hides the effect of yellow or green.

	AaBb	×	AaBb	
	white		white	
			AB	Ab
			aB	ab
	AB		AA BB	AA Bb
			White	White
	Ab		AA Bb	AA bb
			White	White
	aB		Aa BB	Aa Bb
			White	White
	ab		Aa Bb	Aa bb
			White	White
			aa BB	aa Bb
			Yellow	Yellow
			aa Bb	aa bb
			Yellow	Green

AB – 9 White
Ab – 3 White
aB – 3 Yellow
ab – 1 Green

AB – 9 White
Ab – 3 White
aB – 3 Yellow
ab – 1 Green

(b) Recessive epistasis (9:3:4):

This is the type of epistasis where the epistatic allele is recessive, such that its presence in homozygous condition, suppresses the phenotypic expression of the dominant allele located on another locus. This type of epistasis changes the dihybrid phenotypic ratio from 9:3:3:1 to 9:3:4.

Recessive alleles at one locus (aa) mask the phenotypic expression of other gene locus (BB, Bb or bb) such epistasis is called recessive epistasis. The alleles of 'B' locus express themselves only when epistatic locus 'A' has dominant allele like AA or Aa. The phenotypic ratio is 9: 3: 4.

Example:

Complete dominance at both gene pairs, but one gene, when homozygous recessive, is epistatic to the other.

In Mouse coat colour.

Gene pair A: colour dominant over albino.

Gene pair 'B' agouti colour dominant over black.

Interaction: homozygous albino is epistatic to agouti and black.

Agouti 9/16

Black 3/16

Albino 4/16

(c) Duplicate recessive gene (9: 7):

If both gene loci have homozygous recessive alleles and both of them produce identical phenotype the F₂ ratio 9:3:3:1 would be 9:7. The genotype aaBB, aaBb, AAbb, Aabb and aabb produce same phenotype. Both dominant alleles when are present together only then they can complement each other. This is known as complementary gene.

Examples:

Complete dominance at both gene pairs, but either recessive homozygote is epistatic to the effect of the other gene.

In sweet pea flower colour:

Gene pair 'A' – Purple dominant over white.

Gene pair 'B' – Colour dominant over colourless (white).

Agouti (gray) AABB		×	white albino aabb	
		↓	Aa Bb × Aa Bb	
			Agouti Agouti	
	AB	Ab	aB	ab
AB	AABB Agouti	AABb Agouti	AaBB Agouti	Aa Bb Agouti
Ab	AABb Agouti	AAbb Albino	AaBb Agouti	Aabb Albino
aB	AaBB Agouti	AaBb Agouti	aaBB Black	aaBb Black
ab	AaBb Agouti	Aabb Albino	aa Bb Black	aabb Albino
9/16		3/16	4/16	
Agouti		Black	Albino	

Interaction: Homozygous recessive of either gene A or B produce white

9/16 Purple
 7/16 White
 White flowers × White flower
 CCpp × PPcc
 CcPp × CcPp
 Purple
 CP = 9 Purple
 Cp = 3 White
 cP = 3 White
 cp = 1 White
 9 : 7
 Purple : White

	CP	Cp	cP	cp
CP	CCPP Purple	CCPp Purple	CcPP Purple	CcPp Purple
Cp	CCPp Purple	CCpp White	CcPp Purple	Ccpp White
cP	CcPP Purple	CcPp Purple	ccPP White	ccPp White
cp	CcPp Purple	Ccpp White	ccPp White	ccpp White

(d) Duplicate dominant genes (15:1):

If a dominant allele of both gene loci produces the same phenotype without cumulative effect, i.e., independently the ratio will be 15:1.

Example:

Complete dominance at both gene pairs, but either gene when dominant, epistatic to the other.

Seed capsule of shepherd's purse (Caps ell a bursa-pestoris)

Gene pair 'A': Triangular shape dominant over ovoid

Gene pair 'B': Triangular shape dominant over ovoid (double recessive)

15/16 = Triangular
 1/16 = ovoid (topshaped)
 AABB × aabb
 Triangular × Ovoid
 AaBb × AaBb
 Triangular ↓
 AB = 9 = Triangular
 Ab = 3 Triangular
 aB = 3 Triangular
 ab = 1 Ovoid (top shaped)
 15 : 1
 Triangular : Ovoid

	AB	Ab	aB	ab
AB	AABB Triangular	AABb Triangular	AaBB Triangular	AaBb Triangular
Ab	AABb Triangular	AAbb Triangular	AaBb Triangular	Aabb Triangular
aB	AaBb Triangular	AaBb Triangular	aaBB Triangular	aaBb Triangular
ab	AaBb Triangular	Aabb Triangular	aaBb Triangular	aabb ovoid

(e) Dominant and recessive interaction (13: 3):

Sometimes the dominant alleles of one gene locus (A) in homozygous and heterozygous (AA, Aa) condition and homozygous recessive alleles bb of another gene locus (B) produce the same phenotype.

The F_2 ratio will become 13: 3. The genotype AABB, AaBB, AAbb, Aabb and aabb produce one type of phenotype and genotype aaBb, aaBB will produce another type of phenotype.

Example:

Complete dominance at both gene pairs, but one gene when dominant epistatic to the other, and the second gene when homozygous recessive, epistatic to the first.

Feather Colour of Fowl

Gene pair 'A': colour inhibition is dominant to colour appearance.

Gene pair 'B': colour is dominant to white.

Interaction:

Dominant colour inhibition prevents colour even when colour is present, colour gene, when homozygous recessive prevents colour even when dominant inhibitor is absent.

13/16 = White		3/16 = coloured	
White leg horn		White plymouth Rock	
AABB		aabb	
↓		↓	
AaBb		AaBb	
White		White	
AB = 9 white			
Ab = 3 coloured			
aB = 3 white			
ab = 1 white			
13		3	
White		Coloured	

	AB	Ab	aB	ab
AB	AABB White	AABb White	AaBB White	AaBb White
Ab	AABb White	AAbb* Coloured	AaBb White	Aabb* Coloured
aB	AaBB White	AaBb White	aaBB White	aaBb White
ab	AaBb White	Aabb* Coloured	aaBb White	aabb White

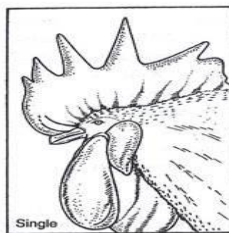
NON EPISTATIC INTER ALLELIC GENETIC INTERACTION:

In some cases two pairs of genes determine same phenotype but assorted independently, produce new phenotypes by mutual epistatic interaction. The F_2 ratio remains the same as 9:3:3:1.

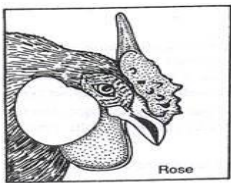
Shape in Poultry (Fig. 39.2)



Walnut



Single



Rose



Pea

Fig. 39.2. Various types of combs in cock.

Example:

Each gene pair affecting the same character complete dominance at both gene pairs, new phenotypes resulting from interaction between dominants, and also from interaction between both homozygous recessives.

Gene Pair A: Rose comb dominant over non rose.

Gene Pair B: Pea comb dominant over non pea.

Interaction:

Dominant of rose and pea produce walnut comb.

Homozygous recessive for rose and pea produce single comb.

Rose comb	×	Pea comb	
AAbb (Rrpp)	↓	aaBB (rrPP)	
	×	AaBb (RrPp)	AaBb(RrPp)
		Walnut	Walnut
		AB	Ab
		aB	ab
9/16 = Walnut	AB	AABB	AABb
3/16 = rose	Ab	AABb	AAbb*
3/16 = Pea	aB	AaBB	AaBb
1/16 = single	ab	Aabb*	aabb*
AB = 9 = Walnut		Walnut	Walnut
Ab = 3 = Rose		Rose	Rose
aB = 3 = Pea		Pea	Pea
ab = 1 = Single		Single	Single
	9	3	3
	Walnut	Rose	Pea
			Single

According to Bateson and Punnett such result is obtained because by combination of homozygous recessive genes (bb) and a homozygous or heterozygous dominant AA or Aa the Rose comb is produced; and by combination of homozygous recessive (aa) and homozygous or heterozygous dominant condition BB or Bb produce pea comb while the single type comb is produced by double recessive aabb genes. 'A' gene determines the shape of rose comb and 'B' gene determines the shape of Pea comb, but when both genes combine a new shape Walnut appears. In the cross between two types of Walnut chickens 4 phenotypes appear. The genes here do not determine themselves in the development of character but modify the character determined by a basic gene. These genes are called supplementary or modifying genes.

INHERITANCE OF COMPLEMENTARY GENES

These are two or more genes which interact together in order to control a single characteristic in an organism.

Inheritance of these genes therefore does not agree with Mendel's laws of inheritance.

Although these genes control a single characteristic, they show independence assortment.

Therefore these genes are passed on from the parents to the offspring in a normal Mendelian fashion. The best example of complementary genes is genes which control the shape of combs in chicken.

In chicken there are four types of combs namely;

- walnut comb
- single comb
- pea comb
- rose comb

These four types of combs are controlled by the two genes located at two loci situated on different chromosomes and which interact together to give rise to the four comb types. The shape of the combs is controlled by two genes which are represented by two alleles shown below;

Let **P** represent the allele for **pea comb**

Let **R** represent the allele for **rose comb**

The pea comb develops in the presence of the **P**-allele and in the absence of the **R**-allele while the rose comb develops phenotypically in presence of **R**-allele and in the absence of the **P**-allele. When both alleles, P and R, are present together a walnut comb develops. A single comb appears only in the homozygous double recessive condition

Consider a cross between a pea comb shaped cock with a rose combed hen whose F1 offspring are then selfed. What is the phenotypic ratio obtained in F2?

Parental phenotype: pea combed cock x rose combed hen

Parental genotype (2n): PPr x $ppRR$

Meiosis

Gametes (n) all Pr all pR

Fertilization

Offspring genotype (2n): $PpRr$

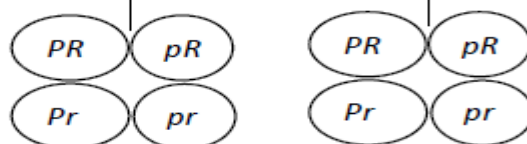
F₁ phenotype: all walnut combed chicken

F₁ parental phenotype: walnut combed chicken x walnut combed chicken

F₁ parental genotype (2n): $PpRr$ x $PpRr$

Meiosis

Gametes (n)



Punnett square to show fusion of gametes to form F₂ genotypes and phenotypes

O ⁺ O	PR	Pr	pR	Pr
PR	PPRR Walnut comb	PPRr Walnut comb	PpRR Walnut comb	PpRr Walnut comb
Pr	PPRr Walnut comb	PPrr Pea comb	PpRr Walnut comb	Pprr Pea comb
pR	PpRR Walnut comb	PpRr Walnut comb	ppRR rose comb	ppRr rose comb
pr	PpRr Walnut comb	Pprr Pea comb	ppRr rose comb	pprr single comb

F₂ genotypic ratio: 9P_R_:3P_rr: 3ppR_:1pprr

F₂ phenotypic ratio: 9 walnut combed: 3 pea combed: 3 rose combed: 1 single combed

In this inheritance, the genes are usually situated at different loci at different chromosomes from where they interact together and give rise to four distinct phenotypes for a single characteristic.

The walnut comb results from a modified form of co-dominance in which atleast one dominant allele of either pea comb or rose comb is present.

This is an incidence where by a 9:3:3:1 phenotypic ratio is obtained for a single characteristic. Although this ratio is this pattern of inheritance differs from the hybrid inheritance because;

- The F1 progeny (offsprings) resembles neither parents i.e. they are all walnut comb shaped unlike their parents.
- The F2 progeny also contains two new phenotypes which do not exist in the F1 parents namely walnut and a single comb shaped and these appear in a higher ratio as compared to the rose and the pea comb

Examples

In oats the inheritance of color is controlled by the epistatic gene which has two alleles, one allele being dominant for color appearance while the other allele is for no color formation (white or albino) i.e. the hypostatic gene is responsible for color deposition or type of color. Where by black is dominant over white

Consider a cross between homozygous black oat plant with a homozygous white oat plant and then the F1 plants are selfed to get F2.

- Were out the phenotypic ratio of the F2 generation
- How many individuals are found in each of the phenotypic classes obtained in F2 if 130 individuals were found in F2?

Let **G** represent the allele for color formation (epistatic)

Let **g** represent the allele for no color formation (albino\ white)

Let **B** represent the allele for black

Let **b** represent the allele for grey

Parental phenotype: black oat plant x white oat plant

Parental genotype (2n): $GGBB \times ggbb$

Meiosis

Gametes (n) all GB all gb

Fertilization

Offspring genotype (2n): $GgBb$

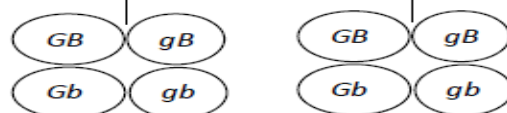
F1 phenotype: all black oats

F1 parental phenotype: Black oat plant x Black oat plant

F1 parental genotype (2n): $GgBb \times GgBb$

Meiosis

Gametes (n)



Punnett square to show fusion of gametes to form F2 genotypes and phenotypes

	GB	Gb	gB	gb
GB	GGBB Black oats	GGBb Black oats	GgBB Black oats	GgBb Black oats

Gb	GGBb Black oats	GGbb Grey oats	GgBb Black oats	Ggbb Grey oats
gB	GgBB Black oats	GgBb Black oats	ggBB white oats	ggBb white oats
gb	GgBb Black oats	Ggbb Grey oats	ggBb white oats	Ggbb White oats

F₂ phenotypic ratio: 9 black oats: 3 grey oats: 4 white oats.

i. $\frac{\text{black oats}}{\text{total}} \times 480 = \frac{9}{16} \times 130 = 73.125 \text{ plants}$ Black oats =

ii. $\frac{\text{grey oats}}{\text{total}} \times 480 = \frac{3}{16} \times 130 = 24.375 \text{ plants}$ Grey oats =

iii. $\frac{\text{white oats}}{\text{total}} \times 480 = \frac{4}{16} \times 130 = 32.5 \text{ plants}$ White oats =

Question2

In white leghorn fowl plumage colour is controlled by two sets of genes, including the following.

W (white) dominant over **w** (colour)

B (black) Dominant over **b** (brown)

The heterozygous F₁ Genotype **WwBb** is white. Account for this type of gene interaction and show the phenotypic ratio of the F₂ generation

LINKAGE OF GENES

Linked genes are more than two genes located on the same chromosome but controlling different characteristics and are inherited together as a single block.

Linkage is the occurrence of more than one gene on the same chromosome which are inherited together along with the chromosome as a single block. **Or**

Linkage is the existence of genes determining different characteristics on the same chromosomes such that the two are inherited together.

Types of linkage

1. Complete linkage

This is a condition where two or more genes are located so close to each other on a chromosome that they are inherited together since they pass to the same gamete. No genetic recombination occurs between them. (No crossing over)

2. Incomplete linkage

Condition where two or more genes are located relatively further apart on the same chromosome that during crossing over, new genetic recombination occur between them. These genes may not be inherited together.

Linked characteristics are the ones controlled by genes located on the same chromosome and so are transmitted together with the chromosome from generation to generation.

Although these genes occur on the same chromosome, each one controls the specific characteristic irrespective of the other. However, linked genes do not show independent assortment during gamete formation and therefore the phenotypic ratio obtained in **F₂ is 3:1 instead of the expected 9:3:3:1** for the two linked characteristics.

Sometimes crossing over occurs, thereby separating the linked genes on the chromosomes leading to the formation of recombinant gametes during meiosis and this gives an F₂ phenotypic ratio of 9:3:3:1 for the linked characteristics.

Example

In drosophila flies the genes controlling body color and the length of wings occur on the same chromosomes and are linked together. Consider a cross between a pure breeding grey bodied long winged fly with a black bodied vestigial winged fly whereby the grey bodied is female while the black bodied is male. If all the F₁ flies obtained here grey bodied and long winged what are the phenotypic and genotypic ratios of the F₂ flies

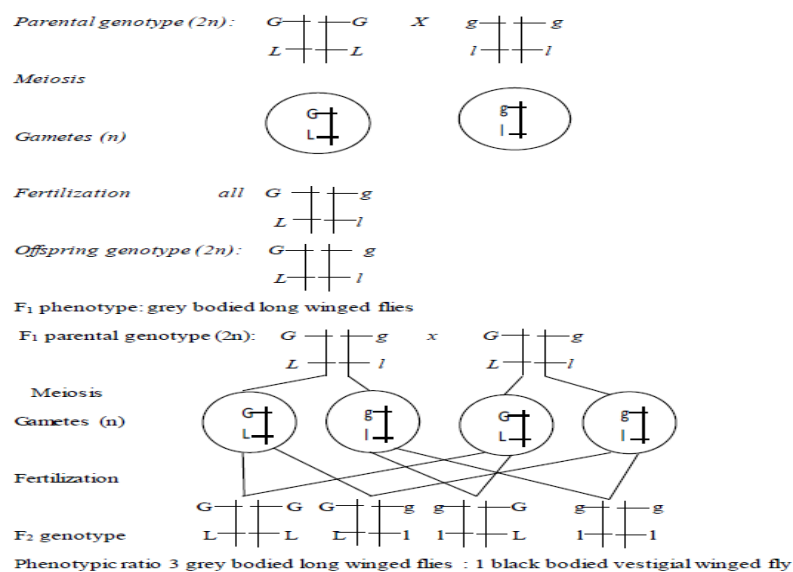
Let **G** represent the allele for grey body

Let **g** represent the allele for black body

Let **L** represent the allele for long wings

Let **l** represent the allele for vestigial wings.

Parental phenotype: *black oat plant X white oat plant*



Phenotypic ratio **3** grey bodied long winged flies: **1** **black** bodied vestigial winged fly

The above results are correct if no crossing over during gamete formation.

In case the genes are not completely linked together in the chromosome crossing over can occur between the non - sister chromatids so as to produce recombinant gametes and this gives a phenotypic ratio in F₂ of the 9:3:3:1 as shown below.

F₂ genotypes: punnet square to show fusion of gametes

	GL	Gl	gL	gl
GL	GGLL Grey body long wings	GGLl Grey body long wings	GgLL Grey body long wings	Gg Ll Grey body long wings
Gl	GGLl Grey body long wings	GGLl Grey body vestigial wings	Gg Ll Grey body long wings	Gg ll Grey body vestigial wings
gL	GgLL Grey body	Gg Ll Grey long	ggLL Black long	gg Ll black long
	long wings	body wings	winged	winged
Gl	Gg Ll Grey body long wings	Gg ll Grey body vestigial wings	gg Ll Black body long winged	gg ll Black body vestigial winged

F₂ phenotypic ratio: 9 grey body long winged flies: 3 grey body vestigial winged flies: 3 black body long winged flies: 1 black body vestigial winged flies

CROSSOVER VALUE AND CHROMOSOME MAPS

The recombinant gametes are formed by crossing over between non-sister chromatids in meiosis I. Such recombinant gametes lead to recombinant offsprings in the phenotypes. Recombinant gametes and recombinant offsprings occur in lower numbers compared to the parental gametes and parental offsprings because crossing over occurs by chance. The percentage of recombinant offsprings in the progeny (total number of offsprings) gives a cross over value which indicates the relative distance between the genes on the chromosomes and the likely hood of undergoing crossing over.

Cross over value (C.O.V) = $\frac{\text{number of recombinants}}{\text{Total number of offsprings}} \times 100\%$

The distance between genes on the chromosome is measured in arbitrary units known as **map units**. 1 map unit = 1 cross over value (C.O.V)

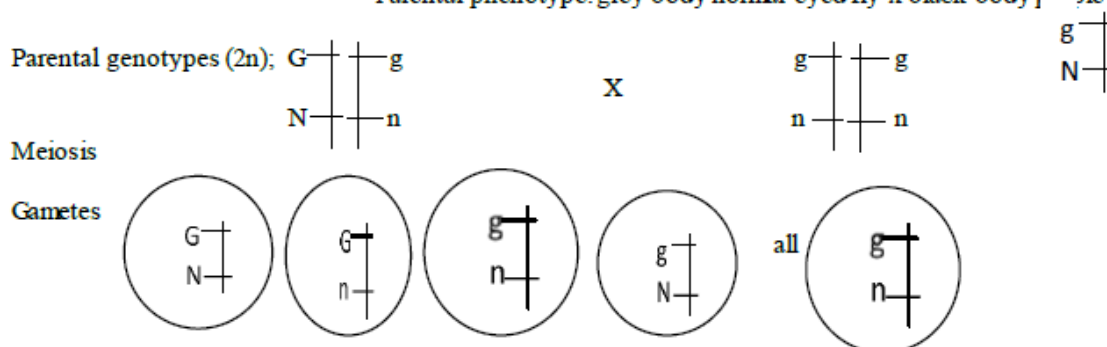
The larger the C.O.V, the more separated the two genes are on the chromosomes and the higher the chances of crossing over taking place. The illustration of the distance between the genes on the chromosome gives the chromosome map i.e. a figure that shows a relative distance between the genes on the same chromosomes. The illustration of distance between the genes on the chromosome is the **chromosome map**.

Example 1

In drosophila flies the genes controlling body color and eye color occur on the same chromosome and are linked together. In an experiment, a heterozygous female fly for grey body and normal eyes was crossed with a black body and purple eyed fly. In these flies , grey body is dominant over black while normal eyes are dominant over purple flies. If 1000 offsprings were obtained from this cross as shown in the table below;

Expected number	Phenotype	Genotype	Number obtained
250	Grey, normal eyes	GgNn	480
250	Grey, purple eyes	Ggnn	18
250	Black, normal eyes	ggNn	17
250	Black, purple eyes	Ggnn	485

a) Parental phenotype: grey body normal eyed fly x black body purple eyed fly



	GN	Gn	gN	Gn
gn	GgNn Grey normal eyes	Ggnn Grey purple eyes	ggNn black normal eyes	Ggnn Black purple eyes.

Test cross phenotypic ratio

1 grey normal eyed fly: 1 grey purple eyed: 1 black normal eyed: 1 black purple eyed.

The obtained results in the test cross differ from the expected ones because the genes are linked together on the chromosomes and were separated by crossing over which occurs by chance hence resulting into formation of fewer recombinants compared to the parents.

Expected number	Phenotype	Genotype	Number obtained
250	Grey, normal eyes	GgNn	480
250	Grey, purple eyes	Ggnn	18
250	Black, normal eyes	ggNn	17
250	Black, purple eyes	Ggnn	485

b)

$$\text{Cross over value} = \frac{\text{number of recombinants}}{\text{total number of offsprings (total progeny)}} \times 100\%$$

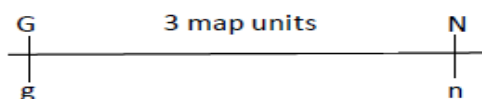
$$= \frac{18+17}{1000} \times 100\%$$

$$= 3.5\%$$

Therefore 3.5% of 3.5 map units

c)

$$1 \text{ C.O.V} = 1 \text{ map unit}$$



Example 2

Further experiment on these flies indicated that the genes for body color, length of wings and eye color are on the same chromosomes. Using the information in the table below calculate the cross over value and illustrate the distance between the genes.

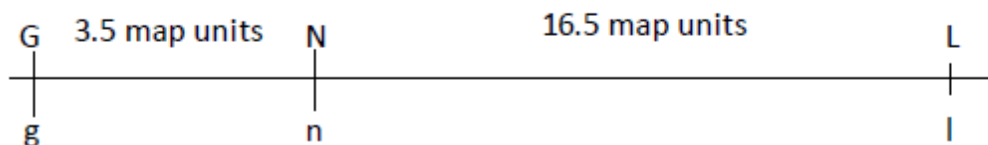
<i>Expected</i>	<i>Phenotype</i>	<i>Genotype</i>	<i>Obtained</i>
250	Grey, long	Gg Ll	400
250	Grey, vestigial	Gg ll	95
250	Black, long	gg Ll	105
250	Black, vestigial	gg ll	400

$$\text{Cross over value} = \frac{\text{number of recombinants}}{\text{total number of offsprings (total progeny)}} \times 100\%$$

$$= \frac{95+105}{1000} \times 100\%$$

$$= 20 \%$$

Therefore 3.5% of 3.5 map units



NOTE: Drawing the chromosome map is also called **gene mapping** where the position of genes are shown on the chromosomes as well as the distance separating with them. Sometimes it is possible to indicate many genes on chromosome and their distances of separation. Consider the cross over values involving for different genes P,Q,R and S.

The distance separating these four genes is shown below;

P-Q = 24%

R-P = 14%

R-S = 8%

S-P = 6%

Draw the chromosome map to show the position of these chromosomes.

Answer. Draw the chromosome map for these genes

a. Insert the positions of the genes with the smallest cross over value in the middle of the chromosome map.

b. Examine the next largest cross over value and insert both possible positions of its genes on the chromosomes relative to either S or P.

c. Repeat the procedure for all the remaining cross over values until you reach the largest cross over values.

Importance of cross over values

- i) Used to show the relative positions of linked genes of a chromosome.
- ii) Used to construct a gene/chromosome map
- iii) Used to show the possibility of crossing over occurring between linked genes
- iv) Used to show the distance between linked genes. I.e. a cross over value of 1 % is equivalent to the distance of 1 unit.

FACTORS THAT AFFECT CROSSING OVER

1) The relative distance between the genes on the chromosome. When the genes are far apart from each other on the chromosome, they have high chances of forming chiasmata in between thereby leading to genetic exchange on the other hand when genes are very close to each other on the chromosome, their chances of forming chiasmata is limited.

2) The position of the centromere on the chromosome. If the genes are very close the centromere their chances of undergoing genetic exchange are limited. However, if the genes are far away from the centromere, there are high chances that they can be exchanged by crossing over.

3) Temperature. Crossing over decreases with increase in temperature because the process of meiosis requires suitable temperature that can promote efficient crossing over.

4) Age of the organism. Increase in age lowers the chances of crossing over. Meiosis is more efficient in grown up adults before menopause stage in females and before senescence in male.

5) Mutagens. These can decrease or increase the rate of crossing over. The chances of crossing over are greatly reduced by presence of chemical substances that inhibit chiasmata formation thereby preventing cross over e.g. in drosophila flies.

INHERITANCE OF SEX

The sex of an organism is determined by two factors namely; environmental conditions and genetic factors.

Environmental determination of sex

In lower animals, sex can be determined by environmental factors such as temperature, salinity, type of food e.t.c. for example in tadpoles the eggs laid in cool places develop into males while those laid in warm places develop into females.

Genetic sex determination

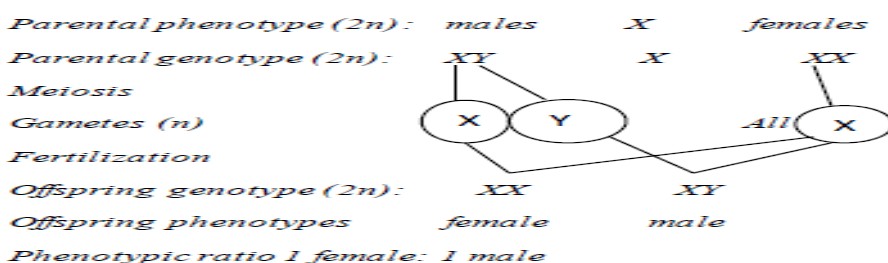
The sex organs development can be determined by the sex genes or the chromosomes. Under chromosomal sex determination, sex can be determined by;

- The number of chromosomes. E.g. in bees the females are diploid and have 32 chromosomes while the males are haploid and have 16 chromosomes. In grasshoppers the females have 24 chromosomes while the males have only 23 chromosomes.
- The sex chromosomes. In heterogametic organisms there are two sex chromosomes that determine the sex of an individual namely the X and Y chromosomes e.g.

The females are **XX** and are described as **homogametic** while the males are XY and are described as **heterogametic**. Therefore in these organisms it is the presence of the Y chromosome that makes one a male and its absence makes one a female. This implies that it is the type of sperm (whether X or Y) that fertilizes the egg which determines the sex of the offspring.

In birds sex is determined by the X and Y chromosomes except that the females are XY while males are XX.

In grasshoppers sex is only determined by X chromosomes where by the males are XO i.e. they lack only one X chromosome while the females are XX.



SEX LINKED CHARACTERISTICS

This is the transmission of characteristics (from parents to offsprings) whose genes are located on the sex chromosomes i.e. the genes controlling such a character are transmitted along with those that determine sex on the same chromosome.

Most sex linked characters are controlled by genes located on the X chromosome and very few are controlled by genes located on Y chromosome. Examples of sex-linked characteristics include; haemophilia, red green colourblindness, brown teeth, **Duchenne muscular** dystrophy e.t.c. Sex linked characteristics can therefore be defined as those whose genes controlling them occur on the sex chromosome and yet they do not determine sex.

Sex linked characters are often expressed more in males than females. This is because the males being heterogametic have a non-homologous portion of the X chromosome while the sex

linked allele is located and therefore such an allele cannot be suppressed in the phenotype by any other dominant allele. the condition.

Why sex-linked traits are most common in males among humans

In humans, males have one X chromosome and one Y chromosome, while females have two X chromosomes.

Most sex-linked traits are determined by recessive genes, carried on the non-homologous portion of the X chromosome, for which the Y chromosome is genetically empty.

In males a recessive allele on the X chromosome is expressed phenotypically, because the Y chromosome cannot carry the dominant allele.

In females a recessive allele on one X chromosome is usually masked/suppressed by the dominant allele on the other X chromosome, such heterozygous females are carriers and phenotypically normal.

Females only express that trait phenotypically in the double recessive state when the recessive allele is present on each of the two X chromosomes. But since the recessive alleles determining sex-linked traits are rare in human populations, this is quite unlikely to occur.



This implies that the genes of the sex linked characters of the males are located in the non-homologous portion and therefore whenever a recessive allele of these characters appears, it has to be expressed in the phenotype since it does not have a counterpart allele that can suppress its phenotypic expression.

In the case of females, the X chromosomes are completely homologous to each other and this gives chances of development of carrier females (heterozygous females) for sex linked characters who may never express the characteristic in the phenotype as the recessive sex linked allele would be suppressed by the dominant allele on the counterpart X chromosome.

Sex linked characters are determined by recessive alleles. However sex linked characteristics undergo a characteristic **cross pattern of inheritance** i.e. the fathers transmit their sex linked characters to their grandsons through their daughters who are carriers this implies that the father will not transmit the sex linked character to his sons but instead to his daughters. This is because

the son only inherits the father's Y chromosome and not the X chromosome that controls the sex-linked characteristics.

Although sex linked characters are mostly carried on the X chromosome there are a few of them which are carried on the Y sex chromosome and these are called **holandric characters** i.e. development of many hairs in the nostrils and ears.

Why there are more colourblind individuals than hemophiliacs.

Haemophilia is potentially lethal and therefore is selected against as it has a killing effect sufferers being less likely to survive to sexual maturity and so produce offspring. Colour blindness is not lethal and thus not selected against; sufferers survive to sexual maturity and produce offspring.

Haemophiliacs may choose not to have children knowing the fatal risk that their offspring may suffer the disease. Colourblind individuals freely produce children as there is no special risk associated with being colourblind.

Haemophiliac females rarely survive to child-bearing age due to the special risk associated with menstrual bleeding; hence only carrier females pass on the disease. With colorblindness both (female) sufferers and carriers can pass on the condition

Worked examples

Consider a normal man who marries a female whose father was having haemophilia and the mother was homozygous normal. Using suitable genetic symbols, work out the phenotypes and genotypes of their offspring. What is the probability that this couple will produce a haemophilic boy?

Let H represent allele for non haemophilic (normal condition)

Let h represent allele for haemophilic

Parental phenotype: haemophiliac father X Non haemophiliac mother

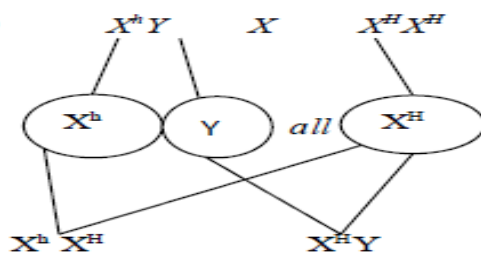
Parental genotype (2n)

Meiosis

Gametes

Fertilisation

Offspring genotype



Parental phenotype: normal man X carrier woman

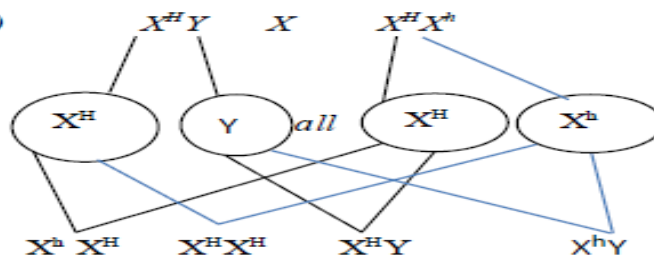
Parental genotype (2n)

Meiosis

Gametes

Fertilisation

Offspring genotype



Offsprings phenotype: normal girl: normal girl: normal boy: haemophiliac boy

Offsprings phenotypic ratio 3 normal: 1 haemophiliac

Male phenotypic ratio; 1 normal: 1 haemophiliac

Probability = $\frac{1}{2}$

Note:

Haemophilia is a condition whereby blood takes too long to clot after an injury leading to excessive bleeding of the victim. This makes hemophiliac individuals rare in population as most of them die before reproductive age.

Color blindness is a condition whereby an individual fails to see the colors or fails to distinguish between particular colors e.g. red, green color blindness where the red and green cone cells of the eye retina are defective due to some sex linked gene in an individual which does not allow such an individual to distinguish between red and green. Colour blind individuals are more common in the population than haemophiliacs because haemophiliacs have higher chances of dying before reaching reproductive age to pass on their genes to the next generations whereas colorblind individuals survive and reach reproductive age in most cases which enables them to reproduce and pass on their gene of colour blindness to the next generation which increases their number in the population. Besides, haemophiliac people may not choose to marry due to the lethal gene they have, thereby becoming unable to pass on their genes to the next generation.

Example 2

Green color blindness is sex linked in man. A normal man married a color blind woman. Using suitable genetic symbols work out the genotypes and phenotypes of their children.

Let C represent the allele for normal color vision

Let c represent the allele for color blindness

Parental phenotype: normal man X colour blind woman

Parental genotype (2n)

$X^C Y$ X $X^c X^c$

Meiosis

Gametes

X^c Y all X^c

Fertilisation

Offsprings genotype

$X^c X^c$ $X^c Y$

SEX LIMITED AND SEX INFLUENCED CHARACTERISTICS

Sex limited characteristics are the ones which occur particularly in one sex. These characteristics occur at a later stage in the life of an organism e.g. in human beings they normally occur at puberty.

In human beings the males have the following sex limited characteristics beard, deep voice, hairs in the ears and nostrils, porcupine characteristics e.t.c. In human females these characteristics include development of breasts, widening of the hip girdles e.t.c.

Sex influenced characteristics are those whose dominance is determined by the sex of the bearer e.g. baldness of the head occurs in males and not in females because the genes determining it are dominant in males and recessive in females.

MULTIPLE ALLELES

This is another form of co-dominance. Multiple alleles refer to more than two possible alleles of which any two can occur at a single locus at any one time.

Examples of x-tics controlled by multiple alleles include;

a. Blood groups in humans

b. Coat color in rabbits

c. Eye color in rabbits and mice

Inheritance of ABO blood system

The ABO blood group system is controlled by three alleles of a gene **I** (isohaemagglutinin) occur at a single locus any time. These alleles are A or I^A , B or I^B and O or I^O . These alleles I^A and I^B are equally dominant while the allele. I^O is recessive to both.

The transmission of thee alleles occurs in a normal Mendelian fashion.

The table below summarizes the possible phenotype and blood group.

GENOTYPE	PHENOTYPE
$I^A I^A$ (AA)	Blood group A (homozygous)
$I^A I^O$ (AO)	Blood group A(heterozygous)
$I^B I^B$ (BB)	Blood group B
$I^B I^O$ (BO)	Blood group B
$I^A I^B$ (AB)	Blood group AB (co-dominant)
$I^O I^O$ (OO)	Blood group O

Physiology of the blood groups in humans

Human blood contains blood group antigens and blood group antibodies. Some of these specifically determine blood groups e.g. allele **A** determines the production of antigen A, allele B determines the production of antigen **B** and allele **O** does not code for the production of any antigens. Antigens **A and B** occur on the plasma membranes of red blood cells. These antigens have corresponding protein molecules known as blood group antibodies (agglutinins) in blood plasma. These antibodies can react with the antigens under the lock and key hypothesis should they be similar to the antigens brought into the recipient's blood, leading to the formation of a precipitate or an agglutinate in blood.

Therefore an individual should not have blood group antibodies corresponding or similar to his blood group antigens in order to avoid agglutination. Consequently, individuals should have the following antibodies not corresponding to their antigen to avoid blood clotting.

Blood group	A	A	B	B	AB	O
Antigen	A	A	B	B	AB	None
Antibody	A	A	B	b	None	a and b

Example

1. A man having blood A marries a woman having blood group AB. What are the possible genotypes and phenotypes of their offsprings if the man is heterozygous for blood group A?

Let I^A represent the allele for formation of antigen A

Let I^B represent the allele for formation of antigen B

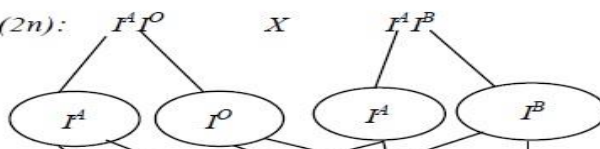
Let I^O represent the allele for no formation of antigen A or B

Parental phenotype: blood group A x blood group AB

Parental genotype (2n):

Meiosis

Gametes (n)



Offspring genotype (2n): $I^A I^A$ $I^A I^B$ $I^A I^O$ $I^O I^B$

F2 genotypic ratio: $1I^A I^A$: $1I^A I^B$: $1I^A I^O$: $1I^O I^B$

F2 phenotypic ratio: 2 blood group A: 1 blood group AB: 1 blood group B

Question 2

A boy has blood group A and his sister

has blood group O. which combination of genotypes and phenotypes do you think their parents have. Show your working.

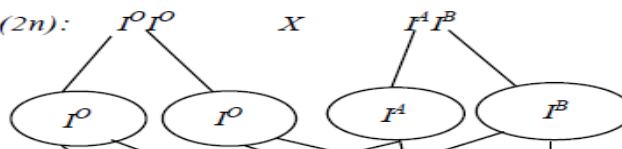
Parental phenotype: blood group O x blood group AB

Parental genotype (2n):

Meiosis

Gametes (n)

Fertilization



Offspring genotype (2n): $I^O I^A$ $I^O I^B$ $I^A I^O$ $I^B I^O$

F2 genotypic ratio: $2I^A I^O$: $2I^B I^O$ = $1I^A I^O$: $1I^B I^O$

F2 phenotypic ratio: 1 blood group A: 1 blood group B

Inheritance of ABO blood groups in man

ABO blood groups are determined by the multiple alleles **A**, **B**, and **o**, of an autosomal gene, whose locus is represented by the symbol **I** for **Isohaemagglutigen**, whereby **alleles A** and **B** are equally dominant, while **o** is recessive to both **A** and **B**.

Inheritance of any of the following allelic combinations via fertilization, results in possession of any of the four blood groups; **A**, **B**, **AB** and **O**.

Blood group A is determined by inheritance of genotype $I^A I^A$ or $I^A i$ through fertilization, determining production of **antigen/agglutinin A** on the red blood cell/erythrocyte cell membrane, and possession of **agglutinin anti-B/antibody b** in plasma.

Blood group B is determined by inheritance of genotype $I^B I^B$ or $I^B i$ through fertilization, determining formation of **antigen/agglutinin B** on the erythrocyte cell membranes, and possession of **agglutinin anti-A/antibody a** in plasma.

Blood group AB is determined by inheritance of genotype $I^A I^B$ through fertilisation, determining formation of **antigens/agglutinogens A and B** on the erythrocyte cell membranes, and absence of agglutinins in plasma.

Blood group O is determined by inheritance of genotype ii through fertilization, determining absence of agglutinogens on the erythrocyte cell membrane, and possession of both **agglutinins a and b** in plasma.

The importance of blood groups

a. They are important during **blood transfusion** where they are used to prevent agglutination (precipitation) of blood of the recipient. To avoid agglutination, the donors blood group should be compatible (matching with) to that of the recipient by having the donors blood group antigen that is different from the blood group antibody of the recipient.

When the recipient's gets antibodies from the donor, such antibodies become diluted in the recipient's blood and so cause either minor clotting of blood or no blood clotting at all and so cannot lead to death of the recipient. However, in case the donor introduces an antigen that is similar to the antibody of the recipient, it stimulates the recipient's blood to produce more antibodies which attack and react with the donor's antigen to cause severe blood clotting. Therefore an individual with a specific antigen on the red blood cell membrane does not possess its corresponding antibody in the blood plasma to avoid agglutination.

Blood plasma permanently contains two blood group antibodies a and b which do not correspond with a specific antigen in blood to avoid agglutination e.g. a person with blood group A has antigen A and antibody to avoid agglutination. A person with blood group B cannot donate blood to a person of blood O because antigen B in the donor's blood will be attacked by antibody b in the recipient's blood leading to agglutination. The same applies to blood group A and blood group AB donors to blood group O recipients.

It is possible for blood group A to donate blood to blood AB, because the donors blood, blood group A, has antigen A which cannot stimulate the recipient's blood group AB to attack antigen A since blood group AB individuals lack antibodies that can attack antigen A to cause an agglutination.

A person of blood group **AB** cannot donate blood to a person of blood group **O**. This is because the donor's blood has antigen **A** and antigen **B**, which stimulate the recipient's blood to

produce corresponding antibodies **a and b**, which then attack and react with antigen A and B in the recipient's blood.

Blood group **AB** individuals can receive blood from all other individuals having other blood groups. Therefore individuals with blood group AB are called **universal recipients**. This is because such individuals have no antibodies in their blood plasma that can react with antigens A and B in the donor's blood.

Individuals with blood group O can donate blood to all other blood groups and are therefore called **universal donors**.

This is because blood group O individuals do not have any antigens in their red blood cells that can react with antibodies in the blood plasma of the recipient to cause agglutination. The table below summarises the possible and impossible blood transfusions.

Recipient		Donor's blood group			
Blood group	Antibody in plasma	A	B	AB	O
A	B	✓	X	X	✓
B	A	X	✓	X	✓
AB	None	✓	✓	✓	✓
O	a and b	X	X	X	✓

✓ = compatible with recipients blood

X = Incompatible with recipient i.e. agglutination occurs

b. They are used in **settling court cases** about who the father of the child is (i.e. paternity suits). Although blood groups cannot prove beyond reasonable doubt who the father of the child is it is possible to use their inheritance to show that an individual could possibly be the father of the child.

Consider a mother who is of blood group O having child of blood group O and the child produced also with blood group O. she claims that the father is a man whose blood group is AB. Since the child is blood group O its only possible genotype is IOIO and it must therefore have inherited one IO allele from each parents. Since the man is of blood group AB he cannot donate the IO to the child and therefore he cannot be the father of the child. Even if the father was found to be of another blood group such as blood group A still the evidence will be insufficient because any other man can possess such a blood group and donate the IO allele to the child. Therefore a DNA test should be carried out to confirm who the father of the child is.

c. Blood groups can also be used as an **evidence of evolution**. This is because organisms of different species having similar blood group systems such as the ABO system are believed to have originated from the same ancestor in the course of evolution for example humans, chimpanzees, gorillas, Baboons e.t.c.

THE RHESUS BLOOD GROUP SYSTEM

The rhesus blood group system is also inherited in a similar way to the ABO blood group system. Individuals with red blood cells with the D-antigens (Rhesus factor) are said to be rhesus positive (**Rh+**) however **Rh+** allele is taken to be dominant over the rhesus negative (**Rh-**) allele. The Rhesus factor is controlled by three alleles C, D and E which determine the production of D-antigens on the surface of the red blood cells. Allele mainly determines the production of D-antigens and it is this antigen which is the fundamental determinant of blood grouping under the rhesus blood group system.

Marriage complications of the Rhesus system

If an Rh+ man marries an Rh- woman, most of their children are likely to die immediately after birth or before birth. The first child usually survives because the time is too short for the mother to produce enough antibodies known as anti D agglutinins which can pass to the foetus to cause death. If a mother becomes pregnant after the first child, the Rh+ foetus formed can die due to antibodies of the mother entering the foetal circulation. This is because during the first pregnancy, especially the time of giving birth, the blood of the child which is Rh+ may mix with that of the mother which is Rh-, thereby introducing D-antigens in the mother's blood. Also, some of foetal erythrocytes of the first child with D-antigens in them may cross the placenta and enter the body of the Rh- mother towards the end of the gestation period. D-antigens will then stimulate the mother's blood to produce many antibodies called anti D-agglutinins which attack and react with the D-antigens introduced in the mother's blood if the mother becomes pregnant again and the child is Rh+. These antibodies in the mother's blood will pass via the placenta and enter the foetal blood circulation, where they will attack and react with D-antigens in the child's blood causing the red blood cells of the child to clump together, this disease is known as **haemolytic disease of the new born** (erythroblastosis foetalis). This results into acute anaemia of the foetus which can lead to death of the foetus.

The problem may be solved in two major ways;

- a. The mother may be injected with anti-D-agglutinins in the first 72 hours after her first born so as to make her immune system insensitive towards D-antigens.
- b. By carrying out proper intermarriages where by Rh+ man marries Rh+ woman and Rh- woman gets married to Rh- woman.

Another blood group system in humans called the MN blood group system is controlled by 2 alleles **M and N** which are co-dominant. M and N alleles also determine the production of antigens respectively. Individuals therefore have the following genotypes if this blood group system **MM, NN, MN**.

ASSIGNMENT

1. Suppose a man having blood group A marries a woman who is heterozygous for blood group B what are the possible genotype and phenotypes.
2. A boy has blood group A and his sister has blood group B. what are the possible phenotypes and genotypes of their parents.
3. If a father has blood group A and the mother blood group AB what are the possible genotypes and phenotypes of the offspring.

VARIATION

This is the description of the differences in phenotypic and sometime genotypic characteristics shown by organisms belonging to the same species or natural population due to interaction between the genes and the environment.

Variations can be clearly seen among sexually reproducing organisms due to some differences in genetic constitution that occur during meiosis.

Variations are important because they make organisms better adapted to their environment. This is because some variations within the population are favorable (beneficial) to the organisms possessing them making such organisms better adapted or fit to survive in their environment and this gives a selective advantage to those organisms possessing them. Other variations are unfavorable because they are disadvantaged in the environment and organisms possessing such. The first organisms therefore survive, grow and reproduce and pass on their favorable characteristics to the next generation. If this continues for a long time it leads to the emergence of new species in the population having good characteristics and therefore better adapted to the environmental change. Variation is therefore a raw material for evolution during which new species are formed.

Importance of variation

- To identify and distinguish individuals
- To ensure the survival of a species. Variation introduces differences in an organism which may cause it to be better suited to adapt to changes in the environment.

Examples of variation in humans:

- Height -weight - shape of nose - dimples - eye colour - blood group
- Skin colour - finger print - ability to roll the tongue - type of ear lobe

Variation can be due to genetic factors, environmental factors or the interaction of both genetic and environmental factors.

TYPES OF VARIATION

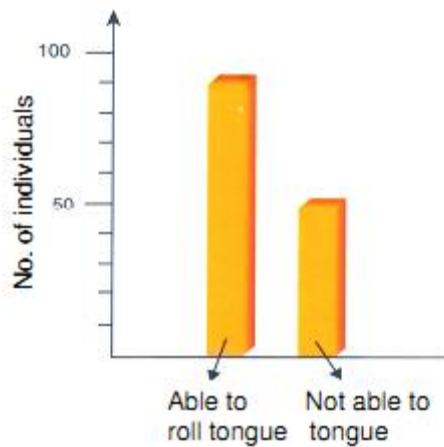
There are two types of variation namely;

- Continuous variation
- Discontinuous variation

Continuous variation

This is the type of variation whereby characteristics in a given population show a smooth gradation between two offsprings with the intermediate phenotype being the majority in the population and few individuals being at the extremes of the characteristics. This implies that organisms do not show any clear cut differences among themselves.

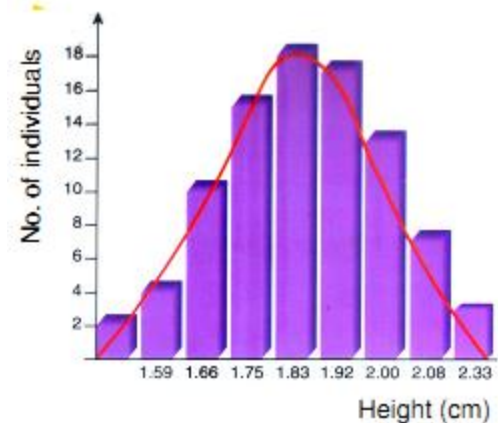
It is brought about by the influence of many genes but can also be influenced by environmental factors. Continuous variation characteristics are therefore influenced by both environmental conditions and genetic factors.



Examples of continuous variation characteristics include skin color, height, weight, intelligence e.t.c. These characteristics are quantitative i.e. they can be measured and are controlled by many genes. These characteristics are therefore described as **polygenic characteristics** i.e. characteristics which are controlled by a number of genes during their transmission i.e. many genes control a single characteristic. These genes are sometimes referred to as **multiple genes**. Although these genes may determine a single characteristic each of them has its own alleles which occur on different loci.

The transmission of x-tics that are controlled by many genes from one generation to another is **called polygenic inheritance** and such characteristics are known as polygenic characteristics. The statistical analysis of these characteristics gives a normal distribution curve shown below;

The above graph shows that continuous variation characteristics appear in the graded pattern and therefore show a smooth graduation. It also shows that most of the individuals in the population lie along the normal.



Discontinuous variation

This is the type of variation where individuals show clear cut differences among themselves in the population with no intermediate phenotypes between them but instead they are grouped into distinct categories. These characteristics are therefore qualitative and cannot be measured. Such characteristics include sex, blood groups in man, tongue rolling, e.t.c.

This variation is controlled by a **single gene** and cannot be influenced by environmental conditions i.e. they are purely genetically controlled.

CAUSES OF GENETIC VARIATION

This is caused by the **gene reshuffling** and **mutations**.

Reshuffling of genes refers to the random orientation of chromosomes at the equator of the spindle during meiosis which changes the positions of the genes on the chromosomes.

Reshuffling of genes include the following;

I. **Crossing over:** this is the exchange of genetic material between the non-sister chromatids of homologous chromosomes during pachytene stage of prophase I of meiosis. This produces new linkage groups and so provides a major source of genetic recombination of alleles on chromosomes which results into formation of recombinant gametes and leads to variation in the offsprings formed. When the gametes undergo random fertilization, offsprings with different genetic constitution are produced.

II. **Independent assortment.** During independent assortment in during metaphase 1, chromosomes are distributed randomly at the equator and segregate (separate). It is by pure

chance as to which chromosome from each homologous pair ends up in a daughter cell at the end of meiosis and therefore all sorts of allele combinations are possible in the gametes. This reshuffles the existing alleles thereby producing new genetic recombination's in of the gametes and the offsprings formed from these gametes when they fuse randomly during fertilisation. Independent assortment can therefore be defined as the random orientation of the chromatids of homologous chromosomes (bivalents) on the equator of the spindle during metaphase 1 of meiosis which determines the direction in which the pairs of chromatids move during anaphase 1. This is so because after random arrangement on the equator of the spindle the chromosomes subsequently segregate (separate) independently thereby leading to the mixing of genes in addition during metaphase II the orientation of the pairs of chromatids is again random at the equator of the spindle and determines which chromosomes migrate to the opposite poles of the cell during anaphase II.

III. Fertilization. Fertilization occurs randomly between the male and female leading to mixing of genes in different combinations.

IV. **Mutation.** Mutations change the genotype of an organism with respect to a specific characteristic as it produces new alleles in the population hence making it to vary due to the combination of mutant and non-mutant gametes during random fertilisation.

V. Genetic drift. This refers to a loss of genes from a small population or the change of genes of a small population by chance alone and not natural selection which results into the change of the gene frequency of the small population. This changes the phenotypic appearance of the organisms thereby making them to vary.

VI. Cross breeding. This mixes genes from different individuals resulting into the formation of hybrids (heterozygotes) with improved qualities compared to the parents. Cross breeding can be defined as mating of organisms that are pure breeding in which one has better x-tics than another which results into the formation of the hybrid offspring.

MUTATION

This refers to the sudden or spontaneous genetic changes which occur in the genetic constitution of an organism. These changes are brought about by mutagens. Mutations change the genotype of an organism with respect to a given characteristic as it produces new alleles in the population. Mutations cause permanent genetic variations unlike reshuffling of genes whose genetic variations are temporary as they can be undone (removed) in subsequent generations due to chromosomes rearranging themselves alongside with their genes.

During mutation, some genetic material may be lost, doubled, inverted, translocated (moved), and mixed, resulting into mutants having different genetic constitution from the non-mutants.

The mutants formed transmit these mutated genes to their offsprings through random fertilisation which makes the offsprings become different from the non-mutants.

Individuals or cells resulting from mutations are known as mutants. The sudden changes in the genetic constitution of an organism are brought about by substances called **mutagens**. The common mutagenic agents include the following;

- Gamma rays
- Alpha and beta particles
- X-rays
- Cosmic rays
- Ultra violet rays
- Excessive heat

- Chemicals such as caffeine and heroin
- DDT and other insecticides
- Colchicine
- Marijuana
- Opium
- Cocaine
- Formaldehyde
- Some food preservatives, color and sweeteners
- Accidents.

Note: mutations usually occur in germ cells during gamete production and so lead to the formation of mutant gametes. When these gametes fuse randomly with mutant or non-mutant gametes of another parent, a mutant offspring is formed which must have unique characteristics compared to the parents. Such mutations are known as **gametic mutations**. Some mutations may occur in somatic cells and are therefore known **somatic mutations** e.g. cancer.

Role of mutation in evolution

Mutation is the sudden, spontaneous and unpredictable change in the amount, arrangement, or structure of the DNA of an organism.

Mutation leads to increased genetic variation in the gene pool of a population.

Somatic mutations occur in somatic cells that do not produce gametes and are only inherited by daughter cells produced by mitosis and so are lost when the organism dies, and do not contribute to evolution.

Mutations occurring in gamete cells and gamete-producing germ cells are inherited by subsequent generations, and contribute to evolution.

Inheritable mutations increase genetic variation, and those mutations which confer a competitive advantage to mutants/bearers are selected for by natural selection, persist and reproduce more offspring; while mutations which confer a competitive disadvantage to mutants/bearers are eliminated as they fail to survive or reproduce.

The resultant change in allele frequency in the population gene pool from generation to generation leads to evolution.

TYPES OF MUTATIONS

There are two types of mutations (germ mutations) namely;

1. Chromosomal mutations
2. Gene mutations (or point mutations)

Somatic mutations cannot be inherited while gametic mutations can be transmitted from parents to the offspring indeed most of the gene and chromosomal mutations are gametic and can therefore be inherited or they are usually recessive.

Chromosomal mutations

This refers to the changes that occur in the chromosome number or chromosome structure but can be transmitted from the parents to the offspring.

Chromosome mutations usually occur during prophase I of meiosis where a number of mistakes are made on the chromosome structure i.e. chromosomes break and join wrongly. It can also arise during anaphase I and II where by some chromosomes may fail to separate and move to opposite poles which brings about an increase in the number of chromosomes or polyploidy i.e.

an increase in the number of chromosomes beyond the normal diploid number. The process by which chromosomes fail to separate during anaphase I of meiosis is known as non-disjunction. Chromosomal mutations are divided into the following categories;

a. Mutations that change the chromosome structure.

b. Mutations that change the chromosome number.

a. Mutations that change the chromosome structure

Such mutations include the following;

i. Deletion

This is a form of mutation where part of a chromosome breaks and gets lost leading to the formation of a number of chromosomes that is shorter than the original chromosome. This is the most dangerous form of mutation because it leads to loss of genes from the chromosome.

In human beings deletion leads to cat cry syndrome where the voice box fails to develop properly.

ii. Inversion

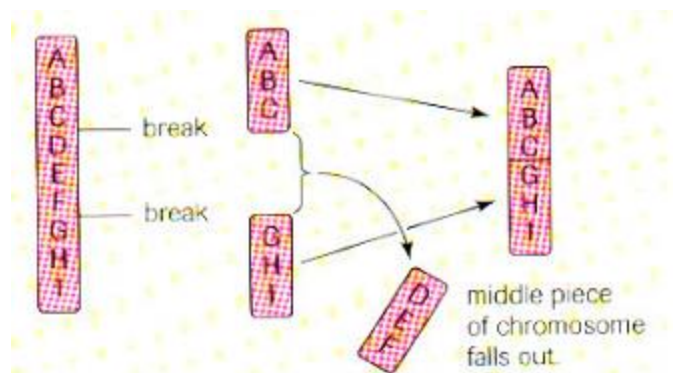
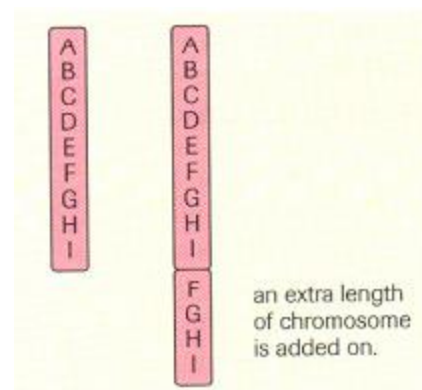
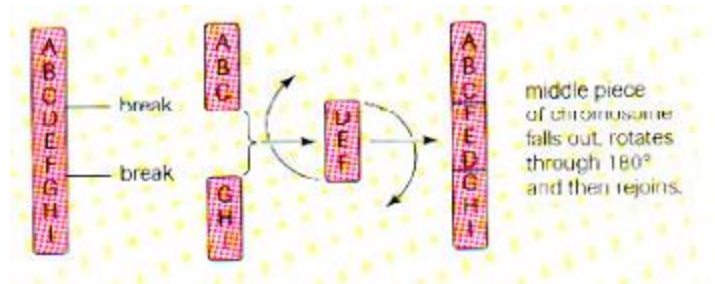
This is the form of mutation where part of a chromosome breaks, rotates through 180 degrees and rejoins in the reverse way, this in turn changes the sequence of genes on the chromosomes as well as the sequence of bases on the DNA strand which makes the offspring vary.

iii. Duplication

This is a form of mutation whereby a portion of chromosomes bearing certain gene is doubled. This form of mutation causes over amplification of certain phenotypes whose genes have been duplicated. This form of mutation is of importance in crop and animal husbandry since it increases yields and improves other characteristics.

iv. Translocation

This is a form of mutation whereby a portion of a chromosome breaks and is moved to join another chromosome which maybe homologous or non-homologous.



b. Mutations that change the chromosome number

These are mutations that affect the whole chromosome and change the chromosome number in the cell. This normally occurs during meiosis at anaphase I and II where two of the same type of chromosomes or chromatids fail to separate and are transmitted together into a single gamete leaving the other gamete empty a concept known as **non-disjunction**. The other chromosomes not affected by non-disjunction are usually distributed normally by meiosis into gametes formed.

Non-disjunction results into formation the formation of gametes either with an extra number of chromosomes ($n+1$) ($n+2$) ($n+3$) e.t.c. or a less number of chromosomes ($n-1$). Therefore, this condition where by half of gametes contain extra number of chromosomes while the other half of gametes formed during meiosis contain a chromosome missing is known as **aneuploidy**.

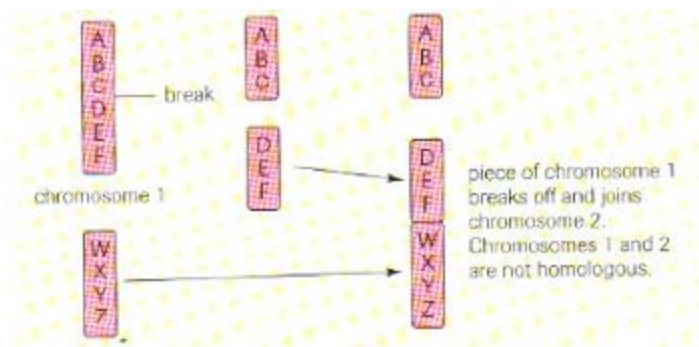
If a chromosome is present in triplicate in the fertilised egg (so that the cell has a total of $2n+1$ chromosomes), the aneuploidy cell formed is said to be **trisomic** and if a chromosome is missing, so that the zygote cell formed has $2n-1$ chromosomes, the aneuploid cell is said to be **monosomic**. Mitosis will subsequently transmit this variation to all embryonic cells (somatic cells) leading to the formation of an organism with variation in the form of a set of symptoms caused by the abnormal dose of genes associated with extra or chromosomes missing.

Some organisms have more than two complete sets of chromosome missing in each of their cells and such organisms are called **polyploids**.

Illustration

Note: The zygote produced with odd number of chromosomes in the above cross containing less than the diploid number of chromosomes usually fails to develop. But those with extra sets of chromosomes though odd numbered or even numbered usually develop and in most cases this produces severe abnormalities. In humans, non-disjunction causes the following abnormalities.

1. Down's syndrome (mongolism)



This disease is also referred to as mongolism and it is caused by an extra autosomal chromosome in position number 21 of the homologous chromosome pairs.

Mongolism occurs as result of non-disjunction in chromosome number or during anaphase I of meiosis in gametes formation leading to formation of abnormal gametes with 24 chromosomes. In such a case if a normal gamete fertilizes an abnormal gamete, the zygote formed will have 47 chromosomes instead of the normal 46. This form of syndrome occurs in both males and females. It can take place during sperm production but it's more common during oogenesis.

Mongolism is due to a type of mutation known as **translocation** in which chromosome 21 is translated or moved to chromosome 14 in most cases or chromosome 22 in some cases.

Most non disjunction occurs in meiosis I where it causes failure of the whole chromosome 21 to separate if it occurs in meiosis II the chromatids fail to separate leading to Down's syndrome. Mongolism causes miscarriage in mothers and the chances of it to occur increases with age of the females. This form of syndrome results in individuals having the following characteristics;

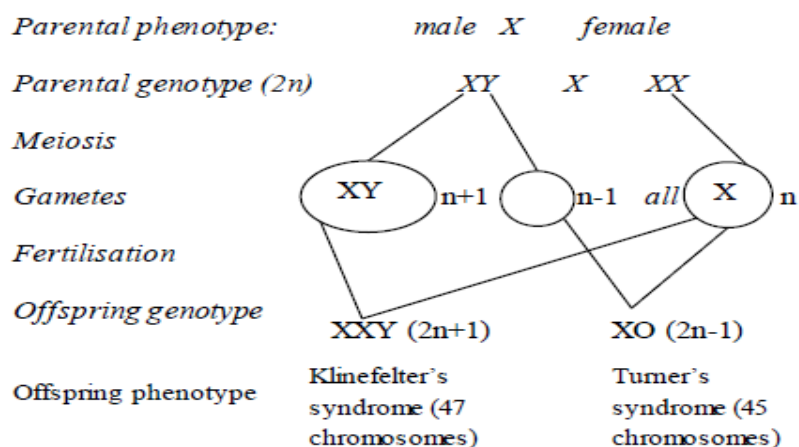
- They are mentally retarded
- They have a low resistance to infections and therefore have a short life span
- They have frequent saliva flow from the mouth
- They have slit eyed appearance.

2. HETEROSOME NON-DISFUNCTION

This is non-disjunction of sex chromosomes which produces a variety of aneuploid conditions which include the following;

a. Klinefelter's syndrome (XXY)

This results from non-disjunction of the sex chromosomes pair in either males or females resulting into a male with 47 chromosomes and genotype XXY. It may occur if a normal Y sperm fertilizes an abnormal egg. It may also occur if an abnormal sperm (XY) fertilizes a normal egg (X). This is illustrated below;



Individuals suffering from Klinefelter's syndrome have the following characteristics;

- They are infertile (sterile) and therefore they don't produce sperms although erection and ejaculation occurs.
- Their testes are usually very small compared to the normal males i.e. the testes fail to grow to the expected size.
- They have very subnormal intelligence
- They tend to be taller than the average height
- They have little facial hair
- They possess some female secondary sexual characteristics e.g. developed breasts and wide hips.
- Their trunk may show signs of obesity

b. Turner's syndrome (XO)

This monosomy condition occurs in females resulting into a female with 45 chromosomes and genotype XO. It occurs as a result of non-dysfunction which results into an abnormal egg or an abnormal sperm. If an abnormal sperm fertilizes a normal egg this condition occurs and if an abnormal egg that is empty is fertilized by a normal sperm containing X chromosomes this condition again occurs. Individuals with Turner's syndrome vary from others by having the following characteristics;

- They do not show female secondary characteristics such as developed breasts, menstruation, widening of hips e.t.c.
- They are infertile with no ovaries but with a small uterus.
- They have a short height than the women average height.
- They have a webbed neck.
- They are usually of normal intelligence

c. Triple X syndrome (XXX)

This also occurs in females due to non-disjunction which results into formation of an abnormal egg containing XX chromosomes and if such an egg is fertilized with a normal X sperm the triple X female occurs with 47 chromosomes and these individuals have the following characteristics;

- They are fertile females
- They are mentally normal
- They are physically normal
- They have a very high sex libido

d. XYY syndrome

This condition occurs in males with genotype XYY. It occurs in case the Y chromosome undergoes duplication and fails to separate at anaphase I. This may result into production of abnormal sperms containing YY which if they fertilize a normal X egg and XYY syndrome occurs. Individuals with this syndrome vary from other males by having the following characteristics;

- They are usually very aggressive and therefore common in prisons and security forces.
- They are fertile males.
- They are giants.

- They are mentally and physically normal

POLYPOIDY (EUPLOIDY)

This is a condition whereby cells of organism possess extra sets of chromosomes beyond the normal diploid number. Polyploidy therefore makes the genetic constitution of an organism multiplied to become $3n$, $4n$, $5n$, $6n$ e.t.c.

Polyplody is a useful phenomenon in plant breeding where the chromosome number is increased so as to improve on the vigor (characteristic) of the plant i.e. plants acquire better characteristics such as high yields, high resistance to diseases, quick maturity, high resistance to pests e.t.c. It is more common in plants than animals because the increased number of chromosomes in animal polyploidy causes errors in gamete formation unlike in plants which usually reproduce vegetatively without such errors.

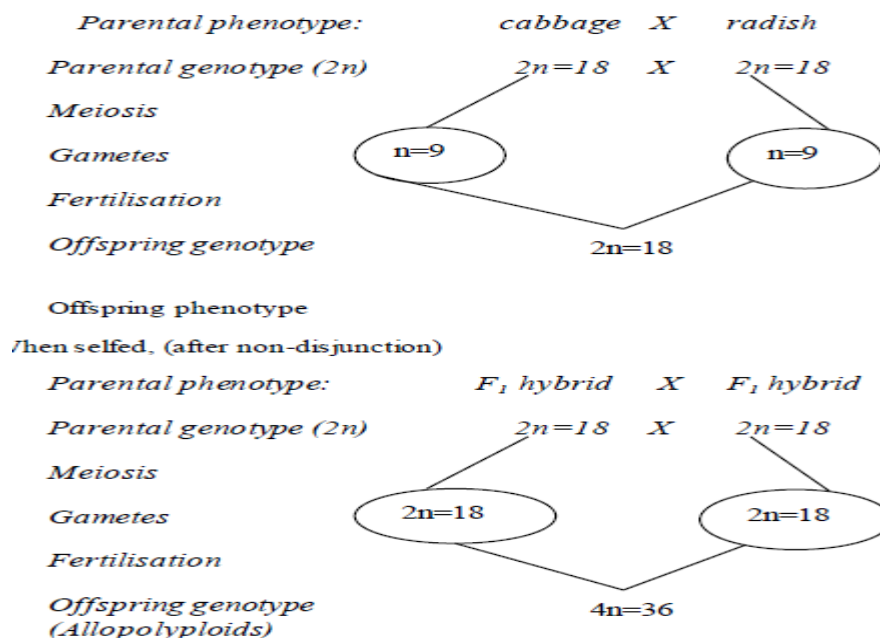
Polyplody brings about genetic variation within a population as it results into the formation of new and different genetic combinations within some individuals of the population. This makes some polyploids within the population better adapted than their original parents and so are favoured by a selection pressure of nature to survive, reproduce and transmit their adaptive variations to their offsprings. However, better adapted polyploids may fail to interbreed with diploid organisms, thereby forming a new species due to possession of extra sets of chromosomes beyond the diploid number.

Polyplody also results into formation of new species via **interspecific hybridisation**, a process in which the F1 hybrids formed are sterile because their chromosomes cannot form homologous pairs being that they arise from organisms of different species, but when a diploid number of chromosomes of F1 hybrids is doubled due to non-disjunction, tetraploids ($4n$) can be formed as F2 hybrids which can interbreed among themselves to produce fertile offsprings. The F2 tetraploids formed by interspecific hybridisation can interbreed among themselves to form fertile offsprings but cannot interbreed with any of the original parents because of having extra sets of chromosomes thereby becoming a new species.

The F2 offsprings formed are described as allopolyploids because they are formed by a type of polyploidy known as allopolyploidy. Allopolyploidy is the one which occurs when two different species interbreed and produce a sterile F1 hybrid whose chromosome number gets doubled by non-disjunction thereby changing the sterile F1 hybrids into fertile F2 hybrids. The allopolyploids formed are fertile with each other but cannot interbreed with a diploid parental species. The F1 hybrids are sterile because the set of chromosomes from one species cannot pair during meiosis with another set of chromosomes from another species.

Illustration

Parental phenotype: cabbage X radish



The tetraploids formed is fertile because homologous pairing of chromosomes can occur in meiosis, as the two sets of parental chromosome present in diploid gametes are produced which contain nine chromosomes from the parental cabbage and 9 chromosome of the parental radish. There are two forms of polyploidy namely; autopolyploidy and allopolyploidy

1. Autopolyploidy

This is where the chromosome number of some individuals in a given species is increased either naturally or artificially by preventing cytokinesis or preventing the formation of spindle fibres during cell division. This can be done artificially using colchicines which prevents formation of spindle fibers thereby increasing the chromosome number. This mutation prevents the tetraploids from successfully interbreeding with the diploid plants of the original population leading to the reproductive isolation. However, the tetraploids can still produce fertile offsprings by self-pollination or by mating with other tetraploids

Autopolyploid can be as fertile as diploids if they have an even number of chromosomes and they can be infertile if they have odd number of chromosomes because they cannot form homologous pairs.

Colchicines and other related drugs have been used in breeding in certain varieties of tobacco and tomatoes whose cells have a large nucleus.

2. Allopolyploidy

This is a condition which arises when the chromosome number in the sterile hybrids gets doubled and produces fertile hybrids. Sometimes the F₁ offsprings formed may be sterile but if these individuals are crossed with another related

During meiosis in F₁ hybrids chromosomes from each parent cannot pair together to form homologous chromosomes hence the F₁ hybrids produces gametes with a diploid set of chromosomes. This brings about allopolyploid as illustrated below;

The allopolyploid is fertile because homologous pairing of chromosomes can occur in meiosis as the two sets of parental chromosomes are present. Allopolyploid is an example of interspecific hybrid

GENE MUTATIONS (Point mutation)

Gene mutations are caused by changes to the sequence of nucleotides in DNA.

Addition, deletion and substitution which occur to the sequence of nitrogenous bases in DNA will affect the nature of proteins that are synthesized.

Mutated genes which cause defective proteins / enzymes to be synthesized may cause the death of the organism. Such genes are called lethal genes.

Examples of gene mutations in the human population

Sickle-cell anaemia

The haemoglobin gene undergoes a mutation to form a mutant recessive gene.

The mutant gene produces abnormal haemoglobin, causing the red blood cell to become sickle-shaped.

Sickle-shaped erythrocytes are not efficient in transporting oxygen and the patient suffers from anaemia.

A person who is homozygous recessive (possesses two mutant alleles) will suffer from severe life-threatening anaemia.

Albinism

A condition caused by a mutation to the gene which produces the pigments found in the skin, hair & eyes. Individuals with this condition are called albinos. Due to their lack of pigmentation, albinos are very fair-skinned, have white hair and red eyes.

The gene which causes albinism is recessive and is found on an autosome. Therefore, an individual needs to be homozygous .

Haemophilia

Haemophilia is an inherited blood disease.

The condition is caused by a gene mutation on the X-chromosome.

Haemophiliacs are unable to produce certain blood clotting factors.

ization i.e. form of sympatric speciation which occurs when a new species is produced by the crossing of individuals from two unrelated species.

Gene mutations (point mutation)

This is a sudden change in the sequence of nuclear nucleotides or bases of DNA. These mutations are of the following types;

1. Substitution. This type of mutation whereby one or more bases of nucleotides may be replaced with wrong nucleotides or bases. Sickle cell anemia in man is an example of a substitution gene mutation where an individual gets abnormal hemoglobin S in the red blood cells. Such individuals have a wrong better polypeptide chain in their hemoglobin due to the substitution

where by glutamic acid that causes the formation of normal hemoglobin is replaced by another amino acid called **valine** which caused formation of hemoglobin S in sickle cell anemia in individuals.

2. Deletion. This is a form of gene mutation where a section of DNA is lost. This results into wrong transcription and wrong translation processes in protein synthesis. This mutation is dangerous because it leads to absence of certain structures or wrong physiological processes taking place in an organism.

3. Inversion. This is where a group of nucleotides in DNA becomes reversed after rotating through 180 degrees.

4. Insertion. This is where one or more nucleotides may be fixed in a particular DNA strand.