GENETICS

Genetics is the scientific study of heredity and variation among organisms. It is the branch of science that accounts for the occurrence of similarities and differences among organisms of the same species; as well as explaining how traits are transmitted to off springs from their parents.

Inheritance refers to the process by which characters/traits are passed from parents to off springs while **variation** describes the difference in characteristics between organisms of the same species.

The importance of genetics

- It is applied in genetic engineering to produce better breeds and varieties of plants and animals by altering their genetic constitution.
- It is important in courts of law to determine the paternity of the child.
- Genetics forms the basis of blood transfusion to determine compatible blood groups.
- Genetic counseling is important in preventing transmission of genetically determined diseases among married couples. This will help to relieve the families and community of the costs on treatment as well as the suffering of the sick and their families.
- It can be used in identification of criminals by use of finger prints and DNA profiling.
- It is used in molecular biology to manufacture artificial enzymes, hormones and vaccines by manipulating responsive genes from organisms.
- Forms the basis of cloning to increase the number of genetically important plants and animals

DEFINITION OF TERMS USED IN GENETICS

Gene; A gene is the basic unit of inheritance that determines the organisms' characteristics. All the characteristic features of an organism are defined at fertilization by the genes inherited from parents but can be greatly modified by the environment in which the organism lives.

Alleles; Are alternative forms of the same gene.
Most of the genes occur in two alternative forms called alleles one of which is dominant and the other recessive both of which are represented by alphabetical letters just for study purposes.

Dominant gene/allele; Is a gene/allele whose trait is expressed phenotypically even in presence of a different allele. Such genes are always represented by capital letters when performing a genetic cross.

Recessive gene/allele; is a gene/allele whose character is not expressed phenotypically in presence of a different allele but is only expressed in a homozygous recessive state. Recessive genes are always represented by small

alphabetical letters in a genetic cross

- Consider the gene controlling height in garden peas, the allele **T** for tallness is dominant over the allele **t** for shortness. A plant with dominant genes **(TT)** is tall and the one with recessive genes **(tt)** is short while a plant with one of each genes **(Tt)** is also tall

Genotype; is the genetic makeup/constitution of an organism as inherited from the parents. It is determined at fertilization and does not depend on the environment. An organism with similar copies of alleles for a given gene is said to have a **homozygous genotype** e.g. TT, AA, rr etc. while an organism with different copies of alleles for a given gene is said to have a **heterozygous genotype.** E.g. Tt, Aa, Rr etc

Phenotype; Refers to the physical/outward appearance of an organism as determine by the interaction between its genotype and the environment in which it lives. A pea plant which is homozygous tall (represented as TT) but growing on nutrient-poor soils will become stunted and appear short. Such a plant is genotypically tall but the environment in which it grows modified it into a phenotypically short/dwarf plant.

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.**OR** is when the alleles found at a given locus are identical.

Heterozygous; this is a condition where an individual possess non-identical alleles for a particular gene e.g. Tt, Bb OR is when the alleles at a given locus are different

Pure breeding (true breeding), this is where the individuals being crossed are homozygous **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 hybrids

Second filial generation (F2); this refers to the set of off springs that are obtained from crossing mature F1hybrids.

Selfing: This refers to the crossing of offsprings of the same parents.
Test cross is a cross between an organism with an unknown genotype with a

homozygous recessive organism so as to determine the unknown genotype.
This is because phenotypically dominant organisms may ether be homozygous or heterozygous. In such a cross if all hybrids show the dominant trait then the unknown is homozygous dominant. A heterozygous individual will result into a mixture of hybrids in a ratio of 1:1 of dominant to recessive trait.

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 genotypes of the parents have been reversed

MENDEL'S GENETIC EXPERIMENTS AND MONOHYBRID INHERITANCE

Monohybrid inheritance refers to the inheritance of a single pair of contrasting characteristics. Examples include, inheritance of height, blood groups, albinism, sickle cell anaemia, and sex linked characteristics etc.
This mechanism of inheritance was discovered by an Austrian monk and biologist Gregor Johann Mendel who carried out a number of genetic experiments using the garden pea plants *(Pisum sativum);* which he grew in the vegetable garden in his monastery. He later observed many sexually reproducing organisms and found out that they had variations among themselves despite being of the same species.

Why Mendel used garden peas

- They occurred in many varieties with distinct characters.
- The plants were easy to cultivate.
- All their offsprings were fertile.
- They have a short life cycle that they reproduced so quickly.
- The plants also had many contrasting characters with no intermediates.
- Their reproductive structure were enclosed in petals which allowed for production of pure breeding plants due to self-pollination over many generations.

MENDEL'S EXPERIMENTS

In one of his experiments, Mendel crossed tall pea plants with dwarf pea plants. In order to properly manage the cross, Mendel covered the stigma of all flowers of one group, and removed all the anthers from the flowers of another group of pea plants in order to prevent self-pollination, and transferred pollen using a brush. The resultant seeds were planted and he observed that all the F_1 off springs were tall.

He then selfed the F_1 pea plants to get F_2. This generation comprised of a mixture of tall and short pea plants in a ratio of 3 tall: 1short plants.
NB: The **3:1** ratio is known as Mendel's monohybrid ratio of the dominant and recessive characters respectively in the F_2 generation.

Observation;

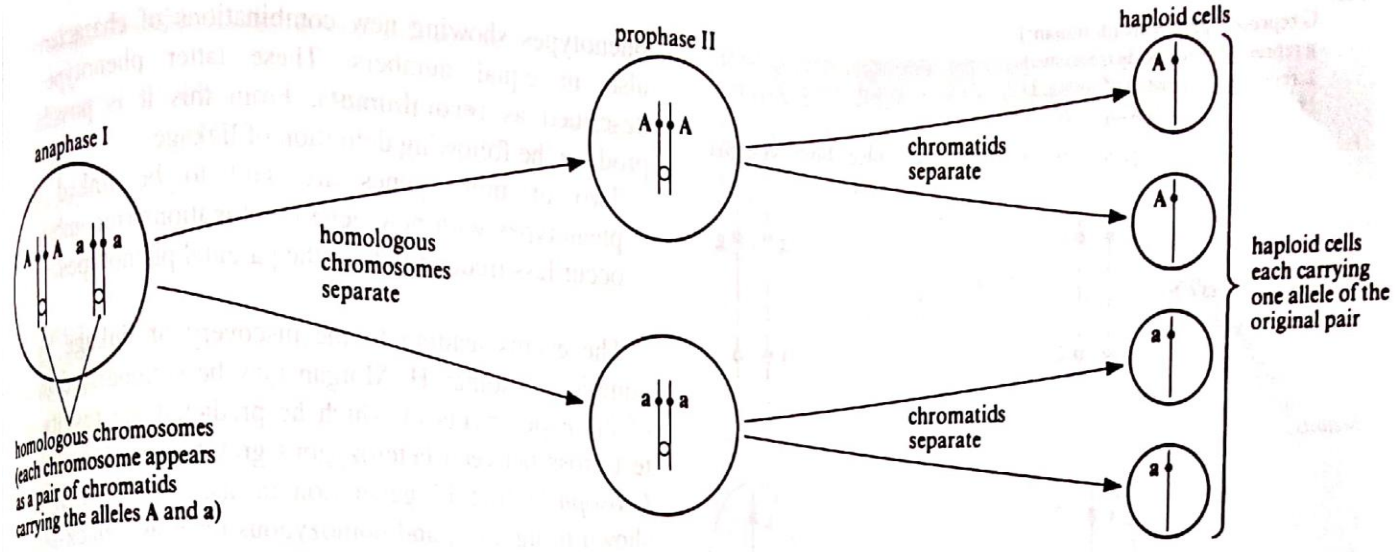
Mendel was able to observe that neither of the F_1 nor F_2 had intermediate phenotypes.

Conclusion;
He then concluded that inheritance is **not** the mixing/blending of features to produce intermediates but rather the process by which **internal factors** of the body **may or may not** express themselves in the phenotype.
From his conclusions, Mendel was able to formulate his first law of inheritance which is well known as the law of monohybrid inheritance/law of segregation/law of particulate inheritance

LAW1 states that ***"The characteristics of an organism are controlled by internal factors which occur in pairs but only one can be carried in a single gamete".***

Later with advancements in technology and microscopy, internal factors later came to be known as **genes** and Mendel's first law was modified. It can **modernly** be stated as follows. "The characteristics of a diploid organism are controlled by alleles which occur in pairs but singly in gametes".

Meiosis explains:
Mendel's first law can currently be explained/accounted for in terms of meiosis. The genes which determine organisms' characters usually occur in two alternative forms called alleles located on homologous chromosome. During anaphase 1 of meiosis, these homologous chromosomes separate (segregate) and move to opposite daughter nuclei. Subsequent cell division results into two gamete cells each containing one of the two alleles; therefore the alleles occur as pairs in body cells but singly in gamete cells.
Illustration:



A full genetic explanation of Mendel's first law and the 3:1 ratio

Let;

T represent the allele for tallness, t represents the allele for shortness

Parents : male x female

Phenotypes : Tall x short

Genotypes : TT x tt

Meiosis

Gametes : T T t t

Random fertilization :

F_1 genotypes : Tt Tt Tt Tt

Genotypic ratio : All Tt
Phenotypic ratio : All tall

To obtain F2 generation
F1 hybrids were selfed as shown below.

Let;

T represent the allele for tallness, t represents the allele for shortness

Parents : male x female

Phenotypes : Tall x tall

Genotypes : Tt x Tt

Meiosis

Gametes : T t T t

Random fertilization :

F_2 genotypes : TT Tt Tt tt

Genotypic ratios: 1TT : 2Tt : 1tt;
Phenotypic ratios: 3tall : 1 Short

Mendel carried out many other experiments on peas and other organisms and all gave consistent results as shown below:

NB: It became so obvious to predict which trait of a given pair is dominant over the other. In a cross starting with pure breeding parental stocks, all the F_1 hybrids show the dominant trait. In addition, a larger proportion of the F_2 hybrids show the dominant trait while those showing the recessive one are always fewer

WORKED EXAMPLES

1. In a garden pea plant there are two forms of heights. When a pure breeding tall pea plant was crossed with a short pea plant all the offsprings obtained where tall when the offsprings were selfed a phenotype ration was obtained in F_2.
   a. Using suitable genetic symbols, workout the genotypes and phenotypes of the F_2 generation
   b. What are the phenotypic and genotypic ratios of the F_2 generation
   c. Explain how you would determine the genotype of F_1 tall pea plants formed
   d. Suppose 700 pea plants where produced in the F_2 generation
      i. How many were tall?
      ii. How many were short?

2. Suppose a man who is a tongue roller marries a woman who is a non-tongue roller and all the children obtained in F_1 are tongue rollers.
   (a) Work out the phenotypic and genotypic ratio as obtained in F_2 generation.
   (b) What is the probability that the 4^{th} born is a non-tongue roller?

Note; for any genetic cross:
- Appropriate letters are 'let' to represent respective alleles involved
- A cross**(X)** must be indicated to symbolize mating between the parents
- Directive words must be indicated to define each step of the cross
- In case of identical gametes, only one can be indicated

Solutions (a)

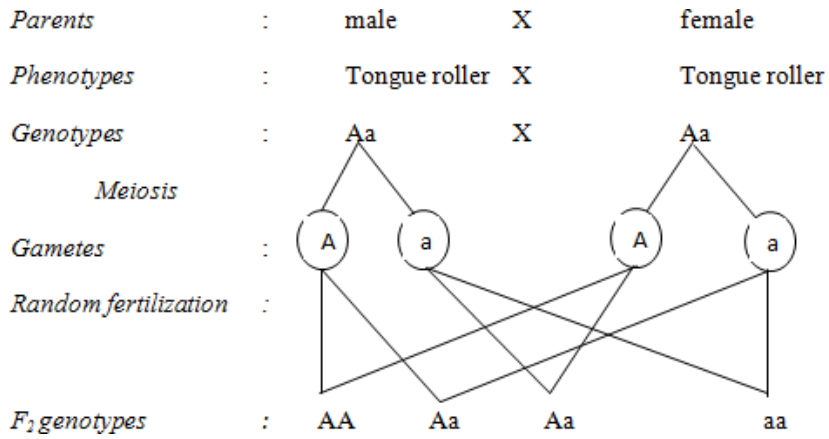
F_1 phenotypes: all tongue rollers

Let;

A represent the allele for tongue rolling, t represent the allele for non-tongue rolling

Parents	:	Male	X	Female
Phenotypes	:	Tongue roller	X	Non-tongue roller
Genotypes	:	AA		aa
Meiosis				
Gametes	:	all A		all a
Random fertilization	:			
F_1 genotypes	:	all Aa		

By selfing the F1 hybrids to obtain F2



Genotypic ratios: 1AA: 2Aa: 1aa;
phenotypic ratios: 3 tongue rollers: 1non-roller

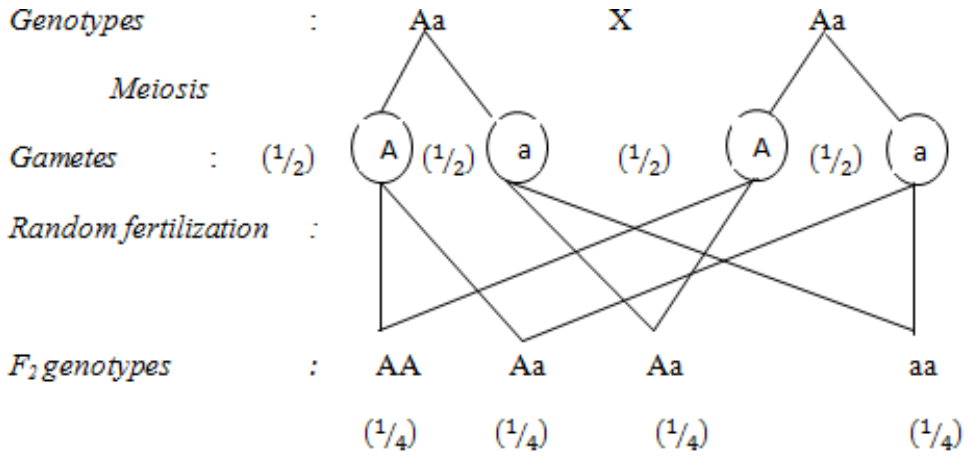
(c) Probability that the 4th born is a non-tongue roller

$= {}^{1}/_{4} \times {}^{1}/_{4} \times {}^{1}/_{4} \times {}^{1}/_{4}$
$= {}^{1}/_{256}$

NB:

- The such crosses can be performed in terms of probability as follows

Each gamete carrying any of the two alleles has 0.5 chance of fusing with the gamete from the other parent,



F_2 phenotypic ratios: $({}^{3}/_{4})$ rollers: $({}^{1}/_{4})$ non-rollers = 3rollers:1non roller

In case of individuals showing the dominant trait, the genotype may either be

homozygous dominant or heterozygous. Such genotypes can be determined by performing **a test cross;** that is, crossing the unknown with a homozygous recessive individual. If the unknown is homozygous, the resultant hybrids will all show the dominant trait but otherwise, a mixture of dominant and recessive traits are produced in a ratio of 1:1

In a test cross, a homozygous dominant individual cannot be used because in such a case; regardless of the unknown genotype, all the resultant hybrids would show the dominant trait

EXAMPLES OF MONOHYBRID INHERITANCE IN MAN

There are many genetically determined abnormalities and diseases that affect man (and other animals). Since these are genetic diseases, they can only be inherited from parents and their occurrence is determined by those genes inherited from parents during fertilization

Examples of such diseases include:

- Sickle-cell anaemia
- Albinism
- Achondroplasia
- Cystic fibrosis and many more

NB: Research has showed that most of, though not all the genetic abnormalities are caused by **recessive genes (alleles)** and the genes responsible for normal conditions are dominant. This implies that for an individual to suffer from such diseases, they must have two copies of the responsive genes (homozygous recessive). The heterozygotes and the homozygous dominant individuals are normal. Though the former are phenotypically normal but their cells contain a copy of the recessive allele and are described as carriers

INHERITANCE OF SICKLE-CELL ANAEMIA

Sickle-cell anaemia is a recessive character caused by a point substitution mutation in which glutamic acid in normal haemoglobin is replaced by valine. Normal haemoglobin **(HbA)** contains an amino acid glutamic acid at position 6 of the ▪-chain. The amino acid is **polar and hydrophilic** which make normal haemoglobin soluble in water. It is coded for by the DNA triplet CTT and its complementary mRNA codon is GAA.

A substitution mutation leads to replacement of T with A making the DNA triplet CAT and its complementary mRNA codon GUA. This tiplet codes for valine which is non-polar and hydrophobic hence reduces the solubility of haemoglobin especially at low oxygen tensions. This abnormal haemoglobin crystallizes into rigid rod-like fibres which distort the normal biconcave shape of RBCs into a crescent/sickle shape. Such abnormal haemoglobin is called **HbS,** It has a very low oxygen- carrying capacity leading to symptoms of anaemia and the disease

is known as **sickle-cell anaemia.**

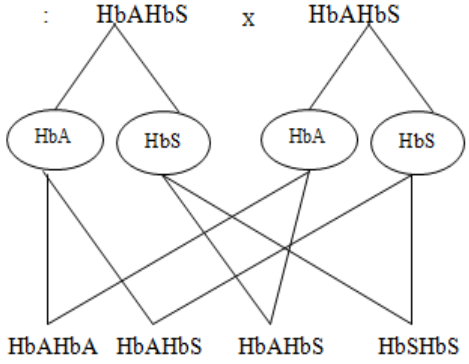
Being a recessive character, for a person to be a sufferer they must possess two copies of the faulty gene (homozygous recessive, i.e. HbSHbS or ss). Heterozygotes (carriers, i.e. HbAHbS or Ss) have one copy of the responsive gene whose effects are masked by the other dominant gene. They don't suffer from the disease symptoms except at exceptionally low oxygen tensions; this is known as **sickle-cell trait**.
It is therefore advisable to avoid exposure of such people to low oxygen environments like crowded places, high altitudes and flying in unpressurised aircrafts.

Question; if two people suffering from sickle cell trait are married, what is the probability that they will produce an anaemic child?

Solution

Let *HbA represent the allele for normal haemoglobin; HbS represent the allele for abnormal haemoglobin*

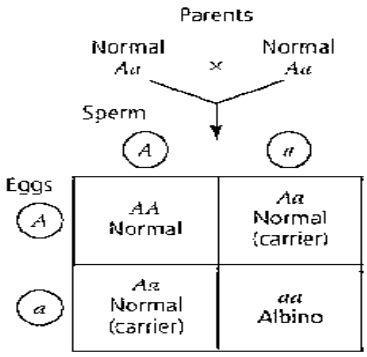
Parents	:	Man	x	Woman
Phenotypes	:	Normal	x	Normal
Genotypes	:	HbAHbS	x	HbAHbS



Genotypic ratios: 1HbAHbA:2HbAHbS:1HbSHbS
Phenotypic ratios: 1normal:2 carriers: 1sickler
Probability of a sickler is $^{1}/_{4}$ = 0.25

Complications due to sickle cell anaemia

1. Anaemia occurs because the sickle cells are destroyed which lowers the amount of oxygen to be carried leading to acute anaemia. This leads to;
    - Fatigue (weakness)
    - Poor physical development
    - Dilation of the heart which may lead to heart failure
2. Interference with circulation of blood because sickle cells get jammed in tiny



capillaries and small arteries. This leads to;

- Heart damage which leads to heart failure
- Lung damage which leads to pneumonia
- Kidney damage which leads to kidney failure
- Liver damage

3. 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.

NB: Despite the above complications suffered by sufferers of sickle cell anaemia, the heterozygotes tend to have an advantage of showing increased resistance to the plasmodium parasite that causes malaria much more than both the sufferers and the normal. This resistance is as a result of two factors:

- The consistent change in oxygen levels between normal and sickle cells makes it difficult for the parasite to adapt. In such cases, the immune system of the body eliminates the parasites before the disease is established rendering resistance to the heterozygotes

This is referred to as the **heterozygous advantage** which increases chances of survival for heterozygotes especially in the tropics where malaria is one of the leading causes of death

INHERITANCE OF ALBINISM

Albinism is a recessive character which results into failure of formation of body pigments.

Albinos have the following characteristics as a result;

- Light-coloured skin
- White hair
- Pink eyes

Qns.

Man with normal skin marries a carrier for albino skin. (i) What is the probability that some of their children will be albinos? (ii) What is the probability that the sister with normal colour is a carrier?

(i) Probability = $^1/_4$; because only one is an albino

(ii) Probability = $^2/_4 = {}^1/_2$; because two are carriers.

INHERITANCE OF CYSTIC FIBROSIS

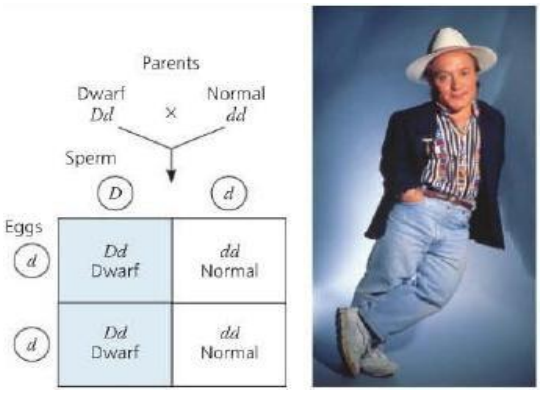
This is a recessive character caused by a mutation resulting into accumulation of abnormally **thick and sticky mucus** that blocks the pancreatic duct, bile duct and air passages.

The mutation occurs on an autosomal chromosome 7 affecting the gene that codes for **a chloride channel protein** in epithelial cells. This results into total absence or malfunctioning of this channel protein hence interfering with chloride ion flow. Chloride ions accumulate in the cells and attract sodium ions towards the opposite charge; this increases the ion concentration, hence osmotic potential of the cells which prevents **osmotic outflow** of water. As a result, the mucus secreted is dry, thick and sticky; blocking small tracts of some body organs. This is known as cystic fibrosis.

In the pancreas, fibrous patches called **cysts** develop (hence the name) and complications include digestive problems due to poor release of pancreatic enzymes, poor absorption of digestive products, chronic lung diseases, reduced fertility etc.

ACHONDROPLASIA (DWARFISM)

Although many harmful alleles are recessive, a number of human disorders are due to dominant alleles. One example is *achondroplasia,* a form of dwarfism that occurs in one of every 25,000 people in the world. Heterozygous individuals therefore have the dwarf phenotype as shown below.



Since this character is dominant (caused by a dominant allele), all people who are not achondroplastic -99.99% of the population are homozygous for the recessive allele. Like the presence of extra fingers or toes mentioned earlier, achondroplasia is a trait for which the recessive allele is much more prevalent than the corresponding dominant allele.

DIHYBRID INHERITANCE AND MENDEL'S SECOND LAW OF INHERITANCE

Dihybrid inheritance refers to the inheritance of two pairs of contrasting characteristics simultaneously.

For instance, in one of his experiments; Mendel crossed pure breeding **tall** pea plants with **red** flowers with pure breeding **dwarf** plants having **white** flowers. All in the F_1 progeny were **tall with red flowers**. This showed just like Mendel had discovered before that the alleles for tallness and red flowers were dominant to those for dwarfness and white flowers respectively.

Mendel went ahead to self-pollinate the F_1 plants and obtained an F_2 progeny, this comprised of a variety of phenotypes as summerised in the table below.

- 315 Tall with red flowers
- 101 Tall with white flowers
- 108 Dwarf with red flowers
- 32 Dwarf with white flowers

These give the respective phenotypic ratios as 9:3:3:1. This is known as **Mendel's Dihybrid ratio**; the ratio of phenotypes in the F_2 generation for a Dihybrid cross.

From this and many other similar crosses, Mendel was able to make the following observations:

- Both phenotypes/characters (height and flower colour) combined in the F_1 but separated and behaved independently in the F_2.
- Two of the F_2 phenotypes resembled one or the other of the parental phenotypes WHILE two new combinations of phenotypes appeared in the F_2; (Tall/white and Dwarf/red). These are known as recombinants.
- The allelomorphic pairs of characteristics (controlled b different alleles of the same gene) occurred in a phenotypic ratio of 3 dominant: 1 recessive.
    - E.g. 3tall: 1 dwarf and 3red: 1 white.

Basing on these observations, Mendel formulated his second law known as the law of independent assortment.

The law states that;

> **"**Any **one of a single pair of characters may combine randomly with either one from another pair"**

Below is a full genetic explanation of the 9:3:3:1 ratio of phenotypes in the F_2 generatioon of a dihybrid cross.

Let; *T represent allele for tallness, t for dwarfness*

R represent allele for red flowers, r for white flowers

Parents	:	Male	X	Female
Phenotypes	:	Tall, red flowered	X	Dwarf, white flowered
Genotypes	:	TTRR	X	ttrr
Meiosis	:	↓		↓
Gametes	:	all (TR)		all (tr)
Random fertilization	:	↘ ↙		
F_1 genotypes	:	All TtRr		

Phenotypic ratios : All Tall with red flowers.

By selfing F1 plants;

Parents	:	Male	X	Female
Phenotypes	:	Tall, red flowered	X	Tall, red flowered
Genotypes	:	TtRr	X	TtRr
Meiosis	:			
Gametes	:	(TR) (Tr) (tR) (tr)		(TR) (Tr) (tR) (tr)

Random fertilization

	TR	Tr	tR	tr
TR	TTRR	TTRr	TtRR	TtRr
Tr	TTRr	TTrr	TtRr	Ttrr
tR	TtRR	TtRr	ttRR	ttRr
tr	TtRr	Ttrr	ttRr	ttrr

F2 offspring phenotypes: 9 tall – red flowered plant
3 tall – white flowered plant
3 short – red flowered plant
1 short – white flowered plant

The F2 phenotypic ratio is 9: 3: 3: 1

NB: When performing a dihybrid cross;

- Alleles of the same gene cannot pass into the same gamete (they segregate during meiosis). I.e. T can only be present with Y or y but not t while t can only be present withy or y but not T as in the above case

- The possible combination of gametes during fertilization is shown in a Punnett square (after the Cambridge geneticist R. C. Punnett). This minimizes errors when listing the combinations.

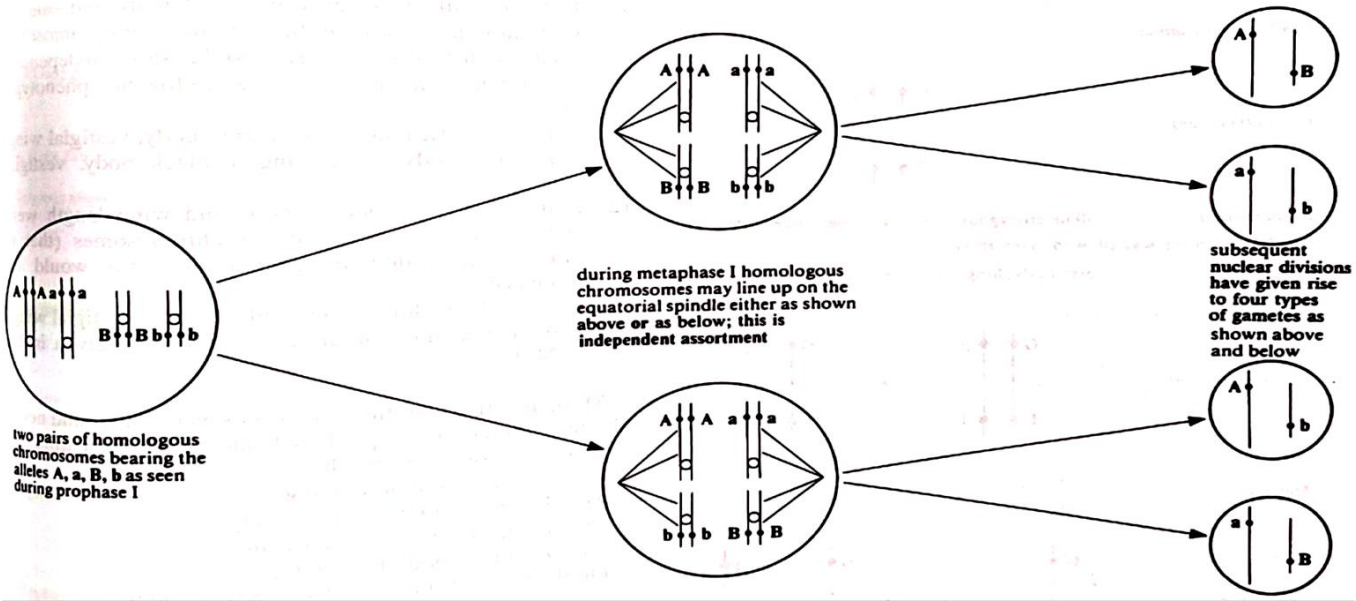
In summary; the following can be noted from Mendel's hypotheses:

- Each characteristic of an organism is controlled by a pair of alleles.
- During meiosis, each pair of alleles segregate (separates) and each gamete receives one of each pair. This is known as the law of segregation.
- During gamete formation, either one of a pair of alleles can pass into the same gamete with either one from another pair. This is known as the law of independent assortment.
- Each allele is transmitted one generation to the next as a discrete unit
- Each diploid organism inherits one allele for each character from each of the two parents.
- If an organism has two unlike alleles for a given gene, one may be expressed (dominant) at total exclusion of the other (recessive).

MEIOSIS EXPLAINS:

Mendel's second law can be explained/accounted for on the chromosomal basis by meiosis.

During formation of gametes by meiosis, the distribution of each allele from a single pair is entirely independent of alleles from other pairs. This in turn depends on the random orientation of homologous chromosomes onto the equatorial spindle in metaphase I. Subsequent separation during anaphase I leads to a variety of allele combinations in gametes. In this process; any one of a single pair of alleles can combine randomly with either one form another pair. **Illustrations**:



NB: For the haploid number of chromosomes = n, the total number of possible combinations in gametes is given by 2^n

WORKED EXAMPLES:

1. 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.

a) Using suitable genetic symbols work out the phenotypes and genotypes that were obtained in F2 generation.

b) Suppose 480 flies were obtained in F2 work out the numbers of the flies for each phenotype class.

c) How many of these flies were recombinants.

Solutions:

Let; *B represent allele for broad abdomen, b for narrow abdomen*

L represent allele for long wings, l for vestigial wings

Parents	:	Male	X	Female
Phenotypes	:	Broad abdomen,		Narrow abdomen,
		Long wings	X	vestigial wings
Genotypes	:	BBLL	X	bbll
Meiosis	:	↓		↓
Gametes	:	all (BL)		all (bl)
Random fertilization	:			
F_1 genotypes	:	All BbLl		

Obtaining F2

Parents	:	Male	X	Female
Phenotypes	:	Broad abdomen		Broad abdomen
		Long winged	X	vestigial winged
Genotypes	:	BbLl	X	BbLl
Meiosis	:			
Gametes	:	(BL) (Bl) (bL) (bl)		(BL) (Bl) (bL) (bl)

Random fertilization

	BL	Bl	bL	bl
BL	BBLL	BBLl	BbLL	BbLl
Bl	BBLl	BBll	BbLl	Bbll
bL	BbLL	BbLl	bbLL	bbLl
bl	BbLl	Bbll	bbLl	bbll

(b) Phenotypic ratios = 9:3:3:1,
Total ratio = (9+3+3+1) = 16
Number of flies = $\frac{Ratio\ in\ the\ phenotype}{Total\ ratio} \times 480\ Flies$
Broab abdomen and long winged $= {}^{9}/_{16} \times 480 = 270$ flies
Broad abdomen and vestigial winged$= {}^{3}/_{16} \times 480 = 90$flies
Narrow abdomen and long winged$= {}^{3}/_{16} \times 480 = 90$ flies
Narrow abdomen and vestigial winged $= {}^{1}/_{16} \times 480 = 30$ flies

(c) Number of recombinants = (90 + 90) flies = 180 flies

SAMPLE QUESTIONS:

1) In guinea pigs, there are two alleles for hair colour and two for hair length. In a breeding experiment, all the F1phenotypes produced from a cross between pure breeding short black-haired and long white-haired parents had short black hair. Explain
   a) Which alleles are dominant
   b) The expected F2 phenotypes

2) Flower in sweet pea plants is determined by two allelomorphic pairs of genes (R, r and S, s). Presence of at least one dominant gene from each pair makes the flowers purple while all other genotypes are purple. If two plants heterozygous for both genes are crossed, what will be the phenotypic ratio of the offsprings **(9:7)**

EXCEPTIONS TO MENDEL'S LAWS

It should however be noted with concern that Mendel's laws of inheritance are not of universal application to all processes of inheritance in organisms, For the work that led to his two laws of inheritance, Mendel chose pea plant characters that turn out to have a relatively simple genetic basis: Each character is determined by one gene, for which there are only two alleles, one completely dominant and the other completely recessive.

But these conditions are not met by all heritable characters, and the relationship between genotype and phenotype is rarely so simple. In this section, we will extend Mendelian genetics to hereditary patterns that were not reported by Mendel. These are referred to as exceptions to Mendel's laws of inheritance because they never produce the 3:1 or the 9:3:3:1 ratios of phenotypes in monohybrid and dihybrid crosses respectively.

LINKAGE

This is the condition when two or more genes are carried on the same chromosome
Such genes form a linkage group and pass into the same gamete during meiosis and are therefore inherited together. As a result, these genes do not show independent assortment (applies to genes on non-homologous chromosomes) and fail to produce the 9:3:3:1 ratio.
Linked characteristics (traits) are characters controlled by genes found on the same chromosomes and therefore inherited together.

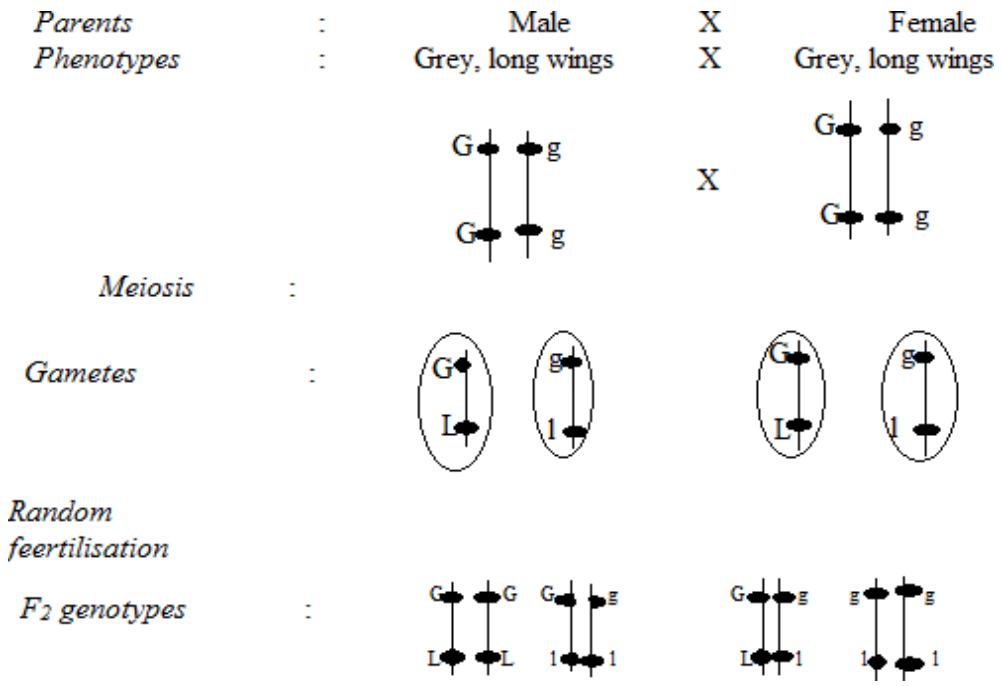
In drosophila, the alleles for grey body and long wings are dominant to those for black body and vestigial wings respectively. If pure breeding grey bodied long winged drosophilae are crossed with pure breeding black bodied vestigial winged drosophila; all in the F1 are grey with long wings. Surprisingly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

Let: G represent allele for grey body, g for black body

L represent allele for long wings, l for vestigial wings

Parents	:	Male	X	Female
Genotype	:	Grey, long winged	X	Black, vestigial winged
		G G L L	X	g g l l
Meiosis	:			
Gametes	:	*all* G L		*all* g l
Random fertilisation	:			
F1 genotypes	:	G g L l		

F1 phenotypes: all grey with long wings
Obtaining F2 generation:

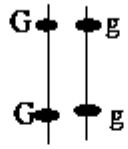
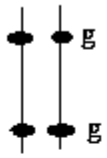
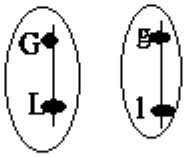
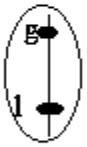
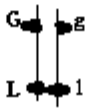
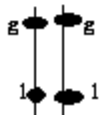


Phenotypic ratio: 3 grey long winged: 1 black vestigial winged.

Surprisingly, the 3:1 ratio of parental phenotypes is never obtained in practice. This is because total linkage is rare. Instead approximately equal numbers of parental phenotypes are obtained with significantly few recombinant phenotypes also in approximately equal numbers.

Definition: Two or more genes are said to be **linked** if recombinant phenotypes occur much less frequently than parental phenotypes.
Total/complete linkage is when the distance between linked genes is not sufficient to allow for successful crossing over.

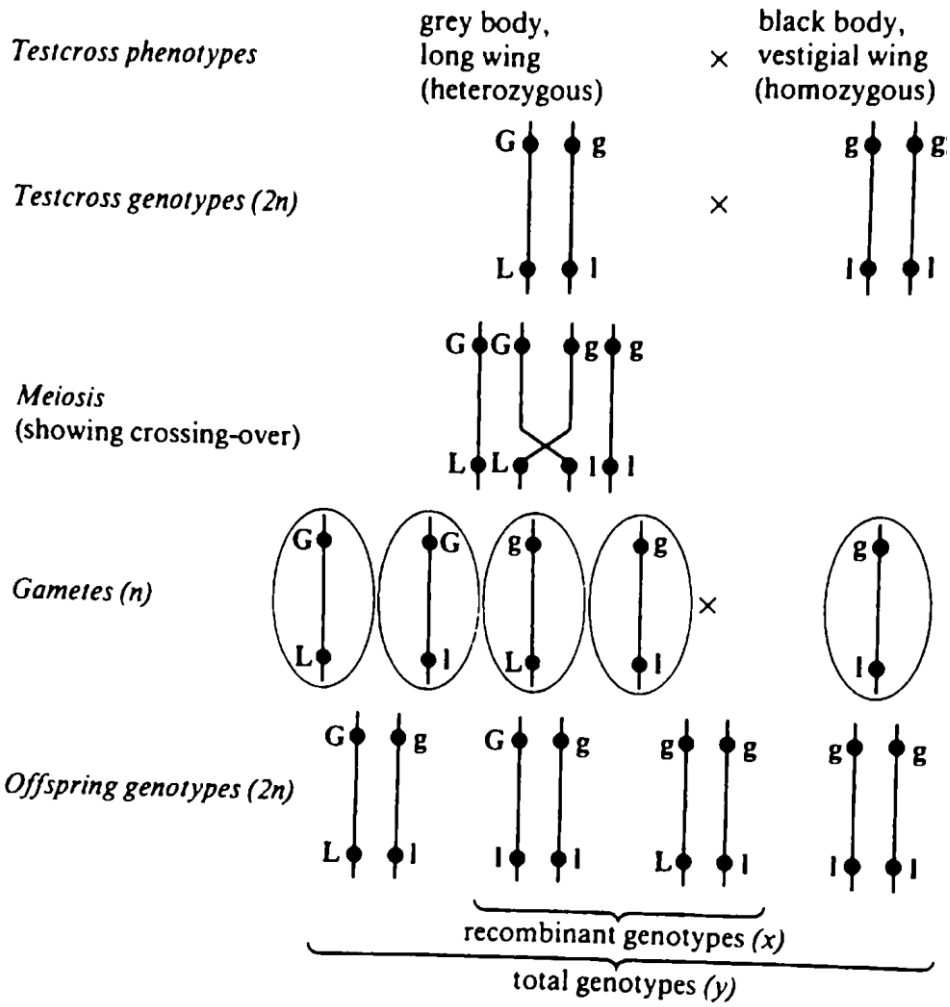
These results were explained by an *American scientist Thomas H. Morgan*. In a cross between a grey, long winged drosophila heterozygous for both traits with a black, vestigial winged drosophila (This is a test cross); Morgan predicted that in the normal Mendelian inheritance. Parental; phenotypes and recombinants would be obtained in a ratio of 1:1:1:1. If genes were completely linked, parental phenotypes would be obtained in a ratio of 1:1 **as shown below**.

Parents	:	Male	X	Female
Phenotypes	:	Grey, long wings	X	Grey, long wings
			X	
Meiosis	:			
Gametes	:			
Random feertilisation				
F_2 genotypes	:			

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

41.5% grey, long winged
41.5% black, vestigial winged
8.5% grey, vestigial winged
8.5% black, long winged

Morgan explained his results in terms of **crossing over**; the responsive genes are located on the same chromosomes (linked) with the alleles of each gene on homologous chromosomes. Alleles were exchanged between homologous chromosomes during meiosis, leading to new gene combinations in gametes hence producing recombinant phenotypes; as shown below.



F2 offspring phenotypic ratio is: 1 grey body long wings : 1 grey body short wings : 1 black body long wings : 1 black body short wings

Sample question:

A homozygous purple-flowered short stemmed plant was crossed with a homozygous red-flowered long stemmed plant and all the F1plants had purple flowers and short stems. When the F1generation was taken through a test cross, the following progeny was produced

53 purple flowered short stemmed 47 purple flowered long stemmed 49Red flowered short stemmed 45 red flowered long stems. Explain the results fully.

CROSSING OVER AND CROSS OVER VALUES

During crossing over, the frequency of crossovers which take place was found to be dependent on the distribution and arrangement of chromosomes. This is given by the **cross over value/frequency aka recombination frequency**.

This is calculated as a percentage ration of recombinants to the total number of offsprings.

$$\boldsymbol{COV = \frac{Number\ of\ recombinants}{Total\ numbe\ of\ offsprings} \times 100}$$

Example
In a test cross carried out on a grey long winged drosophila, the following results were obtained

Phenotype	**Number of offsprings**
Grey, long winged	965
Black, vestigial winged	944
Black, long winged	206
Grey, vestigial winged	185

Solution:

$$\boldsymbol{COV = \frac{number\ of\ recombinants}{total\ numbe\ of\ offsprings} \times 100}$$

$$= \frac{\mathbf{206 + 185}}{\mathbf{965 + 944 + 206 + 185}} \times \mathbf{100}$$

$$= 17\%$$

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

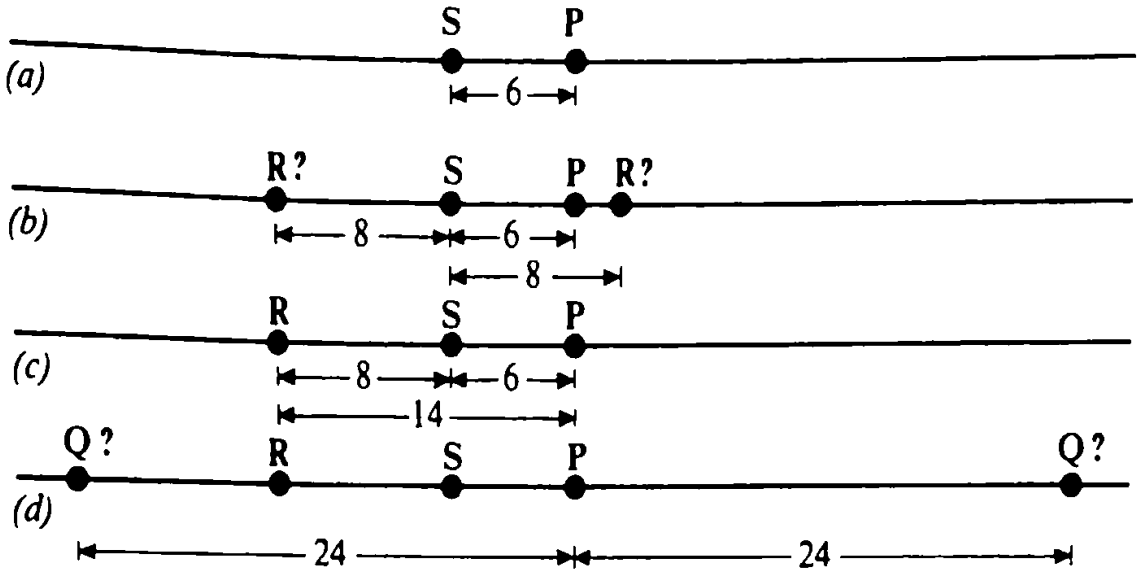
Consider the cross over values involving for different genes P, Q, R and S. The distance separating these four genes is shown below;
P-Q = 24% R-P = 14% R-S = 8% S-P = 6%
Draw the chromosome map to show the position of these chromosomes.

Answer.
Draw the chromosome map for these genes

a. **Insert the positions of the genes with the smallest cross over value first inthe middle of the chromosome map**
b. **Examine the next largest cross over value and insert both possible positionsof its genes on the chromosomes relative to either S or P.**

c. **Repeat the procedure for the entire remaining cross over values until youreach the largest cross over values.**

Illustration



Example
In maize, the genes for coloured seed and full seed are dominant to the genes for colourless and shrunken seed. Pure breeding strains of double dominant variety were crossed with a double recessive variety and a test cross of the f1 generation produced the following results

Coloured full	380
Colourless shrunken	396
Coloured shrunken	14
Colourless full	10

Calculate the distance between the genes for coloured seed and seed shape

DEGREES OF DOMINANCE
In the conventional Mendelian inheritance, each trait is controlled by a pair of alleles located at the same locus of homologous chromosomes, one dominant and the other recessive. In such cases, offsprings always resemble **one or the other of the parents** and phenotypes of the heterozygote and the dominant homozygote are indistinguishable. This condition is called **complete dominance.**

Some traits however are controlled by alleles neither of which shows complete dominance or recessiveness over the other. Such alleles are either equally

dominant (codominant) or incompletely dominant.

INCOMPLETE DOMINANCE

This is when alleles fail to show complete dominance or recessiveness such that their phenotypes **blend (mix) to produce an intermediate** in the heterozygote.

When red snapdragons are crossed with white snapdragon plants (*Antirrhinum);* all the F1 hybrids have pink flowers, while F2 hybrids produced 1 red: 2pink:1white plants as shown below

Note:
Given that both alleles of the same gene are dominant, we let a single letter for the gene and alleles attached as superscripts. I.e. C^R and C^W or simply R and W represent alleles for red and white petals respectively. The third phenotype results from flowers of the heterozygotes (C^R C^W or simply RW) having less red pigment than the red homozygotes.

Let;

R represent the allele for red petals, W represents the allele for white petals

Parents	:	male	x	female
Phenotypes	:	Red petals	x	White petals
Genotypes	:	RR	x	WW
Meiosis				
Gametes		R R		W W
Random fertilization	:			
F_2 genotypes	:	RW RW		RW RW

F1 phenotypes : All pink Obtaining F2

Genotypic ratio: 1RR: 2RW: 1WW
Phenotypic ratio: 1 Red: 2pink: 1 White

Other examples of incomplete dominance include:

Characteristic	**Allelomorphic characteristics**	**Heterozygous phenotype**
Mirabilis Japalla	Red and White	Pink

(4-oclock flower)		
Angora rabbit hair length	Long and short	Intermediate
Plumage colour in Andalusian fowls	Black and splashed white	Blue

CODOMINANCE

This is when alleles fail to show complete dominance or recessiveness such that their phenotypes are independently present in the heterozygote.

During the inheritance of fur/coat colour in short-horned cattle, when red and white cattle are mated, the F1 hybrid has white fur thickly interspersed with red fur. This phenotype is referred to as roan

Let;

R represent allele for red fur, W represents allele for white fur

Parents	:	Bull	X	Cow
Phenotypes	:	Red fur	X	White fur
Genotypes	:	RR	X	WW
Meiosis	:			
Gametes	:	All R		All W
Random fertilization	:			
F_1 genotypes	:	All		RW

F2 phenotypes : All roan calfs

Other examples of codominance include ABO blood groups and Sickle-cell trait

MULTIPLE ALLELES

These are three or more forms of the same gene occurring at the same locus

Most genes are known to occur in two alternative forms (allelic forms) located on the same locus of homologous chromosomes. Some genes are known to occur in more than two allelic forms called **multiple alleles** of which any two can occupy the gene locus in a diploid organism. This is easily noticed for the gene responsible for blood groups in man.

Inheritance of blood groups

Blood group I controlled by autosomal genes. The gene locus represented by the letter **I** (which stands for **isohaemagglutinogen**)

The gene for human blood group is known to occur in three allelomorphic

forms; **A, B** and **o**. Alleles A and B are codominant while o is recessive to both. This is known as the **ABO blood grouping system**, with three alleles producing six possible genotypes and four phenotypes.

Blood group	Possible genotypes
A	Ao/$\mathbf{I^AI^o}$, AA/ $\mathbf{I^AI^o}$
B	Bo/ $\mathbf{I^BI^o}$, BB/ $\mathbf{I^BI^B}$
AB	AB/ $\mathbf{I^AI^B}$
O	oo/ $\mathbf{I^oI^o}$

Sample question:
The father and a mother are known to be heterozygous for blood groups A and B. Show the possible genotypes of their children. If they bear non-identical twins, what is the probability that both twins are of blood group A.

Solution

Let;

A, B and o represent the alleles for blood groups A, B and O respectively

Parents : Father x Mother

Phenotypes : Blood group A x Blood group B

Genotypes : Ao x Bo

Meiosis

Gametes : A, o, B, o

Random fertilization :

Offspring genotypes : AB, Ao, Bo, oo

Offspring phenotypes: Blood groups AB, A, B and O
Probability for a child with blood group A = ¼
Probability for both twins with blood group A = ¼ × ¼ =1/16 = 0.0625

Example:

Work out the possible blood groups of the offsprings produced if a man of blood group A marries a woman of blood group AB

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^+) while those without are called rhesus negative (Rh^+). The allele for Rh^+ allele is dominant over the one for rhesus negative (Rh^-).

If a Rh^+ man marries a Rh^- woman, most of their children are likely to die immediately after birth or before birth because the mother's immune system produces antibodies (anti-D agglutinins) which pass into the foetus and cause death. 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.

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.

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.

LETHAL GENES

Genes are usually known to control a single pair of contrasting traits. Some genes may affect more than one characteristics including mortality. Such genes are responsible for some features necessary for survival but they are simultaneously responsible for lethal effects in the organisms and are therefore called lethal genes.

An example is clearly illustrated in the inheritance of **fur colour in mice**. Wild mice are known to have grey coloured fur (a condition called agouti) or yellow fur. A cross between two yellow mice produces yellow and agouti offsprings in a ratio of **2: 1** respectively.

These results can be explained by the fact that allele for yellow fur is dominant over that for agouti and all living yellow mice are heterozygous for fur colour. The 2:1 ratio of phenotypes is due to the death of the yellow mice that are homozygous for fur colour before birth. This allele is therefore lethal in the homozygous condition.

Let:
Y represent yellow fur (dominant)
y represent agouti fur (recessive)

Parental phenotypes	yellow fur	×	yellow fur
Parental genotypes (2n)	Yy	×	Yy
Meiosis			
Gametes (n)	(Y) (y)	×	(Y) (y)
Random fertilisation			
Offspring genotypes (2n)	YY Yy Yy		yy
Offspring phenotypes	1 yellow fur: 2 yellow fur : 1 agouti fur die before birth		

The homozygous dominant mice die before birth, producing a genotypic ratio of 2: 1 as the phenotypic ratio.

Examination of the uteri of yellow mice pregnant of yellow males revealed dead yellow mice; which are not revealed in yellow mice pregnant of agouti males.

Note:

- The allele for yellow fur is dominant for fur colour but recessive for mortality. It can therefore persist within the population over generations in heterozygous genotypes without phenotypic exposure to environmental elimination.
- Dominant lethal genes are very rare in a population because they are phenotypically expressed for elimination by environment

THE GENE COMPLEX

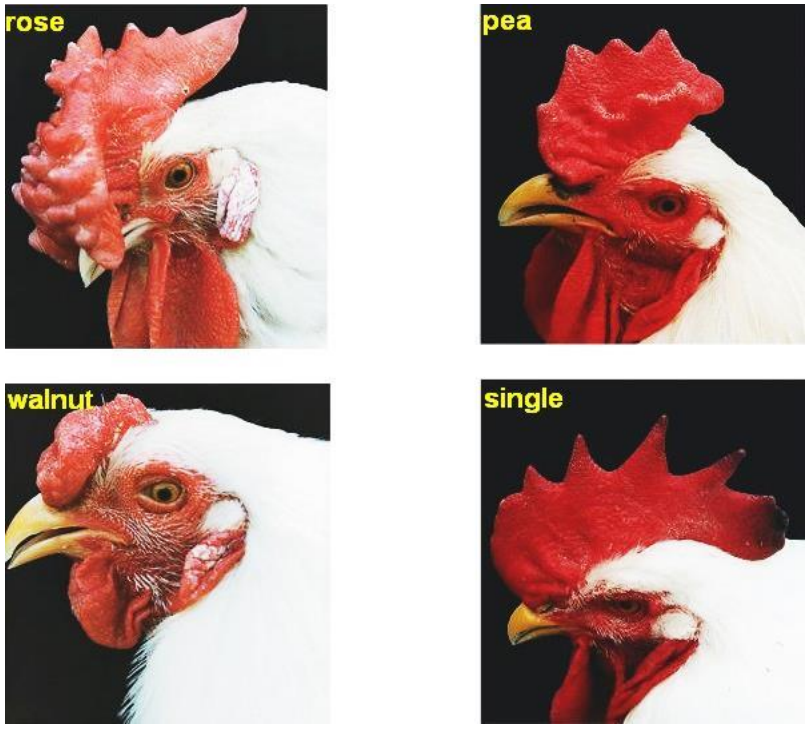
Many characteristics in plants and animals are produced by an interaction of several genes located on different loci; forming a gene complex. A single characteristic may be produced by the interaction of two or more genes occurring at different loci. A good example is shown by the inheritance of comb shape in domestic fowl

In this case, two genes on different chromosomes (loci) interact to produce four

distinct phenotypes of combs. Pea and rose combs are each produced by presence of the dominant forms of their respective genes (P and R respectively) but in absence of the other dominant gene. The walnut and single combs are produced by the interaction of the genes at both loci as summarized below:

Name of comb	**Production**	**Possible genotypes**
Pea comb	Dominant allele P but without dominant allele R	PPrr, Pprr
Rose comb	Dominant allele R but without dominant allele P	ppRR, ppRr
Walnut comb	Dominant alleles for both P and R	PPRR, PpRR, PPRr, PpRr
Single comb	Only by homozygous double recessive condition	pprr

Diagrams of the fowl combs



Starting with pure breeding parents, the following are the expected results for F1 and F2 generations.

Let; *P represent allele for pea comb, p for absence of pea comb*

R represent allele for rose comb, r for absence of rose comb

Parents : Male X Female

Phenotypes : Pea comb X Rose comb

Genotypes : PPrr X ppRR

Meiosis :

Gametes : all Pr all pR

Random fertilization :

F_1genotypes : All PpRr

F1 offspring phenotype: All walnut combed

Obtaining F2

Parents : Male X Female

Phenotypes : Walnut comb X Walnut comb

Genotypes : PpRr X PpRr

Meiosis :

Gametes : PR Pr pR pr PR Pr pR pr

Random fertilization

	PR	Pr	pR	pr
PR	PPRR ●	PPRr ●	PpRR ●	PpRr ●
Pr	PPRr ●	PPrr ■	PpRr ●	Pprr ■
pR	PpRR ●	PpRr ●	ppRR ▲	ppRr ▲
pr	PpRp ●	Pprr ■	ppRr ▲	pprr ○

Phenotypic ratios: 9 walnut: 3Pea: 3rose: 1single Sample

Question
In poultry, the allele for white feathers (W) is dominant over the allele for black feathers (w). The alleles P, for pea comb and R, for rose comb produce their respective phenotypes. If they are present together, the comb shape is modified to walnut and if their recessive alleles are present in homozygous recessive condition, a single comb I produced.
A cross between a black rose comb cock and a white walnut hen produced the following phenotypes:
3white walnut: 3black walnut: 3white rose: 3black rose: 1white pea: 1black pea: 1white single: 1black single. Identify the possible parental genotypes and show clearly how they give rise to the above phenotypes.

EPISTASIS
This is a form of gene interaction where one gene suppresses the effects of another gene at a different locus. The suppressing gene is referred to as an **epistatic gene** (inhibiting gene) while the suppressed gene is called a **hypostatic gene**.

Fur color in mice depends on two **non-allelic genes**, the dominant form of one gene is responsible for coloured fur while its recessive form results into no colour deposition and the phenotype is white (albino). If colour is present, the nature is determined by another gene whose dominant allele produces grey fur (agouti) while the recessive allele produces black fur.

Any of the two colours can be present only and only if their respective alleles are accompanied by the gene for coloured fur. Absence of this gene will result into albinos even if the genes for grey or white are present. The gene for coloured fur is hypostatic to the gene responsible for colour of fur (hypostatic).

This interaction produces three possible phenotypes as summarized below.

Phenotype	**Possible genotypes**
Grey (agouti)	AAGG, AAGg, AaGG, AaGg
Black	AAgg, Aagg
Albino (white)	aaGG, aaGg, aagg

Examples
In oat plants, the inheritance of color is controlled by the gene with two alleles, the dominant results into colour formation while the recessive results into no colour formation (white or albino). The other gene is responsible for the kind of colour, if present with the allele for grey being recessive to one for black.

Identify the nature of gene interaction and show the F1 and F2 outcomes starting with true breeding parental stocks.

Let; *A represent allele for colour, a for absence of colour*

B represent allele for black, b for grey

Parents	:	Male	X	Female
Phenotypes	:	Black	X	White
Genotypes	:	AABB	X	aabb
Meiosis	:			
Gametes	:	all AB		all ab
Random fertilization	:			
F_1genotypes	:	All AaBb		

F1 phenotypes: All black

Obtaining F2;

Parents	:	Male	X	Female
Phenotypes	:	Black oat	X	Black oat
Genotypes	:	BbAa	X	BbAa
Meiosis	:			
Gametes	:	BA Ba bA ba		BA Ba bA ba

Random fertilization

	BA	Ba	bA	ba
BA	BBAA ●	BBAa ●	BbAA ●	BbAb ●
Ba	BBAa ●	BBaa ■	BbAa ●	Bbaa ■
bA	BbAA ●	BbAa ●	bbAA ○	BBAa ■
ba	BbAa ●	Bbaa ■	bbAa ○	bbaa ○

F2 offspring phenotypic ratio: 9 black seeds: 4 grey seeds: 3white seeds

Epistasis also occurs in ***Cucurbita pepo*** in which there are 3 types of fruits colour white, yellow and green. White is found to be dominant over yellow as well as green colour. When yellow is crossed with green, yellow is found to be dominant. Here, the character (colour of fruits) is governed by two pairs of genes.

White and green – white dominant
Yellow and green – yellow dominant
White and yellow - white dominant

Thus white is the dominant epistatic factor and yellow is hypostatic factor.

If white dominant is represented by allele W, and its recessive by allele Y, both non – allelic factors or genes may be represented as follows:-

P	WHITE	×	YELLOW
	WWyy		wwYY
GAMETES	Wy		wY
F_1			
	WwYy		
	WHITE		
GAMETES	WY, Wy, wY, wy		

Selfing

♀ \ ♂	WY	Wy	wY	wy	Genotypes	
WY	WWYY White	WWYy White	WwYY White	WwYy White	WWYY-1 WWYy-2	
Wy	WWYy White	WWyy White	WwYy White	Wwyy White	Wwyy-2 WwYY-2	
wY	WwYY White	WwYy White	wwYy Yellow	wwYy Yellow	WWyy-1 WwYy-4	12 White
wy	WwYy White	Wwyy White	wwYy Yellow	wwyy Green	wwYY-1 wwYy-2	3 Yellow
					wwyy-1	1 Green

(Showing genotypes & phenotypes of F_2 generation)

COMPLEMENTARY GENES

These are two hormone genes that are independent and interact with each other to bring about particular phenotype such that the dominant gene allele from one gene can only bring an effect if the dominant allele from the other gene is also present. In other words, the two genes complement with each other with expression of a particular phenotype.

It was discovered in pea plants when two plants producing only white flowers were crossed.

All the F1 offsprings had purple flowers. On crossing the F1 offsprings, the F2 plants had purple and white flowers in the ratio of 9: 7.
The results arise because one gene C controls the production of a colourless pigment in the precursor and the other gene P is for the conversion of colourless pigment in to the purple colour.
Unless both dominant alleles are present, flowers will be white either because no pigment was formed or it was formed but not converted to the purple colour.

P White flowers × White flowers
CCpp ccPP
Gametes Cp cP
Purple flowers
CcPp
Gametes : CP.Cp, cP, cp Gametes

♀ \ ♂	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 Whie	ccPp White	ccpp White

Gametes

Phenotypic ratio : 9 Purple : 7 White

(Representing a Cross between two varieties of sweet pea)

SUPPLEMENTARY GENES

These are genes that have no effect on their own but can work with other genes to bring about a particular phenotype.
The most studied supplementary genes have been seen coat colour of mice. There are three coat colours namely Black, Albino and agouti controlled by two genes represented by alleles A for albino and B for black.

If the dominant gene A is present alone, it gives the albino coat colour. If present with the dominant gene B, it gives the agouti cat colour and if B is present alone, it gives the black coat colour.

However the presence of even recessive alleles for both genes gives albino coat colour. This shows that the dominant gene A has no effect on its own but supplements the dominant gene B to give the agouti coat colour.

Parent phenotype: Albino coat x Black coat

Parent genotype (2n): Mouse AAbb x mouse aaBB

Meiosis

Gametes (n): Ab, aB

Random Fertilization

F1 genotypes: AaBb

F2 phenotypes: All agouti fur coloured mice

Selfing F1 offsprings
To obtain F2

Parent phenotype: Agouti mouse x Agouti mouse

Parent genotype (2n): AaBb x AaBb

Meiosis

Gametes (n): AB, Ab, aB, ab

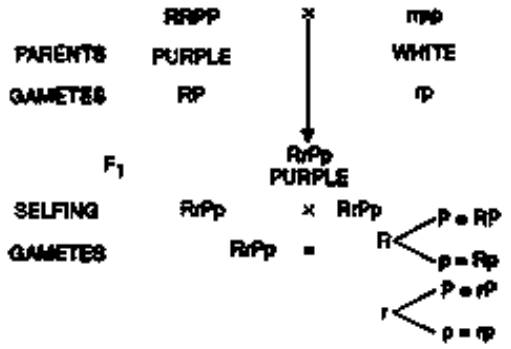
Random fertilization
Using a punnett square

♀ \ ♂	AB	Ab	aB	ab
AB	AABB ●	AABb ●	AaBB ●	AaBb ●
Ab	AABb ●	AAbb ○	AaBb ●	Aabb ○
aB	AaBB ●	AaBb ●	aaBB ▲	aaBb ▲
ab	AaBb ●	Aabb ○	aaBb ▲	aabb ○

● Agouti fur coloured mice with A and B alleles
○ Albino fur coloured mice with A and b alleles
▲ Black fur coloured mice with a and B alleles

F2 phenotypic ratio: 9 agouti : 4 albino : 3 black

In plants, supplementary genes include the development of grain colour in maize which is governed by 2 dominant genes R – for red colour production and P – which is unable to produce colour on its own but it modifies the colour produced by the gene R from red to pink. The recessive allele p has no effect on grain colour. The homozygous state of the recessive allele r (rr) checks the production of red colour, i.e. gives white coloured seeds.



Gametes

F_2 ♀ \ ♂	RP	Rp	rP	rp
RP	RRPP Purple	RRPp Purple	RrPP Purple	RrPp Purple
Rp	RRPp Purple	RRpp Red	RrPp Purple	Rrpp red
rP	RrPP Purple	RrPp Purple	rrPP White	rrPp White
rp	RrPp Purple	Rrpp Red	rrPp White	rrpp White

Gametes

Results : 9 Purple : 3 Red : 4 White

(Cross between Purple and White grain colour in Maize.)

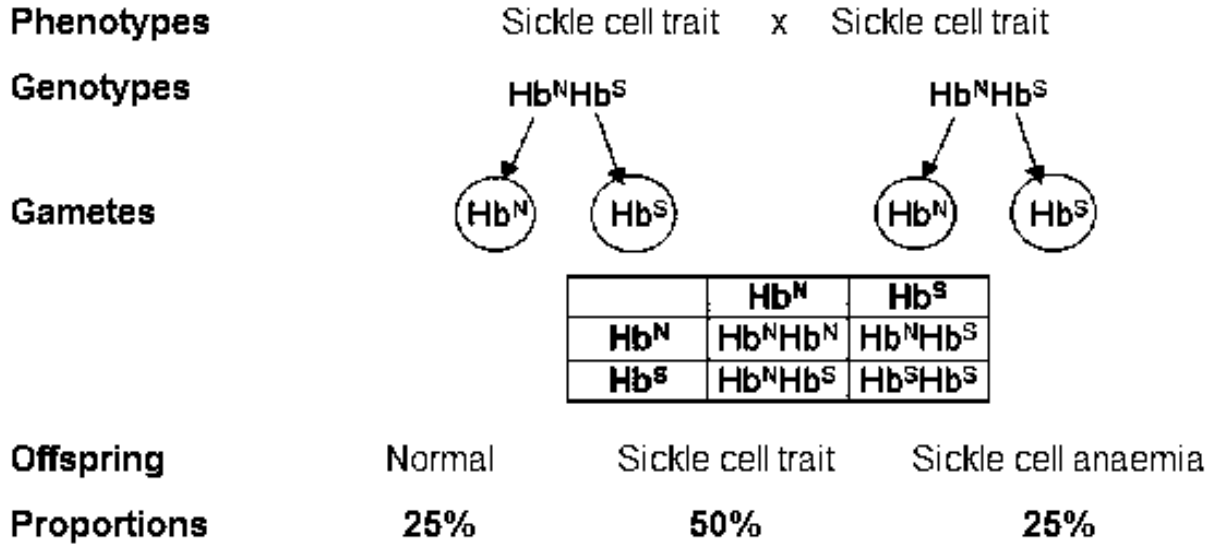
PLEIOTROPY

This is a condition where a gene exerts more than one phenotypic effect. The most characterized is the sickle cell gene. Individuals having the sickle cell gene have an abnormal haemoglobin called the **haemoglobin S** which has a low oxygen carrying capacity and tends to appear crystalline under low temperature and low oxygen tensions. They also have crescent or sickle shaped red blood cells.

Normal red blood cells are either circular disks or biconcave disks. These abnormal red blood cells are not pliable and they tend to break as they squeeze through the tiny capillary, resulting in to severe blood loss which may appear in urine. This condition is called sickle cell anaemia.

Carriers of the sickle cell trait or gene suffer from mild anaemia but they stand

lower chances of suffering from malaria. This is because the causative agent of malaria (plasmodium) cannot survive in their blood. This is called heterozygous superiority, because the homozygous individuals either suffer from malaria or sick cell anaemia. This explains why the gene for sickle cell cannot disappear from the population within a few generations.



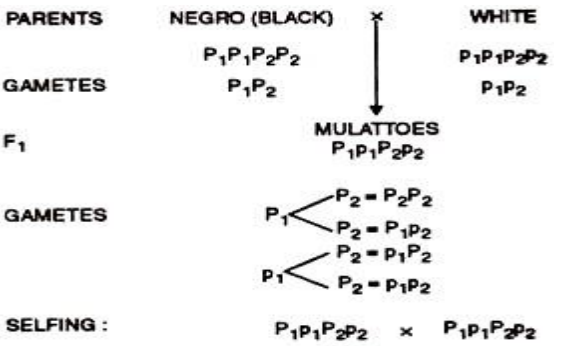
The gene is inherited in the same Mendelian fashion but it is non – Mendelian because of the two phenotypic effects.
Pleiotropy also occurs in the inheritance of phenylketonuria, fruit flies and vestigial genes, chicken and their frizzle traits, the process of pigmentation and deafness in cats. In plants, Pleiotropy involves the inheritance of flower colour, seed coat colour and axil colour of pea plants.

POLYGENIC INHERITANCE
Under normal cases, a single gene or character is controlled by a pair of alleles but there are some characteristics that are controlled by many allelic genes. Called non allelic. These are characteristics of continuous variation where by each gene exerts an effect on the phenotype which becomes cumulative with increasing number of dominant genes.

For example a cross between a negro having three dominant gene pairs for melanin production (BBBBBB) and an albino without gene pairs for melanin production (bbbbbb). All the F1 offsprings will show an intermediate skin colour.
On selfing the F1 offsprings, a gradation of skin colours will be observed ranging from Negros, at one extreme through various skin colours to albinos at the other extreme.

If two gene pairs are used such as $\mathbf{P_1P_1P_2P_2}$ and $\mathbf{p_1p_1p_2p_2}$, the F2 phenotypic

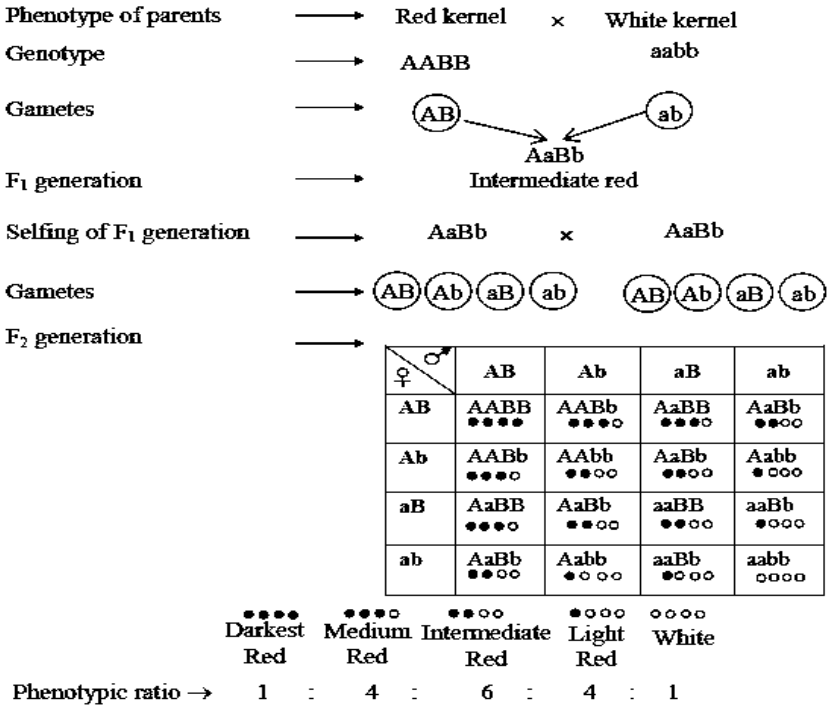
ratio is 1: 4 : 6 : 4 : 1, modifying the expected 9:3:3:1. If a frequency polygon is plotted, a normal bell shaped curve is obtained as shown below.

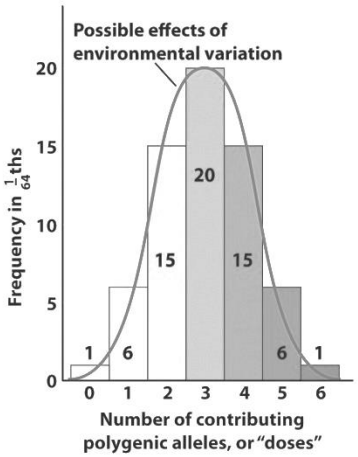


F_2 generation

♀ \ ♂	P_1P_2	P_1p_2	p_1P_2	p_1p_2	Phenotypes
P_1P_2	$P_1P_1P_2P_2$ Black	$P_1P_1P_2p_2$ Dark Mulattoes	$P_1p_1P_2P_2$ Dark Mulattoes	$P_1p_1P_2p_2$ Mulattoes	Black = 1 (Negro)
P_1p_2	$P_1P_1P_2p_2$ Dark Mulattoes	$P_1P_1p_2p_2$ Mulattoes	$P_1p_1P_2p_2$ Mulattoes	$P_1p_1P_2p_2$ Light Mulattoes	Dark Mulattoes = 4
p_1P_2	$P_1p_1P_2P_2$ Dark Mulattoes	$P_1p_1P_2p_2$ Mulattoes	$p_1p_1P_2P_2$ Mulattoes	$p_1p_1P_2p_2$ Light Mulattoes	Mulattoes = 6
p_1p_2	$P_1p_1P_2p_2$ Mulattoes	$P_1p_1p_2p_2$ Light Mul.	$p_1p_1P_2p_2$ Light Mul.	$p_1p_1p_2p_2$ Light White	Light Mulattoes = 4 White = 1

However, if a mating between pure breeding red kernel seeds and pure breeding white kernel seeded plants. All the F1 offsprings had an intermediate red coloured kernels. Selfing the F1 offsprings gives the same F2 phenotypic ratio stated before.





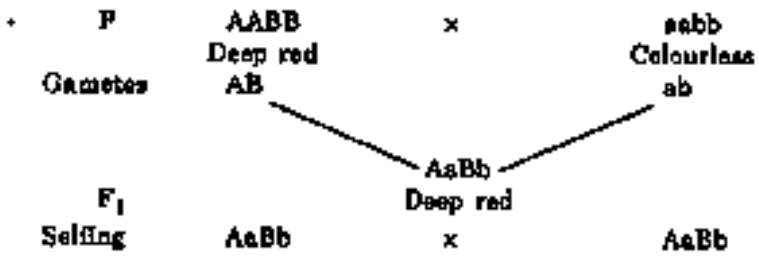
Therefore, polygenic inheritance involves the control of height, skin colour, weight, intelligence and others in animals. In plants they include the colour and shape of the stem, pollen, flower, yield, oil content, size of the seed, time to mature or flower, etc.

POLYMERIC GENES

When two forms genes govern any character separately; their effect is equal but when both the genes are present together, their phenotypes effect is increased or raised as if the effects of the two genes were additive or cumulative. It is notable in this case that both the genes show complete dominance.
Additive or cumulative effect of genes present at different loci Is called **polymerism**.

In wheat, three types of pericarps colour is found namely deep red, light red and colourless. When a cross is made between plants having deep red (AABB) and colourless (aabb) pericarp, the F1 (AaBb) have deep red pericarp due to addidtive effect of both genes A and B

In F2 generation, on an average, $^{9}/_{16}$ plants will have dominant alleles of both the genes A and B; as a result of these plants will produce deep red pericarpd. $^{6}/_{16}$ plants will have light red pericarp since they have either A or B, but not both. The rest $^{1}/_{16}$ plant will be homozygous recessive for both the genes and will be therefore, colourless.



Gametes

♀ \ ♂	AB	Ab	aB	ab
AB	AABB Deep red	AABb Deep red	AaBB Deep red	AaBb Deep red
Ab	AABb Deep red	AAbb Light red	AaBb Deep red	Aabb Light red
aB	AaBB Deep red	AaBb Deep red	aaBB Light red	aaBb Light red
ab	AaBb Deep red	Aabb Light red	aaBb Light red	aabb Colourless

Gametes

Phenotypic ratio : 9 Deep red : 6 Light red : 1 Colourless pericarp

INHERITANCE OF SEX AND SEX DETERMINATION

In man, there are 23 pairs of chromosomes; of these only one pair carries genes for sex determination. These are called sex chromosomes (**heterosomes**) designated X and Y, and the other 22 pairs are called **autosomes**.

A genotype XX is described as **homogametic** and is female while XY is described as **heterogametic** and is a male. During meiosis, the two sex chromosome segregate such that each ovum carries one X chromosome, half of the sperms carry an X chromosome and the other half carry a Y chromosome.

If a sperm carrying an X chromosome fuses with the ovum, the zygote is female and if the sperm is carrying a Y chromosome, the zygote is male. Sex is therefore determined by the sex chromosome carried in the sperm as a matter of chance.

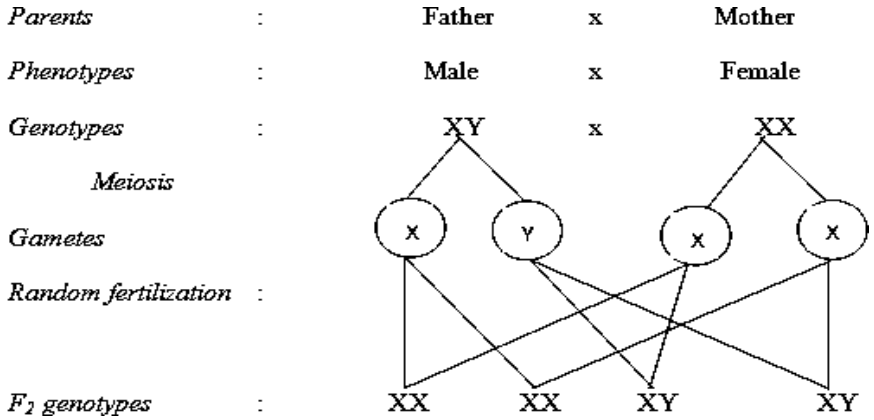
This is called the **X-Y system** and occurs mainly in mammals with humans inclusive. The females are described as homogametic because all their gametes contain the same sex chromosome-the X chromosome while the males are heterogametic because 50% of the gametes produced contain an X chromosome and 50% contain a Y chromosome for sex.

In some animals like birds (including poultry), moths and butter flies; the sex genotypes are reversed. The homogametic genotypes (XX) are male while the

heterogametic genotype (XY) is female.

In some cases, the Y chromosome is completely absent and the heterogametic sex (XO) is male. This is the X-O system as in grass hoppers, cockroaches and some insects. The sex of the offsprings is determined by whether the sperm cell contains an X chromosome or no sex chromosome. This implies that the Y chromosome does not carry genes needed for survival of the organisms.

In some species of bees and ants, there are no sex chromosomes. Females develop from fertilized eggs and are thus diploid while males develop from un fertilized eggs and are haploid, without feathers.
Example:



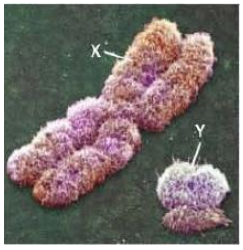
Genotypic ratios: 1XX: 1XY
Phenotypic ratios: 1female: 1male

This shows that there is a 50% chance of any child being a male or female

Environmental determination of sex
Sex is primarily genetically determined as described above but in some lower animals, sex can be determined by environmental factors such as temperature, salinity, type of food etc. for example in some turtles the eggs laid warm sand develop into females while those laid in cool sand develop into females.

SEX CHROMOSOMES

The sex chromosomes are called heterosomes because they are non-identical and are designated X and Y. The X chromosome is rod shaped and much bigger than the Y chromosome which is hook shaped.



The Y chromosome carries genes responsible for secondary male sex characteristics, differentiation of testes and development of genital organs in humans. Actually in some organisms, the Y chromosome is absent and is believed not to carry genes necessary for survival of the organism and is described as genetically inert.

SEX LINKAGE:

In humans, there are several thousands of characteristics each genetically controlled. With only 23 pairs of chromosomes, each chromosome must therefore carry many genes; a phenomenon that does not exclude sex chromosomes. These in addition to genes responsible for sex differences may carry genes determining some other features in the body.

Sex-linked genes are genes carried on sex chromosomes and inherited together with those determining sex. Sex linked traits (characters) are traits determined by genes carried on sex chromosomes and inherited together with those determining sex.
Note: The Y chromosomes don't carry genes, sex linked genes are specifically carried on the X sex chromosomes but not on the Y chromosome.

Many experiments were carried out by Thomas Morgan about sex-linked genes in drosophila. In one of his experiment, Morgan mated a wild type (pure breeding) red-eyed female with a mutant (white eyed) male. All the F_1 hybrids were red eyed. He went on to interbreed the F_1 males and females to obtain an F_2 generation which consisted of red eyed and white eyed offsprings in a ratio of 3:1 respectively. However, all female were red eyed and all the white eyed flies were males though some males were red eyed.

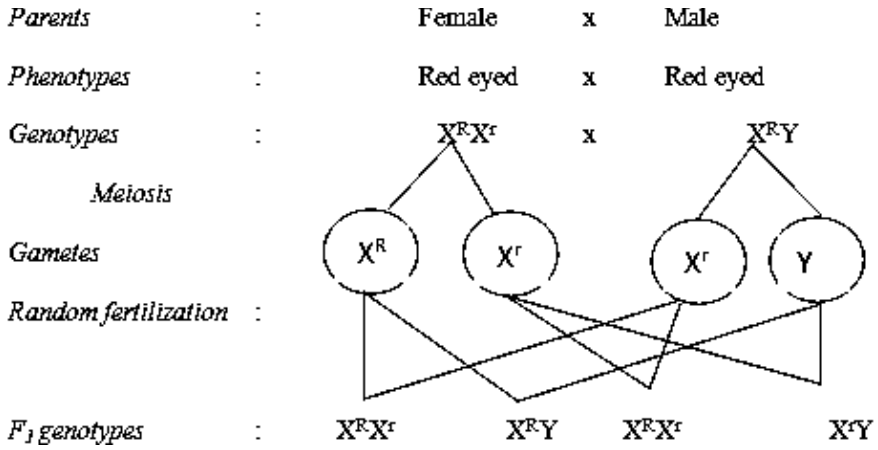
In conclusion, all the F_1 were red eyed; implying that this allele is dominant over that for white. Since in the F_2 all the white eyed were males, this indicates that the gene for eye colour is located on the X chromosome and there is no corresponding locus on the Y chromosome; otherwise some females would also be white eyed

Let;

R represent the allele for red eyes, r represents the allele for white eyes

Parents	:	Female	x	Male
Phenotypes	:	Red eyed	x	White eyed
Genotypes	:	X^RX^R	x	X^rY
Meiosis				
Gametes		X^R		X^r , Y
Random fertilization	:			
F_1 genotypes	:	X^RX^r		X^RY

Obtaining F2 generation



Phenotypic ratios: 3 red eyed: 1 white eyed
Note that all the white eyed are males yet some red eyed are males

Sample question:

1. If the gene for eye colour was autosomal, predict the phenotypes of the F2 hybrids (including sex) in this hypothetical cross. (Show your working).
   a. Perform a test cross on the F1 female fly obtained in the above cross.
   b. What would be the phenotypes of the reciprocal cross between the original parents?
2. In drosophila, the genes for wing length and eye colour are sex-linked; with normal wings and red eyes being dominant to miniature wings and white eyes respectively.
   a. In a cross between a miniature-winged red eyed male and a homozygous normal wing white eyed female; explain the expected appearance of F1 and F2 generations.
   b. Crossing a female from the F1 generation above with a miniature wing white eyed male gave the following results:

   Normal wing white eyed males and females = 35
   Normal wing red eyed males and females = 17
   Miniature wing red eyed males and females = 18
   Miniature wing white eyed males and female = 36

   Account for the appearance and numbers of the phenotypes listed above.

Examples of sex linked characters in man include the following

- Haemophilia
- Red – green or Colour blindness
- Pre-mature balding
- Eye colour in drosophila
- Muscular dystrophy or Duchene muscular dystrophy (BMD

Most of these characters are caused by recessive alleles and in a genetic cross, these must be represented as superscripts on the x sex chromosome.

Characteristics of sex linked characters

- They are determined by recessive alleles while the dominant allele determines normal conditions.
- The genes controlling the character are carried on X chromosomes only. The Y chromosome does not carry any genes.
- In a population, the heterozygotes are females and they are called **carriers**. Carrier females are phenotypically normal and do not suffer the conditions.
- Sex – linked characters are more common among males than females.
- The recessive alleles are transmitted to the male offsprings by their mothers.
- The mothers transmit the recessive alleles to their sons while the daughters inherit one of their X chromosomes carrying either recessive or dominant allele from the father and one being inherited from the mother.
- Offer mainly disadvantageous characters to individuals.
- Frequency of colour blindness is higher than that for haemophilia in a population.

HAEMOPHILIA (BLEEDERS' DISEASE)

Haemophilia is a recessive sex-linked blood disorder that leads to absence of one or more blood clotting factors, leading to prolonged bleeding even from minor cuts.

Just like other sex-linked traits, haemophilia is carried on the X chromosome and the responsive allele is recessive to the normal allele. The condition interferes with formation of blood clotting factors; commonly factor VIII (Anti-Haemophiliac Globulin) whose absence greatly delays the blood clotting process. This results into prolonged bleeding and excess blood loss even from minor cuts which may lead to death.

The allele being recessive, haemophiliac females must inherit two copies of the defective allele while males inherit one copy. The heterozygous females show normal blood clotting and are described as carriers. This is because the other X chromosome carries a dominant allele needed for normal blood clotting which suppresses the recessive allele for haemophilia. The males lack the alternative allele and the recessive allele is automatically expressed phenotypically.

Example: When a carrier woman is married to a normal man

Let;

H represent the allele for normal blood clotting, h represents the allele for haemophilia

Parents : Female x Male

Phenotypes : Normal x Normal

Genotypes : X^HX^h x X^HY

Meiosis

Gametes : X^H X^h X^H Y

Random fertilization :

F_1 genotypes : X^HX^H X^HY X^HX^h X^hY

It can be noted that there is a 50% chance of a daughter being a carrier and a 50% chance of a son being haemophiliac.

Sons can only inherit haemophilia (and other sex linked traits) from their mothers but not fathers as they only inherit the father's Y chromosome and not the X chromosome that carries sex linked genes. Girls can inherit from both parents.

Today, people with hemophilia are treated as needed with intravenous injections of the missing protein.

COLOUR BLINDNESS

It is a recessive sex linked character that leads to inability of the individual to distinguish between colours.

It is caused by a recessive allele, carried on the X chromosome and inherited in the same way as haemophilia. Colour vision is due to presence in the retina of red, blue and green cones needed for seeing the respective colours.

The recessive alleles result into absence of some of these cones which renders inability to identify such colours from other related colours. This is called **colour blindness**; the commonest being red-green colour blindness where individuals lack red and green cones in their eyes.

Example

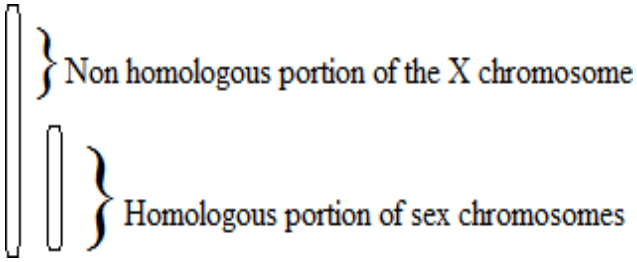
Green colour blindness is sex linked in man. A normal man married a colour blind woman. Using suitable genetic symbols workout the genotypes and

phenotypes of their children Colour blind individuals **are more common** in the population than haemophiliacs despite the two being inherited in the same way.

This is because haemophilia is associated with many lethal effects due to excessive internal and external bleeding which increases chance of dying before reproductive maturity to pass on their genes to the next generations.

Colour blindness exerts less lethal effects as colour vision is not much necessary for survival. Colour blind individuals usually survive to reproductive age and pass the allele to subsequent generations hence increasing the number of colour blind individuals in the population. Also haemophiliacs are advised to desist from reproducing as they may end up bleeding to death which further reduces the numbers of haemophiliacs.

NB: Sex linked characters have been found to occur more commonly in males as compared to females in the human population. Being caused by recessive alleles, the other X chromosome in females may carry a dominant allele to mask the defective allele hence preventing its phenotypic expression in the population. In males however, these genes are carried on the non-homologous portion of the X chromosome for which there is no alternative gene on the Y chromosome. Such genes are automatically expressed in males leading to higher frequencies in males as compared to females.



MUSCULAR DYSTROPHY

DMD is a tragic, wasting disease affecting infant children mainly boys, where muscles of the body are replaced by fibrous tissue, resulting in to progressive body weakness and difficulty in breathing.

This disease can result to death of the sufferer at an early age (before the age of 20 years is reached). The gene for DMD is sex linked and found on the X chromosome. The allele for DMD codes for an enzyme that induces the replacement of muscle by fibres.

Currently there is no cure for muscular dystrophy. But medications like

glucocorticoids prednisone or deflazacort and therapy can help manage symptoms and slow the course of the disease.

SEX LIMITED CHARACTERISTICS

Se limited characters are characters that are more pronounced in one sex than the other.

Though both sexes may carry genes responsible for these characteristics, pronounced expression is strictly limited to one of the two sexes. They are usually carried on autosomes but may largely be influenced by the level of sex hormone in the body.

Examples include;

Facial hair, deep voice, baldness etc in males

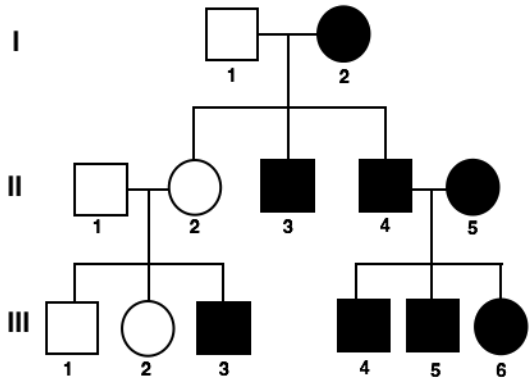
Breasts, lactation, widening of hip bones, high pitched sound etc

PEDIGREE ANALYSIS

This is a chart of the ancestral history of related individuals showing inheritance of sex linked characters such as haemophilia or other characters.

Question

1. The pedigree below tracks Duchene muscular dystrophy (DMD) through several generations. DMD is an X – linked recessive trait.



If individuals 1 – I and 2 – I had another son, what is the chance that he would have DMD?

QUESTIONS

I. Beth and Tom each have a sibling with cystic fibrosis, but neither Beth nor Tom nor any of their parents have the disease. Calculate the probability that if this couple has a child, the child will have cystic fibrosis.

What would be the probability if a test revealed that Tom is a carrier but Beth is not?

2. Joan was born with six toes on each foot, a dominant trait called polydactyl. Two of her five siblings and her mother, but not her father, also has extra digits. What is Joan's genotype for the number-of-digit character? Explain your answer. Use D and d to symbolize the alleles for this character.

3. What would you suspect if Peter was born with polydactyl, but neither of his biological parents had extra digits?

4. Incomplete dominance and epistasis are both terms that define genetic relationships. What is the most basic distinction between these terms?

5. If a man with type AB blood marries a woman with type 0 blood, what blood types would you expect in their children?

6. A rooster with gray feathers is mated with a hen of the same phenotype. Among their offspring, 15 chicks are gray, 6 are black, and 8 are white. What is the simplest explanation for the inheritance of these colors in chickens? What phenotypes would you expect in the offspring of a cross between a gray rooster and a black hen?

7. Genes A, B, and C are located on the same chromosome. Testcrosses show that the recombination frequency between A and *B* is 28% and between A and C is 12%. Can you determine the linear order of these genes? Explain.

VARIATION

Variation describes the differences in characteristics shown by organisms belonging to the same natural population or species

TYPES OF VARIATION

There are two basic forms of variation that occur in any large population basing on the phenotypic differences i.e. discontinuous and continuous variation.

Discontinuous variation

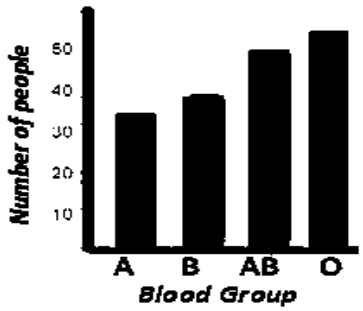
This is s a type of variation in which certain characteristics with in a population exhibit a limited form of difference. This produces individuals showing clear – cut differences with no intermediates between them.

Examples

-blood groups in humans, wing length in drosophila, melanic and light forms in *Biston betularia*, style length in *primula*, sex in animals and plants, tongue rolling, albinism.

Characteristics showing discontinuous variation are usually controlled by one or two genes which may have two or more allelic forms and their phenotypic expression is relatively unaffected by environmental conditions.

Discontinuous variation is also qualitative inheritance because the phenotypic variation is restricted to certain clear – cut characteristics.



Discontinuous data can be plotted on a bar chart. For example, a person can only have one of four blood groups – A, B, AB or O.

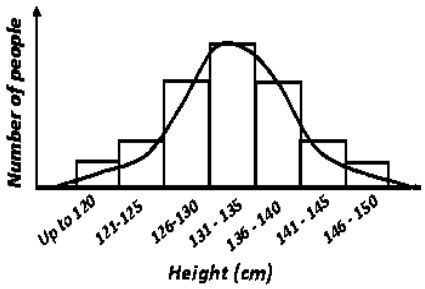
Continuous variation

This is where certain characteristics with in a population vary only very marginally between one individual and the next. This results in a gradation from one extreme to the other.

Examples

Mass, linear dimension, shape and colour of organs and organisms, heart rates, etc.

Characteristics exhibiting continuous variation are produced by the combined effects of many genes (polygenes) and environment factors.



Continuous data is plotted on a histogram, this shows a range of measurements from one extreme to another. The curve that the graph produces is know as normal distribution.

ORIGIN /SOURCES OF VARIATION

The core factors determining a phenotypic characteristic of an organism is the **genotype**. But the subsequent degree of expression allowed to tis genetic potential is influenced by the action of environmental factors during the development of the organism e.g. adequate light, temperature, water and soil conditions. Thus both genotypic/gene reshuffles and environmental factors interact to cause an effect on the phenotype.

Environmental factors

If organisms of identical genotypes are subjected to different environmental influences, they show variety. This is because environmental influences are various and often form gradations. They are responsible for continuous variation with in a population.

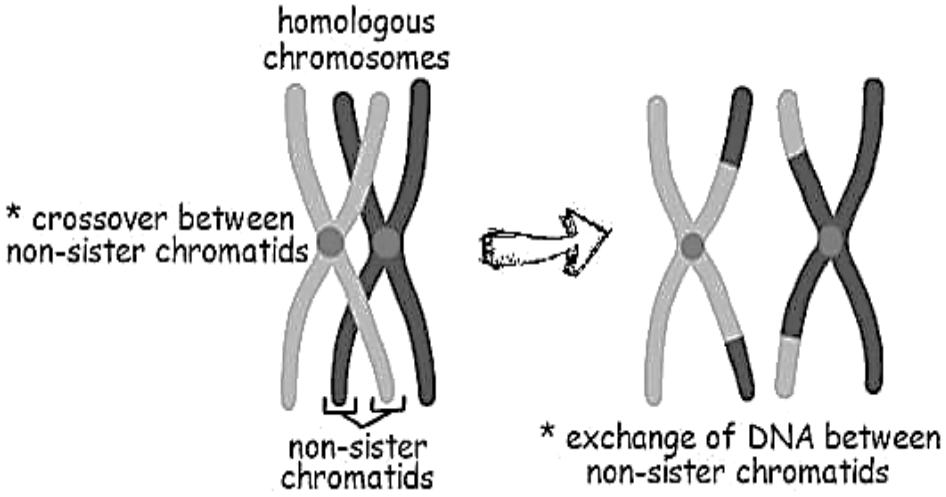
Reshuffling of genes

The sexual process in organisms has three method of causing variety.

- **Crossing over**: The reciprocal crossing over of genes between non – sister chromatids of homologous chromosomes may occur at a point called chiasmata, during prophase I of meiosis.

  As a consequence of this recombination, all four chromatids that comprise the bivalent will be genetically different. Chromatids that consist of a combination of DNA derived from both homologous chromosomes are called **recombinants**. Offsprings with recombinant chromosomes will have unique gene combinations that are not present in their parents.

  This produces new linkage groups and so provides a major source of genetic recombination of alleles.



- **Independent assortment**: The orientation of the chromatids of homologous chromosomes (bivalents) on the equatorial spindle during metaphase I of meiosis determines the direction in which the pairs of chromatids move during anaphase I. This orientation of chromatids is random.
  During metaphase II, the orientation of pairs of chromatids once more is random, and determines which chromosomes migrate to opposite poles of the cell during anaphase II. These random orientations and the subsequent independent assortment (segregation) of the chromosomes give rise to a large number of different chromosome combinations in the gametes.

  Note
  The total number of combinations that can occur in gametes is 2^n – where n = haploid number of chromosomes. Humans have 46 chromosomes (n=23) and thus can produce 8,388,608 different gametes (2^{23}) by random orientation. If crossing over also occurs, the number of gametes combination becomes immeasurable.

- **Random fusion of gametes**: During sexual reproduction, the fusion of haploid male and female gametes to form a zygote is completely random. This zygote can then divide by mitosis and differentiate to form a developing embryo. Thus, any male gamete is potentially capable of fusing with any female gamete.
  As meiosis results in genetically distinct gametes, random fertilization by egg and sperm will always generate different zygotes. This causes variation.

However, these sources of variation do not generate the major changes in genotype which are necessary in order to give rise to new species as described by evolutionary theory. These changes are produced by mutations.

MUTATIONS

These are changes in the amount, arrangement or structure of DNA of an organism.
These produce changes in the genotype which may be inherited by cells derived by mitosis or meiosis from the mutant cell.
Most mutations occur in somatic cells and are not passed from one generation to the next. Only those mutations which occur in the formation of gametes can be inherited. These mutations are the basis of discontinuous variation. Mutations on body cells are called **somatic mutations**.

TYPES OF MUTATIONS

There are two types of mutations i.e. point/gene mutation and chromosomal mutations/ aberrations.

Gene mutations

These describe the changes in the structure of DNA at a single locus on a chromosome. These cause the wrong arrangement of nucleotides on the DNA molecule leading to formation of wrong sequence of amino acids in the protein it makes.
This protein (enzyme) may have a different molecular shape hence unable to catalyze its reaction, and can't form end products of such reactions.
These cause profound effects in organisms e.g. a gene mutation resulting in the absence of pigments such as melanin, leading to albinism, cystic fibrosis and Huntington's disease.

There are many forms of gene mutations

- **Duplication**: a portion of a nucleotide chain becomes repeated. This mostly occurs in plant species
- **Addition**/insertion: an extra nucleotide sequence becomes inserted in the chain. This causes some diseases like fragile X syndrome characterized by moderate intellectual disability, anxiety and hyperactivity. Huntington's disease, myotonic dystrophy and cystic fibrosis.
- **Deletion**: a portion of the nucleotide chain is removed from the sequence. This results in the inheritance of cystic fibrosis cases and the cat cry syndrome (so called because children with this syndrome often have a cry that sounds similar to a cat meowing,
- **Inversion**: a nucleotide sequence becomes separated from the chain. It rejoins in its original position only inverted. The nucleotide sequence of this portion is therefore reversed.
- **Substitution**: one of the nucleotides is replaced by another which has a different organic base. For example, haemophilia, PKU (phenylketonuria) and a single base substitution in beta – haemoglobin gene, causes sickle cell anaemia

Chromosomal mutations

These result from a change in the amount or arrangement or structure of DNA.

a) Changes in whole set of chromosomes

There are two changes that occur to bring about chromosomal mutations i.e. aneuploidy and euploidy (polyploidy)

Aneuploidy

These are changes which involve the loss or gain of single chromosomes.
This causes formation of daughter cells, half having extra chromosomes (n + 1), (2n+1) and so on, whilst the other half having a chromosomes missing (n-1), (2n-1) and so on.
Aneuploidy arises from the failure of a pair or pairs of homologous chromosomes to separate during anaphase I of meiosis, a process called **non – disjunction**. If this occurs, both sets of chromosomes pass to the same pole of the cell and separation of the homologous chromosomes during anaphase II leads to formation of gamete cells containing either one or more chromosomes, too many or too few.

Fusion of either of these gametes with a normal haploid gamete produces a zygote with an odd number of chromosomes. Zygotes with less than the diploid number of chromosomes usually fail to develop, but those with extra chromosomes develop.

Aneuploidy in animals causes abnormalities e.g. in humans chromosomal mutation resulting from non – disjunction is a form of **trisomy** called **Down's syndrome** (2n = 47), this is characterized by mental retardation, coarse and straight hair, short stature and relatively small skull due to poor skeletal development, etc.

Non – disjunction of sex chromosomes can also occur e.g. **Klinefelter's** syndrome with XXY, XXXY or XXXXY genotypes. These individuals appear male but have small testes and no sperm in the ejaculate, little facial hair, voice pitched than normal, some breast development, usually taller than normal, etc. **Turner's** syndrome has one missing X chromosome hence XO genotype. These individuals are aborted but those who survive are phenotypically females but small in stature and sexually immature. Turner's syndrome also seems to be responsible for a relatively high proportion of miscarriage.

Euploidy (polyploidy)

This is where gametes and somatic cells contain multiples of the haploid number of chromosomes. When three sets (3n) of chromosomes are present, the organism is triploid, with four sets (4n), it is tetraploid.

Polyploidy is much more common in plants than in animals e.g. most species of angiosperms are polyploidy. It is rare in animals because increased number of chromosomes in polyploidy makes normal gametes formation during meiosis more prone to error. But plants are capable of propagating themselves vegetatively, they are able to reproduce despite being polyploidy.

Polyploidy is associated with advantageous features such as increased size, hardness and resistance to diseases, drought tolerant plants, short gestation and high yielding plants. This is called **hybrid vigor**. Most domestic plants are polyploids producing large fruits, storage

organs, flowers and leaves.

There are two forms of polyploidy; autopolyploidy and allopolyploidy.

- **Autopolyploidy**: This is a polyploidy in which the increases in sets of chromosomes occurs naturally or artificially with in the same species. The actual number of chromosomes in an autopolyploid is usually an exact multiple of its haploid numbers.
  For example, if chromosomes undergo replication (during interphase) and the chromatids separate normally (during anaphase) but the cytoplasm fails to cleave (during cytokinesis)

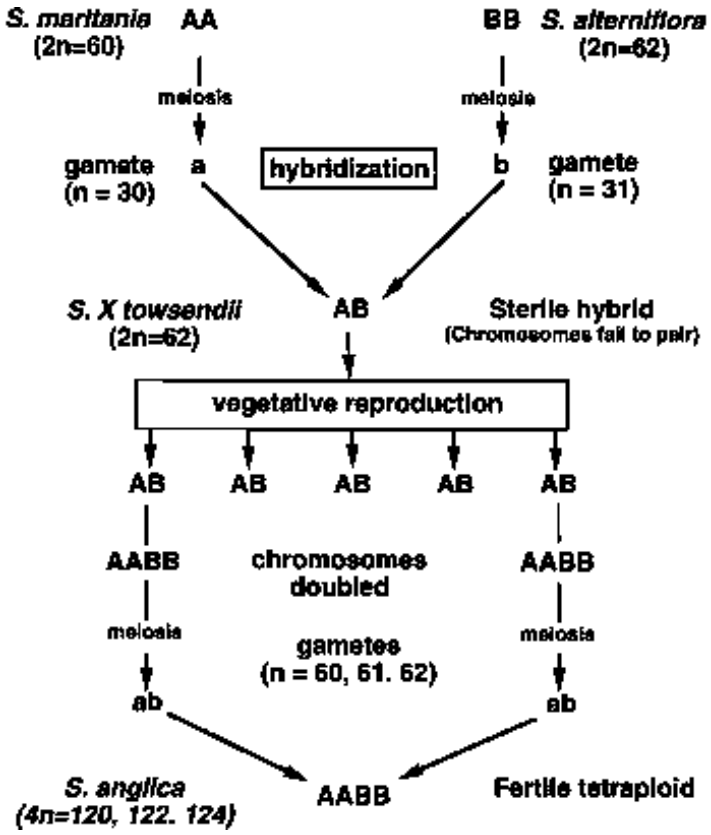
  Autopolyploidy can be induced by a chemical called **colchicine** from corms. Colchicine inhibits spindle formation and so prevents chromosomes separating during anaphase. Autopolyploid can be as fertile as diploids if they have an even number of chromosomes sets.

  A modified form of polyploidy can occur in animals and gives rise to cells and tissues which are polyploidy. This process is called **endomitosis** and involves chromosome replication without cell division. The giant chromosomes in the salivary glands of drosophila and tetraploid cells in the human liver are produced by endomitosis.

- **Allopolyploidy**: this occurs when a polyploidy offspring is derived from two distinct parental species. This condition arises when the chromosome number in a sterile hybrid becomes doubled and produces fertile hybrids. The F1 hybrids produced from different species are sterile since their chromosomes cannot form homologous pairs during meiosis i.e. possess uneven chromosome pairing, a process called **hybrid sterility**.

  However, if cytokinesis fails to occur in one of the gametes, the hybrid offspring will have paired chromosomes from that parental species. If the hybrid interbreeds with a member of the other parent species, all chromosomes from both parent species will be paired. The resulting offspring will now be fertile and have the combined chromosome composition of both parental species.

  Most allopolyploid species have a diploid chromosome number which is the sum of the diploid numbers of their parental species. For example rice grass, *Spartina anglica* (2n = 122) is a fertile allopolyploid hybrid produced from a cross between *Spartina maritima* (2n = 60) and *Spartina alterniflora* (2n = 62). The F1 hybrid formed from the latter two species is sterile and is called *Spartina townsendii* (2n = 62) as shown below.



Another case of allopolyploidy occurs in the species of wheat used to make bread, *Triticum aestivum* (2n = 42) which was selectively bred by crossing a wild variety of wheat, einkorn wheat (2n = 14) with a wild grass (2n = 14), a different species of wheat, emmer wheat (2n = 28) was produced. Emmer wheat was crossed with another species of wild grass (2n = 14) t0 produce *Triticum aestivum*, which actually represents the hexaploid condition (6n) of the original einkorn wheat.

b) Changes in chromosome structure

There are four mays or mutations that cause changes in the structure of the chromosome namely

- **Deletion**: a portion of a chromosome is lost
- **Inversion**: a portion of the chromosome becomes deleted but becomes reattached in an inverted position.
- **Translocation**: a portion of the chromosome becomes deleted and rejoins at a different point on the same chromosome or with a different chromosome.
- **Duplication**: a portion of the chromosome is doubled resulting in repetition of a gene sequence.

CAUSE OF MUTATIONS

The natural mutation rates can be increased artificially by certain chemicals or energy sources. Any agent of which induces mutations is called a **mutagen** and the resulting individual is a **mutant**.
They include

- High energy radiations like UV light, X – rays and gamma rays.
- High energy particles such as alpha and beta particles, and neutrons, cosmic radiations, etc.
- Chemicals like colchicine, formaldehyde, nitrous acid and mustard gas, caffeine, drugs, pesticides and food preservatives.

IMPLICATIONS OF MUTATION

- Many cases of mutations are lethal and prevent development of the organism.
- Some chromosome mutations bring certain gene sequences together, causing beneficial characteristics.
- Gene mutations may lead to several alleles occupying a specific locus, this increases both the heterozygosity and size of the gene pool of the population, leading to increased variation within a population.
- Gene reshuffling as a result of crossing over, independent assortment, random fertilization and mutations may increase the number of continuous variation, though evolutionary implications may be short-lived since the change produced are rapidly diluted.
- Some gene mutations may increase discontinuous variation which has profound effect on changes in the population.
- Most gene mutations being recessive to the normal allele, this forms genetic equilibrium with the rest of the genotype and the environment as a result of withstanding selection over many generations.