MASAKA S.S
S.6
BIOLOGY

Genetics is a branch of biology concerned with the study of genes, genetic variation, and heredity in organisms.

Gregor Mendel was the scientist who discovered genetics in the late 19th-century. Mendel studied trait inheritance, patterns in the way traits are transmitted from parents to offspring. He described how organisms inherit traits as discrete units of inheritance.

Using pea plants (*Pisum sativum*),he studied very many traits using these plants.

Advatages of using peas in breeding experiments

- The plants can easy to cultivated.
- They have several varieties with distinct characteristics.
- Their reproductive structures are completely enclosed by the petals to allow self- pollinating. This leads to the production of generation of the same characteristic after generation, a phenomenon known as **pure breeding**.
- Artificial cross-breeding between varieties is possible and to produce fertile hybrids.

Important terms used in genetics

Gene. This is the basic unit of inheritance of a given characteristic.

Allele. This is the alternative form of the same gene responsible for determining contrasting characteristic.

Locus. This is the position of an allele within a DNA molecule or on a chromosome.

Homozygous. This is a diploid condition in which the alleles at a given locus are identical.

Heterozygous. This is a diploid condition in which the alleles at a given locus are different.

Phenotype. This is the observable characteristics of an individual usually resulting from the interaction between the genotype and the environment in which development occurs.

Genotype. This is the genetic constitution of an organism with respect to the alleles under consideration.

Dominant allele. This is the allele which influences the appearance of the phenotype even in the presence of an alternative allele.

Recessive allele. This is the allele which influences the appearance of the phenotype only in the presence of another identical allele.

F_1 generation. This is the generation produced by crossing homozygous parental stocks.

F_2 generation. This is the generation produced by crossing two F_1 organisms.

Monohybrid inheritance and the principle of segregation

Monohybrid inheritance is the inheritance of a single characteristic determined by one gene.

Inheritance of pod colour in peas

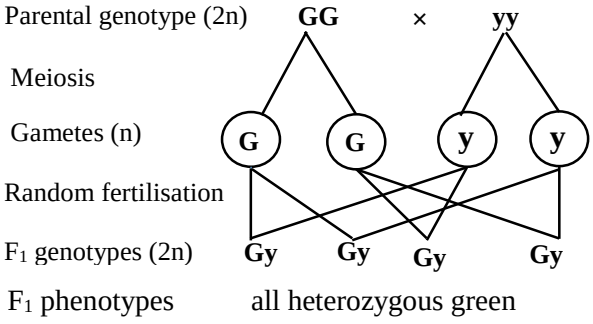
When pea plants with green pods are bred repeatedly with each other and they consistently give rise to plants with green pods, they are called **pure breeding** for the character of green pods.

Pure breeding strains can be bred for almost any character. Organisms are always homozygous (i.e. they have two alleles that are the same) for that particular gene.

If these pure breeding green pod plants are then crossed with pure breeding yellow pod plants, all the offspring, known as the **first filial** or **F_1 generation**, turn out to produce green pods. This means that the allele for green pods is dominant to the allele for yellow pods, which is therefore recessive. When the heterozygous plants (Gg) of the F_1 generation are crossed with one another (F_1 intercross), the offspring (known as the **second filial** or **F_2 generation**) are always in an approximate ratio of 3 plants with green pods to each 1 plant with yellow pods.

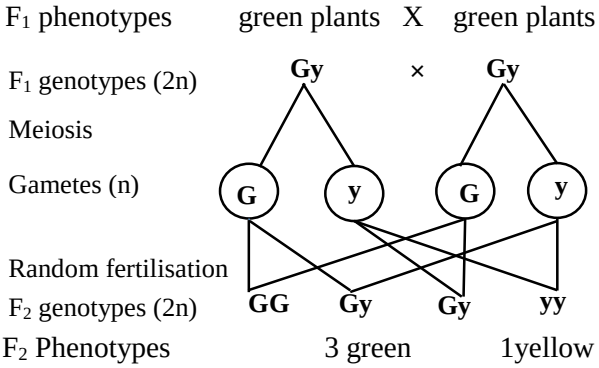
Let **G** represent allele for green colour (dominant)
Let **y** represent allele for yellow colour (recessive)

Parental phenotypes Green plants X yellow plants



F_1 phenotypes all heterozygous green

pollination of F_1 generation



The 3: 1 ratio of dominant phenotypes to recessive phenotypes is called the **monohybrid ratio**.

Mendel's conclusions

1. Since the original parental stocks were pure breeding, the character (colour) must have possessed two factors responsible for colour.
2. The F_1 generation possessed one factor from each parent which was carried by the gametes.
3. These factors do not blend in the F_1 generation but retain their individuality.
4. The green factor is dominant to the yellow factor, which is recessive.

The separation of the pair of parental factors, so that one factor is present in each gamete, is known as **Mendel's first law,** or the **principle of segregation**.

This states that: In diploid organisms, characteristics are determined by factors that occur in pairs, only one of each pair of factors can be present in a single gamete.

These factors determining characteristics, such as flower position, are regions of the chromosome known as **genes**. By convention, the initial letter of the dominant characteristic is used as the symbol for the gene and its capital form (e.g. A) represents the dominant form of the gene (the dominant allele) while the lower case (e.g. **a**) represents the recessive allele.

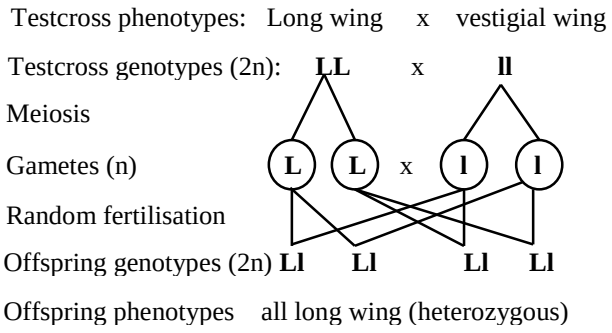
Test cross. This is a genetic cross between a homozygous recessive individual and a corresponding suspected heterozygote to determine its genotype.

Example in the fruit fly, *Drosophila*, the long wing is dominant to the vestigial wing. The genotype of a long wing *Drosophila* may be homozygous (**LL**) or heterozygous (**Ll**). In order to establish the correct genotype, the fly is test crossed with a double recessive (**ll**) vestigial wing fly. If the test cross offspring are all long-winged, the unknown genotype is homozygous dominant. A ratio of 1 long wing: l vestigial wing indicates that the unknown is heterozygous.

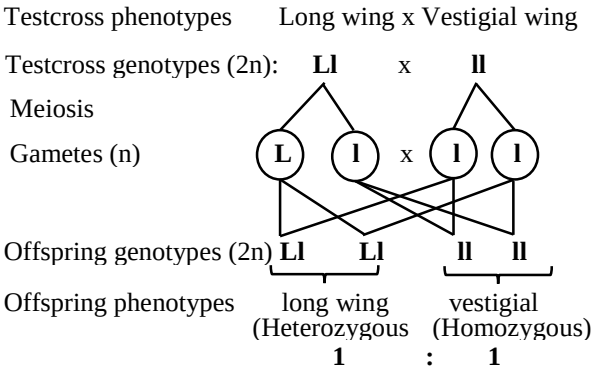
A full genetic illustration of how to determine the genotype of an organism showing a dominant characteristic

Let: **L** represent allele for long wing
l represent allele for vestigial wing

Homozygous long wing parent



Heterozygous long wing parent



Monohybrid inheritance in humans

The following conditions in humans are inherited in normal Mendelian fashion.

Albinism. This is a condition in which external pigment fails to develop, resulting in the person having light skin, white hair and pink eyes. It is caused by a recessive gene (**a**) and exerts its effects in the homozygous state (**aa**). The allele for normal skin (**A**) is dominant. The genotype of a normal person is **AA** or **Aa** and albino **aa**. Because the character is controlled by a recessive allele (a), individuals with genotype **Aa** are referred to as **carriers**.

Cystic fibrosis. This is a disorder of the mucus-secreting cells of the pancreas and other organs. It is caused by a recessive allele which is transmitted in the normal Mendelian fashion. The condition is usually fatal before the individual reach adulthood.

Achondroplasia. This is characterised by shortened and deformed legs and arms. It is brought about by a dominant allele. It exerts its effects in both homozygous and heterozygous state.

Others include;

- haemophilia,
- huntington's disease,
- lactose intolerance,
- phenylketonuria, and
- rhesus blood groups.

Dihybrid inheritance and the principle of independent assortment

This is the simultaneous inheritance of two characters. Mendel using pea shape and pea cotyledon colour as the characteristics, crossed pure- breeding (homozygous) plants having round and yellow peas with pure-breeding plants having wrinkled and green peas. The F_1 generation seeds were round and yellow. Self-pollination of the F_1 plants produced a variety of characteristics. He collected a total of 556 seeds from the F_2 generation which showed the following characteristics:
315 round and yellow, 101 wrinkled and yellow, 108 round and green, 32 wrinkled and green.

The proportions of each phenotype approximated to a ratio of 9:3:3:1. This is known as the **dihybrid ratio**.

Let

R represents round seed (dominant) Y represent yellow seed (dominant)
r represents wrinkled seed (recessive) y represent green seed (recessive)
Parental phenotypes: Round seed and yellow seed (homozygous) x wrinkled seed and green seed (homozygous)
Parental genotypes (2n): **RRYY** X **rryy**
Meiosis
Gametes (n) all **RY** X r**y**
Random fertilization:
F_1 genotypes (2n) all **RrYy**
F_1 phenotypes: All heterozygous round and yellow seeds

Intercrossing F_1 offspring

F_1 phenotypes round and yellow seed X round and yellow seed
F_1 genotypes (2n) **RrYy** X **RrYy**
Meiosis Segregation with independent assortment
Gametes (n) **RY** **Ry** **rY** **ry** X **RY** **Ry** **rY** **ry**

Punnet square showing random fusion of F_1 gamete

	gametes	**RY**	**Ry**	**rY**	**ry**
Random fertilization	**RY**	**RRYY** Round yellow	**RRYy** Round yellow	**RrYY** Round yellow	**RrYy** Round yellow
	Ry	**RRyY** Round yellow	**RRyy** Round green	**RrYy** Round yellow	**Rryy** Round green
F_2 genotypes(2n)	**rY**	**RrYY** Round yellow	**RrYy** Round yellow	**rrYY** wrinkled yellow	**rrYy** wrinkled yellow
	ry	**RrYy** Round yellow	**Rryy** Round green	**rrYy** wrinkled yellow	**rryy** wrinkled green

F_2 genotypes: 9 round yellow: 3 round green: 3 wrinkled yellow: 1 wrinkled green seeds

Two deductions are made from the above observations.

1. Two new combinations of characteristics appeared in the F_2 generation: wrinkled and yellow, and round and green.
2. The ratios of each pair of **allelomorphic** characteristics (phenotypes determined by different alleles) appeared in the monohybrid ratio of 3:1, that is 423 round to 133 wrinkled, and 416 yellow to 140 green.

The two pairs of characteristics (seed shape and colour), whilst combining in the F_1 generation, separate and behave independently from one another in subsequent generations. This forms the basis of **Mendel's second law** or the **principle of independent assortment** which states that: *any one of a pair of characteristics may combine with either one of another pair.*

In another experiment

Pure breeding tall pea plants possessing coloured flowers were crossed with dwarf plants possessing white flowers.

F_1 generation. All tall pea plants had coloured flowers.

F_2 generation. 96 tall coloured; 31 tall white; 34 dwarfs coloured and 11 dwarf white. This gives an approximate ratio of 9:3:3:1.

The two pairs of characteristics, tall-dwarf and coloured white combine in every possible way.

Let **T** represent allele for tallness.

Let **t** represent allele for dwarf.

Let **C** represent allele for coloured plants

Let **c** represent allele for white plants

Parental phenotype Tall coloured X Dwarf white

Parental genotye (2n) **TTCC** X **ttcc**

Meiosis

Segregation Segregation

Gametes (n) all **TC** X **tc**

Random fertilisation

F_1 generation (2n) all **TtCc**

F_1 phenotype all tall coloured

When F_1 generation are self-pollinated (selfed)

Parental phenotypes Tall coloured X tall coloured

Parental genotypes (2n) **TtCc** X **TtCc**

meiosis Segregation with independent assortment

Gametes (n) **TC** **Tc** **tC** **tc** X **TC** **Tc** **tC** **tc**

Punnet square to show the fusion of the F_1 gametes

	Gametes	**TC**	**Tc**	**tC**	**tc**
Random fertilisation	**TC**	**TTCC** Tall coloured	**TTCc** Tall coloured	**TtCc** Tall coloured	**TtCc** Tall coloured
	Tc	**TTCc** Tall coloured	**TTcc** Tall white	**TtCc** Tall coloured	**Ttcc** Tall white
F_2 generation (2n)	**tC**	**TtCC** Tall coloured	**TtCc** Tall coloured	**ttCC** dwarf coloured	**ttCc** dwarf coloured
	Tc	**TtCc** Tall coloured	**Ttcc** Tall white	**ttCc** dwarf coloured	**ttcc** dwarf white

F_2 phenotypes 9 tall coloured 3 Tall white 3 Dwarf coloured 1 Dwarf white

Test cross

This is used to establish the genotype of tall plants by crossing it with dwarf white plants in this case (ttcc)

Parental phenotypes tall coloured x dwarf white

Parental genotypes (2n) **TtCc** x **ttcc**

meiosis

Gametes (n) **TC** **Tc** **tC** **tc** X all **tc**

Punnet square to show fusion of gamets in the test cross

	Gametes	**TC**	**Tc**	**tC**	**tc**
Random fertilisation	**tc**	**TtCc** 1 Tall coloured	**Ttcc** 1 Tall white	**ttCc** 1 Dwarf colured	**Ttcc** 1 Dwarf white

Phenotypic ratio of the test cross offsprings is 1: 1: 1: 1

Mendel's second law: The law of independent assortment

Each of the two alleles of one gene may combine randomly with either of the alleles of another gene.

Explanation. For independent assortment, the genes concerned are carried on different chromosomes. During metaphase I, homologous chromosomes line up side by side on the spindle prior to separating. Different pairs of homologous chromosomes behave independently of each other. The way one pair of homologous chromosomes arranges themselves on the spindle and subsequently separate has no effect on the behaviour of any other pair of chromosomes.

Summary of Mendel's hypotheses

1. Each characteristic of an organism is controlled by a pair of alleles.
2. If an organism has two unlike alleles for a given characteristic, one may be expressed (the dominant allele) to the total exclusion of the other (the recessive allele)
3. During meiosis, each pair of alleles separates (segregates) and each gamete receives one of each pair of alleles *(the principle of segregation).*
4. During gamete formation in each sex, either one ofapair of alleles may enter the same gamete cell (combine randomly) with either one of another pair *(the principle of independent assortment).*
5. Each allele is transmitted from generation to generation as a discrete unchanging unit.
6. Each organism inherits one allele (for each characteristic) from each parent.

NB: The mechanism of dihybrid inheritance and the typical dihybrid ratio of 9:3:3:1 only apply to characteristics controlled by genes on different chromosomes. Genes situated on the same chromosome may not show this pattern of independent assortment.

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

Linkage

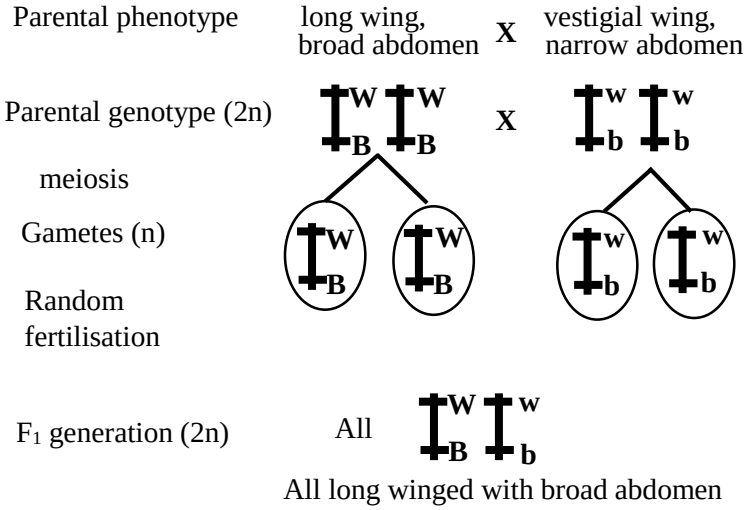
In Drosophila, the allele for the broad abdomen is dominant over the allele for the narrow abdomen, just as the allele for longwing is dominant to the allele for the vestigial wing.

When the long-winged broad fly is crossed with a vestigial-winged, narrow fly. All the F_1 offspring have long wings and broad abdomens. When two flies from the F_1 generation are mated. The F_2 generation fails to produce the 9:3:3:1 ratio. Instead, the offspring are mainly long-winged, broad flies and vestigial, narrow flies in the proportion of 3:1.

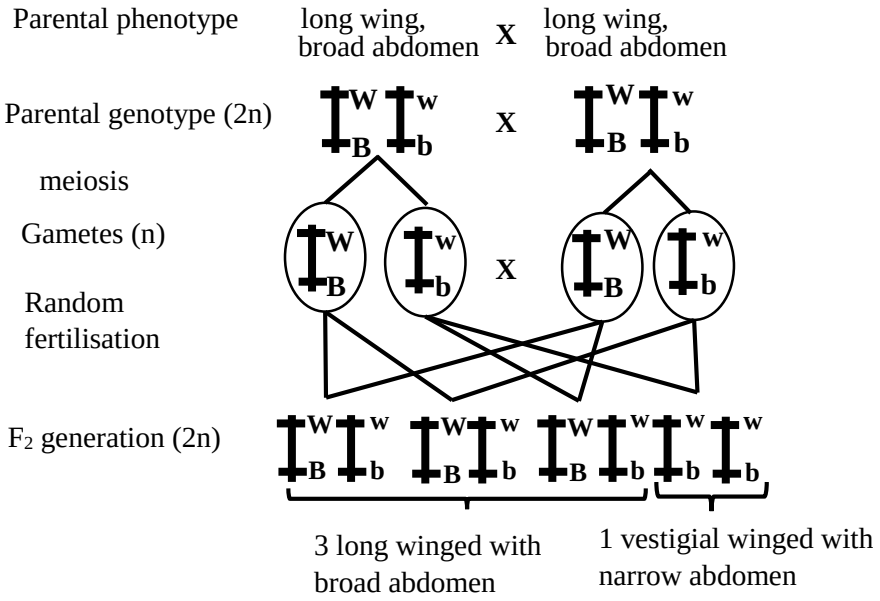
This takes place because the genes determining the length of the wings and the width of the abdomen are located on the same chromosome, resulting in their being transmitted together. These are called **linked genes**. Independent assortment does not take place during this situation.

Individual chromosomes usually contain a very large number of genes controlling a wide variety of characteristics. These genes linked together on the same chromosome for a **linkage group**.

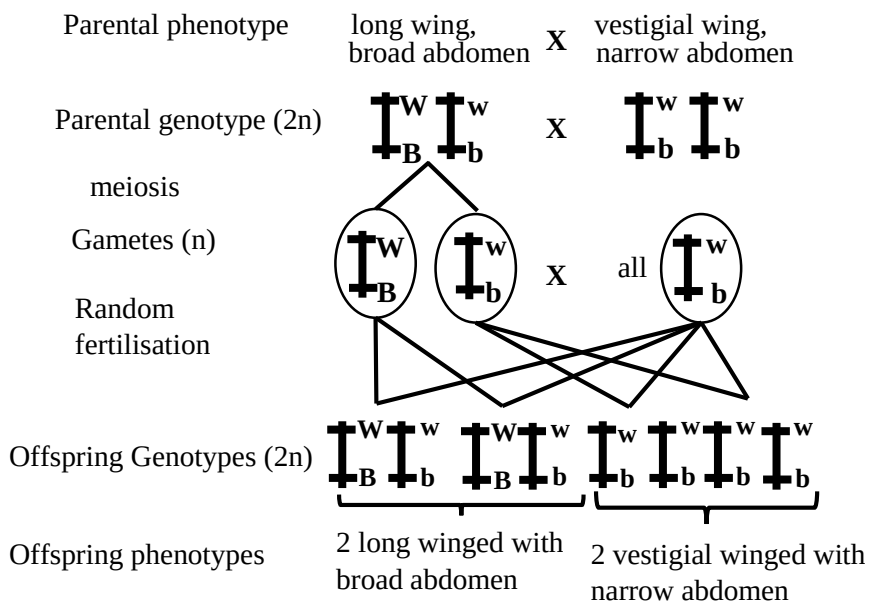
Let **W** represent allele for the long wing.
Let **w** represent allele for the vestigial wing.
Let **B** represent allele for the broad abdomen.
Let **b** represent allele for the narrow abdomen.



Selfing F_1 generation



Test cross of F_1 offspring during linkage



Offspring phenotypic ratio is 1:1

In practice, the F_2 3 : 1 ratio and the 1:1 test cross-ratio is never achieved. Four phenotypes are invariably produced. This is because the **total** linkage is rare. Most breeding experiments involving linkage produce approximately equal numbers of the parental phenotypes and a significantly smaller number of phenotypes showing new combinations of characteristics, also in equal numbers. These latter phenotypes are described as **recombinants**.

Crossing over

Complete linkage does not exist. A certain proportion of the offspring shows new combinations as in independent assortment.

In maize. When a maize plant homozygous for kernels which are coloured and smooth is crossed with one having colourless and shrunken kernels. F_1 offsprings all have coloured and smooth kernels.

When F_1 offspring is test crossed with double recessive.

- ✓ Four combinations are expected to be obtained in equal numbers. This happens when genes are on separate chromosomes.
- ✓ Two types of kernels are supposed to be produced if the alleles are carried on the same chromosome, i.e coloured smooth and colourless shrunken.

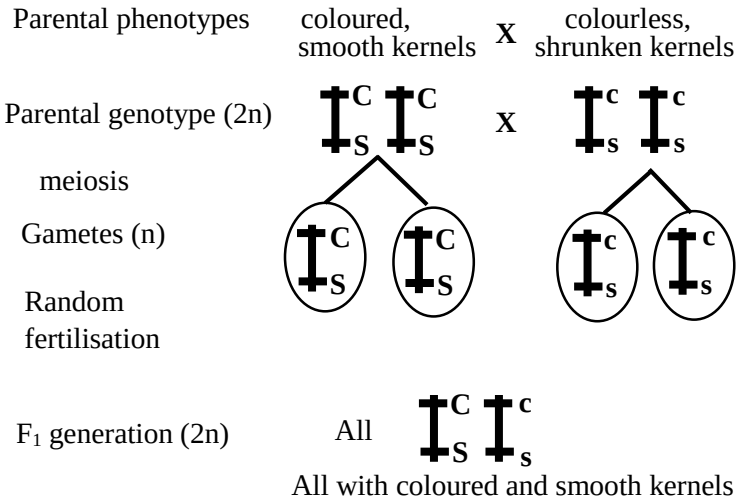
Neither of the above happens. The results obtained are coloured smooth and colourless shrunken kernels, some few coloured shrunken and colourless smooth ones.

All combinations are obtained but not in equal proportions.

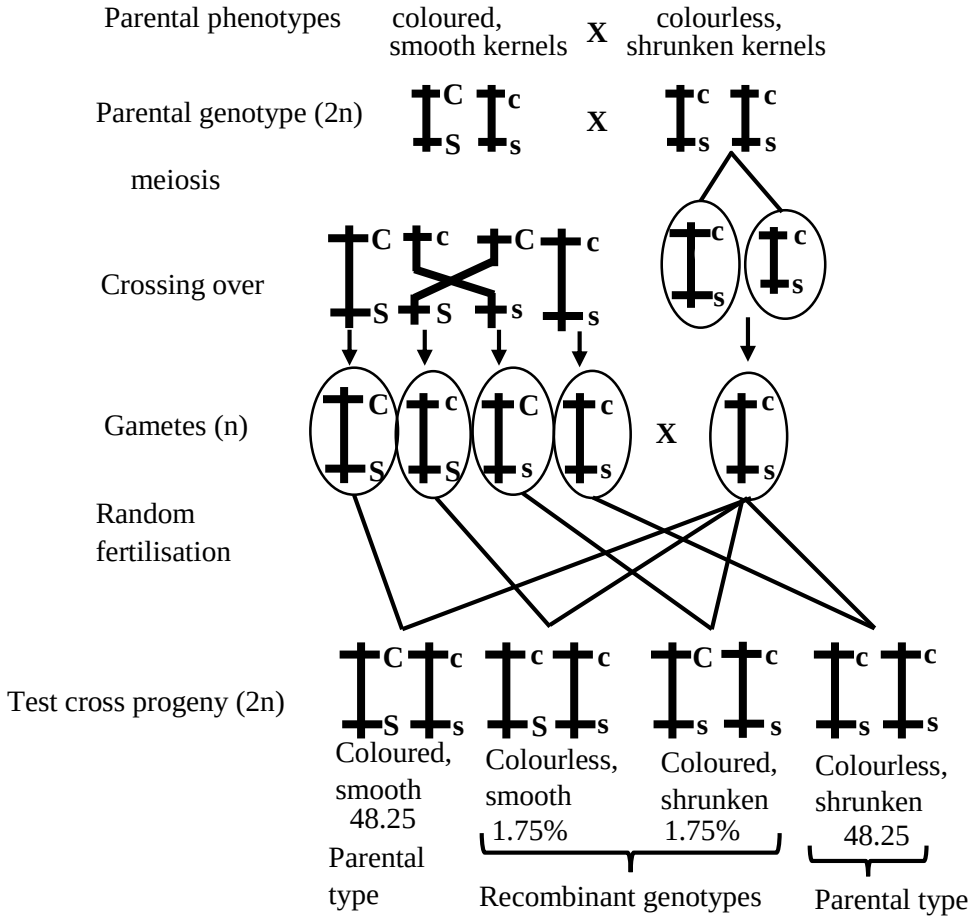
This is because the genes are located on the same chromosome however, during gamete formation some changes take place. This is called **crossing over** and it leads to separation of linked genes which allows them to recombine. A few show new combinations and they are known as **recombinants**.

Let **C** represent allele for coloured kernels.
Let **c** represent allele for colourless kernels.
Let **S** represent allele for smooth kernels.

Let **s** represent allele for shrunken kernels.



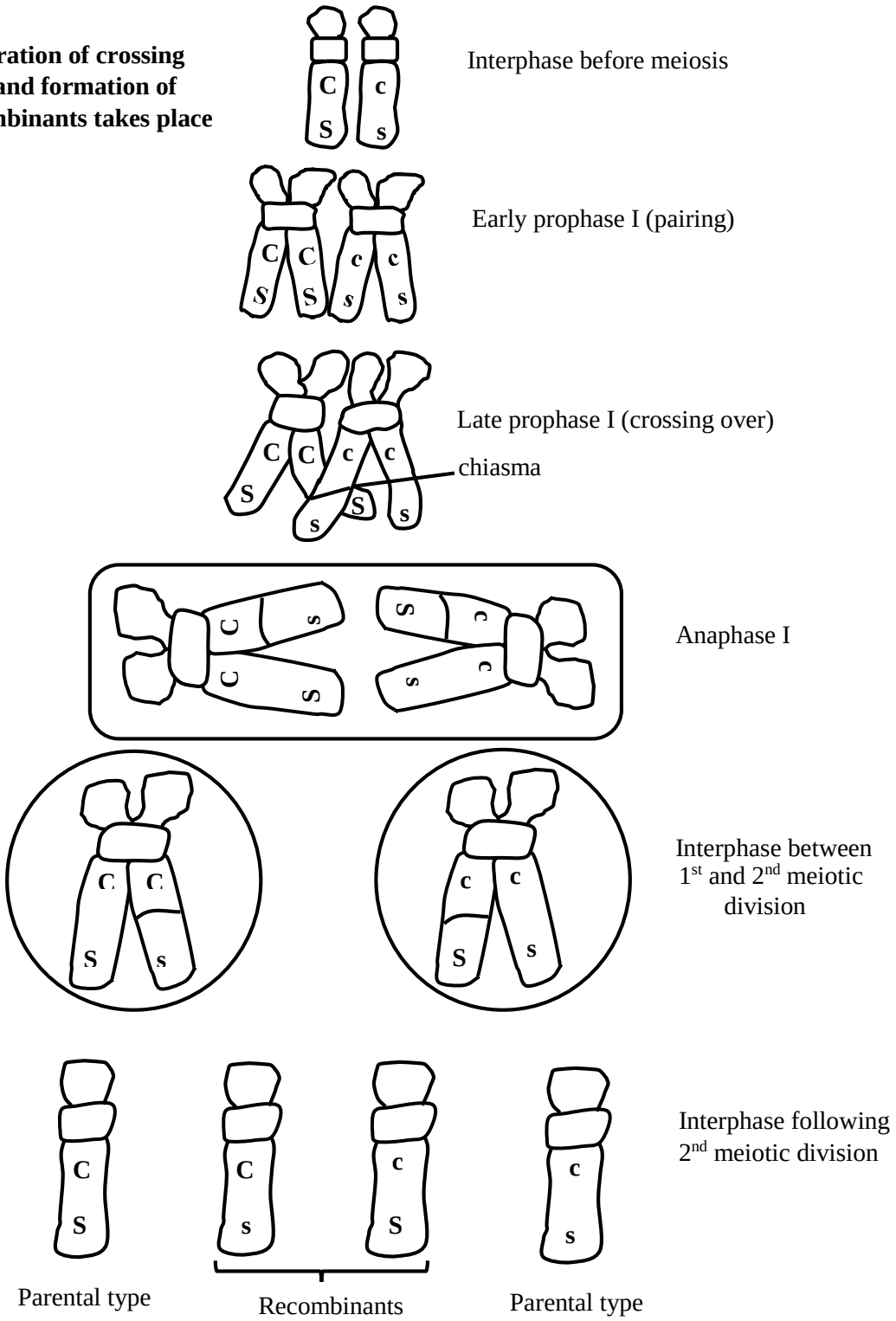
Test crossing F_1 generation



In meiosis I homologous chromosomes become intimately wrapped around one another. They become intertwine and the non-sister chromatids of the homologous chromosomes exchange portions at a point called chiasma. This is called crossing over. The chromatids finish up in separate gametes and after fertilization give rise to new combinations of alleles in the offspring (recombinants).

The number of chiasmata formed in a bivalent during meiosis due to the length of the chromosomes is proportional to the number of possible combinations. This causes genetic variety.

Illustration of crossing over and formation of recombinants takes place



Using the figures obtained from crossing over, it is possible to calculate the recombination frequency of the genes.

The **recombination frequency** is calculated using the formula:

$$\frac{number\ of\ individual\ showing\ recombination}{number\ of offspring}\ X\ 100$$

From the example above the recombination frequency (%) is

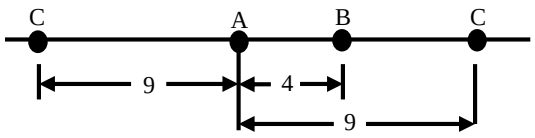
$$\frac{(1.75+1.75)}{(48.25+48.25)+(1.75+1.75)}\ \text{X}\ 100: \frac{3.5}{100}\ X\ 100 = 3.5\%$$

Crossover frequency reflects the relative positions of genes on chromosomes because the further apart linked genes are on the chromosomes, the greater the possibility of crossing-over occurring between them that is the greater the crossover frequency.

Gene mapping

This is the method used to identify the locus of a gene and the distances between genes. It shows the relative positions of genes on chromosomes. Chromosome maps are constructed by directly converting the crossover frequency or value between genes into hypothetical distances along the chromosome. A crossover frequency or value (COV) of 4% between genes A and B means that those genes are situated 4 units apart on the same chromosome. A COV of **9%** for a pair of genes A and C would indicate that they were 9 units apart, but it would not indicate the linear sequence of the genes.

In practice, it is usual to determine crossover values for at least three genes at once, as this **triangulation** process enables the sequence of the genes to be determined as well as the distance between them.



Possible gene loci of A, Band C on basis of the data presented

Calculate the sequence and distances apart of the genes for the following.

P-Q = 24%

R-P = 14%

R-S = 8%

S-P = 6%

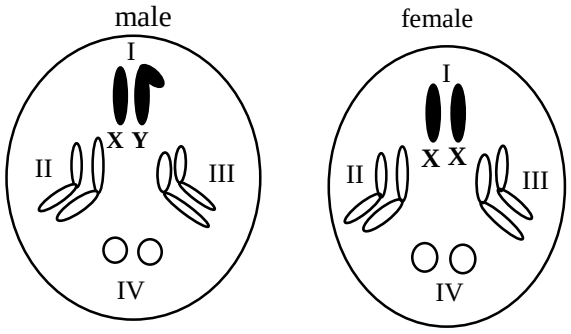
Linkage groups and chromosomes

Drosophila melanogaster has four linkage groups and four pairs of chromosomes. The number of linkage groups found in a particular species corresponds to the number of different types of chromosome (the haploid number) characteristic of the species. In maize, there are 10 pairs of chromosomes and 10 different linkage groups. Genes in any one linkage group are transmitted together but independently of the genes in the other linkage groups.

A linkage group which contains a large number of genes appears large compared to the one carrying few genes. Drosophila melanogaster has two large linkage groups, one which is medium and one which is very small. They are numbered I to IV.

The medium-sized chromosomes in *Drosophila melanogaster* determine sex. They are referred to as the **sex chromosomes.** Females have two rod-shaped identical chromosomes known as **X chromosome** of the genotype **XX**. Males have the two sex chromosomes which are different. One is a rod-shaped **X** chromosome, and the other is a hook-shaped chromosome called the **Y chromosome**. The male genotype has a genotype of **XY**. The same applies to humans but the **Y** chromosome in humans is smaller than the **X** chromosome and is not hooked.

Sex chromosomes are not always identical as other homologous chromosomes. They are different in appearance and referred to as **heterosomes**. Other pairs which are identical in appearance are called **autosomes**.



The females produce only one kind of gamete containing **X** chromosome and they are therefore referred to as

homogametic. The male produces two kinds of gametes, half the sperms produced contain the **X** chromosome and the other half contains **Y** chromosome. The male is therefore **heterogametic**. During random fusion of gametes, half the zygotes receive two **X** chromosomes and give rise to females; the other half receive an **X** and **Y** chromosome and give rise to males.

NB: In birds the male is **XX** and the female is **XY**. In some insects the female of **XX** and the male is **XO**, the Y chromosome is absent.

Sex determination in man

In humans, there are 23 pairs of chromosomes within the cell, each pair of chromosomes is structurally the same except in only one pair, the 23rd pair of the homologous chromosomes, which show structural differences in males and females.

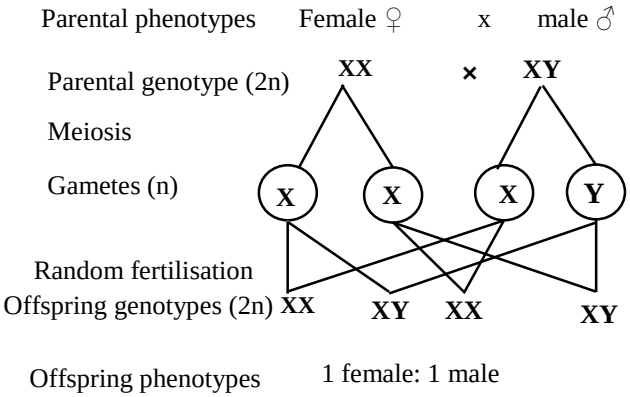
In the production of gametes, the sex chromosomes segregate in typical Mendelian fashion. For example, in mammals each ovum contains an **X** chromosome; in males, one half of the sperm contains an **X** chromosome and the other half contain a **Y** chromosome. The sex of the offspring depends upon which type of sperm fertilises the ovum. Females have **XX** genotype (homogametic) whereas males have **XY** genotype (heterogametic).

The function of the **Y** chromosome appears to vary according to the species. In humans, the presence of a **Y** chromosome controls the differentiation of the testes which subsequently influences the development of the genital organs and male characteristics. In some organisms, however, the **Y** chromosome does not carry genes concerned with sex. It is described as genetically inert or genetically empty since it carries so few genes. In *Drosophila*, the **Y** chromosome is usually empty.

In humans, males receive their **Y** chromosome from the father and therefore they cannot inherit their fathers' sex-linked traits. A female receives an **X** chromosome from the father which she transmits to her children. The children show their grandfather's sex-linked traits. Therefore, a male transmits his sex-linked traits to his grandchildren via his daughters, he cannot transmit them to or through his sons.

Let XX represent female chromosomes

Let XY represent male chromosomes



All genes carried on the sex chromosomes are transmitted along with those determining sex. They are therefore referred to as **sex-linked genes**. They include inheritance of eye colour in *Drosophila*. There are red-eyed and white-eyed strains. Red-eye is dominant over the white.

Examples of sex-linked traits in humans include

- ✓ Red-green **colourblindness**. The allele is recessive.
- ✓ **Haemophilia** or 'bleeder's disease'. The blood takes an abnormally long time to clot, resulting in profuse and prolonged bleeding from even slight wound. It is caused by a recessive allele.

In *Drosophila*, the genes determining male characteristics are carried on the autosomes and their phenotypic effects are masked by the presence of a pair of X chromosomes. Male characteristics, on the other hand, appear in the presence of a single X chromosome. This is an example of **sex-limited inheritance,** as opposed to sex-linked inheritance, and in humans is thought to cause suppression of the genes for the growth of a beard in females.

Sex linkage

Genes carried on the sex chromosomes are called sex-linked. They are inherited together with sex. Examples of sex-linked (X linked) traits in humans include,

- ➢ Haemophilia or Bleeder's disease.
- ➢ Red-green colour blindness.
- ➢ Muscular dystrophy/ Duchenne muscular dystrophy (DMD).

- Premature balding.

Muscular dystrophy/ Duchenne muscular dystrophy (DMD). This is a tragic, wasting disease affecting infant children mainly boys, where muscles of the body are replaced by fibrous tissue, resulting in progressive body weakness and difficulty in breathing. This disease can result in the 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 fibre. The person is restricted to a wheelchair.

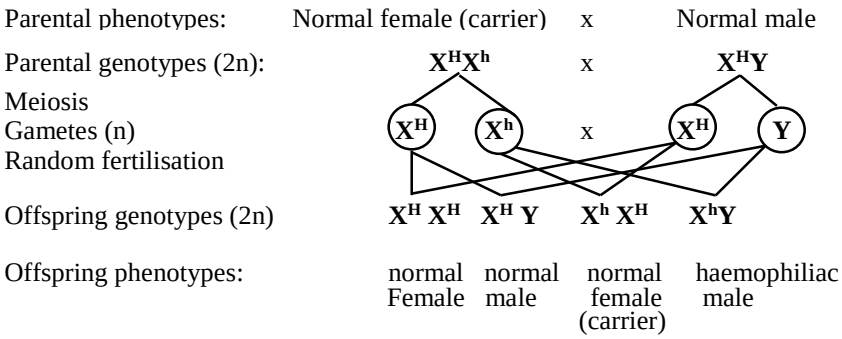
Red-green colour blindness. This is a condition in which a person cannot distinguish between red and green colours. It is caused by recessive allele carried on the X chromosome.

Haemophilia or 'bleeder's disease' is a sex-linked recessive condition which prevents the formation of clotting factor VIII. This delays the clotting process. The gene for factor VIII is carried on the non-homologous portion of the X chromosome and can appear in two allelomorphic forms: normal (dominant) and mutant (recessive). The following possible genotypes and phenotypes can occur:

Genotype	**Phenotype**
X^HX^H	Normal female
X^HX^h	Normal female (carrier)
X^HY	Normal male
X^h	Haemophiliac male

In all sex-linked traits, females who are heterozygous are described as **carriers** of the trait. They are phenotypically normal but half their gametes carry the recessive gene.

Let
H represent normal allele for blood clotting (dominant).
h represent allele for haemophilia (recessive).
XX represent female chromosomes.
XY represent male chromosomes.



Mechanism for the inheritances of sex-linked allele for Haemophilia

Red and white eyes in *Drosophila* is another sex-linked (X linked) trait.
In ***Drosophila*** there are red-eyed and white-eyed strains, red-eye being dominant to white. The result of crossing a red-eyed fly with a white one depends on which parent is red and which is white. If the father is white the F_1 gives nothing but red-eyed flies, males and females being in equal proportions.

Let: **R** represent allele for red colour
r represent allele for white colour
XX represent female organism
XY represent male organism

Parental phenotypes Red-eyed ♀ x white-eyed♂

Parental genotypes (2n) X^RX^R x X^rY

Meiosis

Gametes (n) X^R X^R x X^r Y

Random fertilisation

F_1 genotypes (2n) X^RX^r X^RY X^RX^r X^RY

F_1 phenotypes 1 Red-eyed ♀ : Red-eyed♂

Parental phenotypes white-eyed ♀ x Red-eyed ♂

Parental genotypes (2n) X^rX^r x X^RY

Meiosis

Gametes (n) X^r X^r x X^R Y

Random fertilisation

F_1 genotypes (2n) X^RX^r X^rY X^RX^r X^rY

F_1 phenotypes 1 Red-eyed ♀: 1white-eyed♂

Complete and incomplete dominance

Complete dominance is the inheritance of contrasting characteristics where one gene is dominant over another gene which is recessive, and the organisms produced show distinct phenotypes with no intermediate characteristics. The offspring produced, therefore, belong to one or both of the parental types without any intermediates.

Incomplete dominance

This is a condition whereby some genes don't show complete dominance; one gene is neither dominant nor recessive over the other.

The F_1 hybrids have phenotypes somewhere in between the two parental varieties. This phenomenon occurs when red snapdragons are crossed with white snapdragons: All the F_1, hybrids have **pink flowers**.

Incomplete dominance is found in both plants and animals. In genetics, genes responsible for incomplete dominance are represented by capital letters bu of different kinds.

An example is the production of blue Andalusian fowls by crossing pure-breeding black and splashed white parental stocks. The presence of black plumage is the result of the possession of an allele for the production of the black pigment melanin. The splashed white stock lack this allele. The heterozygotes show a partial development of melanin which produces a blue sheen in the plumage.

The results of a cross between black and splashed white homozygous fowl.

If the F_1 generation is allowed to interbreed, the F_2 generation shows a modification of the normal Mendelian phenotypic monohybrid ratio of 3:1. In this case, a phenotypic ratio of 1:2:1 is produced where half the F_2 generation has the F_1 genotype. This ratio of 1:2:1 is characteristic of examples of incomplete dominance.

Example 1

Let **B** represent the black allele.
Let **W** represent the splashed white allele

Parental phenotypes Black x splashed white

Parental genotypes (2n) **BB** x **WW**

Meiosis

Gametes (n) **B** **B** X **W** **W**

Random fertilisation

F_1 genotypes (2n) **BW** **BW** **BW** **BW**

F_1 phenotypes all blue heterozygotes

Production of F_1 hybrids of Andalusian fowl

Selfing F_1 offspring

F_1 phenotypes Blue x Blue

F_1 genotypes (2n) **BW** x **BW**

Meiosis

Gametes (n) **B** **W** X **B** **W**

Random fertilisation

F_2 genotypes (2n) **BB** **BW** **BW** **WW**

F_2 phenotypes 1 black 2 blue 1 splashed white

Production of F_2 hybrids of Andalusian fowl.

Example 2

Let: **R** represent allele for red colour
W represent allele for white colour

Parental phenotypes Red flowers x white flowers

Parental genotypes (2n) **RR** x **WW**

Meiosis

Gametes(n) **R** **R** x **W** **W**

Random fertilisation

F_1 genotypes(2n) **RW** **RW** **RW** **RW**

F_1 phenotypes All pink

Intercrossing F_1 offspring

F_1 phenotypes Pink flowers x pink flowers

F_1 genotypes (2n) **RW** x **RW**

Meiosis

Gametes (n) **R** **W** X **R** **W**

Random fertilisation

F_2 genotypes (2n) **RR** **RW** **RW** **WW**

F_2 phenotypes 1 red 2 pink 1 white

Degree of Dominance

1. Snapdragon fly with red flowers crossed with white gives plants with pink flowers.

2. Shorthorn cattle crossing individuals with red and white coats give offspring whose coats are composed of a mixture of red and white hairs (roan condition)

3. In Andalusian breed of fowls. A black fowl crossed with a splashed-white one (white with small black patches on it) gives chicks with a blue sheen, the true Andalusian condition.

Between the two extremes of complete dominances and no dominance at all are all shades of partial dominance. The offspring fail to resemble either parent exactly, but closer to one than the other.

Gene interaction

Sometimes a single characteristic is controlled by the alleles of two or more genes interacting with one another. A characteristic which is controlled by more than one gene is known as a **polygenic character** and its transmission is called polygenic inheritance. There are many situations when genes interact to control phenotypic characteristics of organisms, these include;

- Codominance
- Multiple alleles
- Lethal genes.
- Gene complex/simple gene interactions/ complementary genes.
- Epistasis.
- Pleiotropy.

Multiple alleles. There are conditions where a single character may appear in several different forms controlled by three or more alleles, of which any two may occupy the same gene loci on homologous chromosomes. This is known as the **multiple allele** (or **multiple allelomorphs**) conditions and it controls such characteristics as coat colour in mice, eye colour in mice and blood group in humans.

Inheritance of blood groups

Blood group is controlled by an autosomal gene. The gene locus is represented by the symbol **I** (which stands for **isohaemagglutinogen**) and there are three alleles represented by the symbols **A**, **B** and **o.** The alleles **A** and **B** are equally dominant and **o** is recessive to both. The presence of a single dominant allele results in the blood-producing a substance called **agglutinin** which acts as an antibody. Genotype $\mathbf{I^AI^\circ}$ would give rise to the agglutinogen **A** on the red blood cell membrane, and the plasma would contain the agglutinin **anti-B** (the blood group would be **A**).

Table: Human blood groups genotypes

Genotype	Blood group (phenotype)
$\mathbf{I^AI^A}$	**A**
$\mathbf{I^AI^B}$	**A**
$\mathbf{I^BI^B}$	**B**
$\mathbf{I^BI^O}$	**B**
$\mathbf{I^AI^B}$	**AB**
$\mathbf{I^OI^O}$	**O**

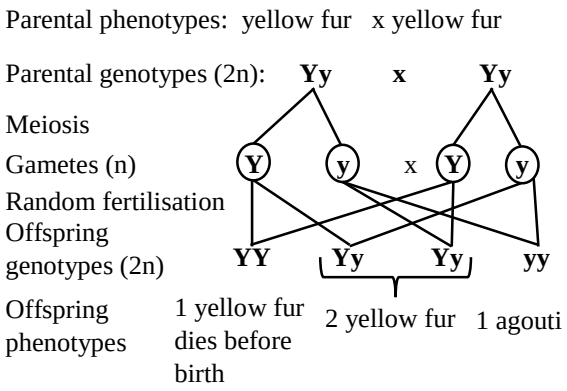
Lethal genes. These are genes that in some (such as homozygous) conditions may prevent the development or cause the death of an organism or its germ cells. They are usually a result of mutations in genes that are essential for growth and development. Lethal alleles may be recessive, dominant, or conditional depending on the gene or genes involved.

In humans and other mammals, a certain recessive gene leads to internal adhesion of the lungs resulting in death at birth. Another example involving a single gene affects the formation of cartilage and produces congenital deformities leading to fetal and neonatal death.

In chickens which are homozygous for an allele controlling feather structure called 'frizzled', several phenotypic effects result from the incomplete development of the feathers. These chickens lack adequate feather insulation and suffer from heat loss. To compensate for this, they exhibit a range of structural and physiological adaptations, but these are largely unsuccessful and there is a high mortality rate.

Lethal genes also occur in mice during the inheritance of fur colour. Wild mice have grey- coloured fur, a condition known as **agouti**. Some mice have yellow fur. Cross-breeding yellow mice produces offspring in the ratio 2 yellow fur: 1 agouti fur. Yellow is dominant to agouti and that all the yellow coat mice are heterozygous. From Mendelian ratio of 3: 1, fetal death of homozygous yellow coat mice takes place. Examination of the uteri of pregnant yellow mice reveals dead yellow fetuses. Examination of the uteri of crosses between yellow fur and agouti fur mice reveals no dead yellow fetuses.

Let **Y** represent allele for yellow fur (dominant)
Let **y** represent allele for agouti fur (recessive)

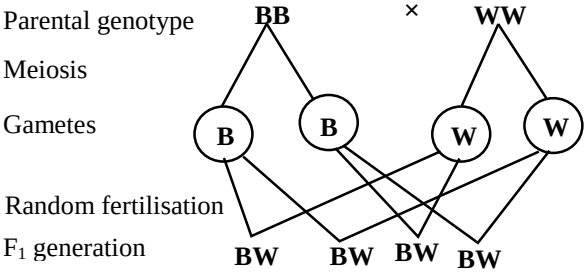


Codominance. This is the condition where two alleles affect the phenotype in separate distinguishable ways. It is a condition whereby the genes controlling contrasting characteristics are neither dominant nor recessive over each other. The F_1 offspring does not show intermediate characteristic but instead both characteristics of the two pure line parents express itself independent and both characteristics appear in the offspring. Like **AB** blood system. The **A** and **B** alleles show equal dominance with respect to one another (co-dominance) but both are dominant to **o**.

A person with the genotype **AA** or **Ao** belongs to blood group **A**.
A person with the genotype **BB** or **Bo** belongs to blood group **B**.
A person with the genotype **AB** belongs to blood group **AB**.
A person with the genotype **oo** belongs to blood group **o**.

The genetic crosses of codominance condition are done like for incomplete dominance, where the two genes are represented with capital letters of different kinds.

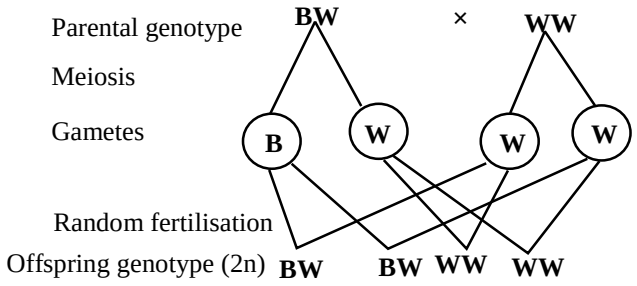
In a monohybrid cross, a male rabbit homozygous for brown hair was matted with a female rabbit homozygous for white hair. Each one of the F_1 offspring had a mixture of brown and white hair.
Let **B** represent the allele for brown hair
Let **W** represent the allele for white hair
Parental phenotype: Brown hair × White hair



F_1 phenotype: All brown and white hair mixture
F_1 genotype: All **BW**

(ii) Supposing there was crossing between the F_1 male and the female brown rabbit, what will be the phenotype and genotype of the offspring.

Parental phenotype: Brown &White hair × White hair



F_1 phenotype: 2 brown and white: 2 white

Partial dominance. Offspring fail to resemble either parent exactly but are closer to one than the other. It occurs between the two extremes of complete dominance and no dominance at all. Alleles do not interact in an all-or- anything manner but shows varying degrees of intermediate expression. There are more blends of partial dominance which led to a wide range of intermediate varieties between two extremes

Gene complex (Complementary genes)

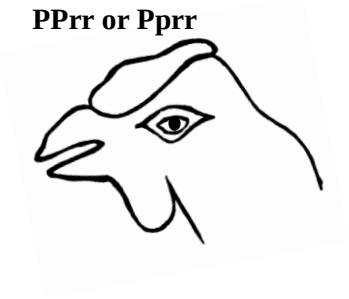
This is the interaction between several genes of an organism to determine certain characteristics. A single characteristic may be controlled by the interaction of two or more genes situated at different loci.

For example, during the inheritance of the shape of the comb in domestic fowl, there are genes at two loci situated on different chromosomes which interact and give rise to four distinct phenotypes, known as **pea**, **rose**, **walnut** and **single** combs.

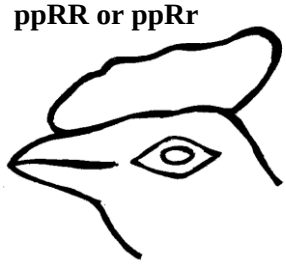
The appearance of the pea comb and rose comb are each determined by the presence of their respective dominant allele (P or R) and the absence of the other dominant allele. Walnut comb results from a modified form of codominance in which at least one dominant allele for pea comb and rose comb is present (that is PR). Single comb appears only in the homozygous double recessive condition (that is **pprr**).

Phenotypes and possible genotypes associated with comb shape in poultry	
Phenotype	Possible genotypes
Pea	**PPrr, Pprr**
Rose	**RRpp, Rrpp**
Walnut	**PPRR, PpRR, PPRr, PpRr**
Single	**pprr**

pea comb
PPrr or Pprr

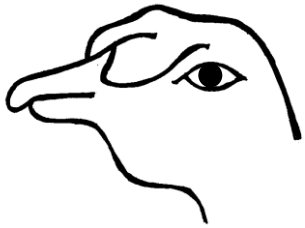


rose comb
ppRR or ppRr



Single Comb pprr

walnut comb PPRR, PPRr, PpRR or PpRr



Let:
P represent presence of pea comb (dominant)
p represent absence of pea comb (recessive)
R represent presence of rose comb (dominant)
r represent absence of rose comb (recessive)

Parental phenotypes Pea comb x rose comb

Parental genotypes (2n) **PPrr** x **RRpp**

Meiosis

Gametes (n) all **Pr** x **Rp**

Random fertilisation

F_1 genotypes (2n) all **PpRr**

F_1 phenotypes all walnut

Selfing F_1 generation

F_1 phenotypes walnut comb x walnut comb

F_1 genotypes (2n) **PpRr** x **PpRr**

Meiosis

Gametes (n) **PR** **Pr** **pR** **pr** **X** **PR** **Pr** **pR** **pr**

Punnet square showing fusion of gametes

Gametes(n)		**PR**	**Pr**	**pR**	**pr**
Random fertilsation	**PR**	**PPRR**	**PPRr**	**PpRR**	**PrRr**
	Pr	**PPrR**	**PPrr**	**PrRr**	**Pprr**
F_2 genotypes(2n)	**pR**	**PpRR**	**PpRr**	**ppRR**	**ppRr**
	pr	**PpRr**	**Pprr**	**ppRr**	**pprr**

F_2 phenotypes: 9 walnut: 3 pea comb: 3 rose comb: 1 single comb

Epistasis

This is a condition where the presence of a gene suppresses the effect of another gene at another locus. Epistatic genes are sometimes called **inhibiting genes** because of their effect on the other genes which are described as **hypostatic**. It occurs when two or more different gene loci contribute to the same phenotype. One gene locus masks or modifies the phenotype of a second gene locus.

In mice and many other mammals, black coat colour is dominant (**B**) to brown (**b**). For a mouse to have brown fur, its genotype must be **bb**. But there is more to the story. A second gene determines whether or not pigment will be deposited in the hair. The dominant allele, symbolized by **C** (for colour), results in the deposition of either black or brown pigment, depending on the genotype at the first locus. But if the mouse is homozygous recessive for the second locus (**cc**), then the coat is white (albino), regardless of the genotype at the black/brown locus. In this case, the gene for pigment deposition is epistatic to the gene that codes for black or brown pigment.

Mating black mice that are heterozygous for both genes (**BbCc**) follows the law of independent assortment and produces 9:3:3:1 ratio. As a result of epistasis, the phenotypic ratio among the F_2 offspring is 9 black to 3 brown to 4 white. Other types of epistatic interactions produce different ratios, but all are modified versions of 9:3:3:1.

Let: B represent allele for black fur (dominant)
b represent allele for brown fur(recessive)
C represent coloured fur (dominant)
c represent albino fur (recessive)

Parental phenotypes black mouse x black mouse

Parental genotypes (2n) **BbCc** x **BbCc**

Meiosis

Gametes		**BC**	**bC**	**Bc**	**Bc**
Random fertilization	**BC**	BBCC **black**	BbCC **black**	BBCc **black**	BbCc **Black**
	bC	BbCC **black**	bbCC brown	BbCc **black**	bbCc brown
	Bc	BBCc **black**	BbCc **black**	BBcc abino	Bbcc albino
Offspring genotypes (2n)	**bc**	BbCc **black**	bbCc brown	Bbcc albino	,bbcc albino

Offspring phenotypes: 9 black: 3 brown: 4 albino

Pleiotropy

This is a condition where genes have multiple effects. In humans, for example, pleiotropic alleles are responsible for the multiple symptoms associated with certain hereditary diseases, such as cystic fibrosis and sickle-cell disease. In the garden pea, the gene that determines flower colour also affects the colour of the coating on the outer surface of the seed, which can be grey or white.

Polygenes

Some characters may be determined by many genes acting together. For example, a character may be determined by five genes each gene having a dominant or recessive allele. An organism inheriting five dominant alleles will lie at one end of the spectrum and one with five recessive alleles will lie at the other. Between these extremes is the continuum of types depending on the relative proportions of dominant and recessive alleles. Polygenes give rise to continuous variation.

PEDIGREE ANALYSIS

Pedigree charts are diagrams that show the phenotypes or genotypes for a particular organism, its ancestors, and descendants. They are commonly used in human families to track genetic diseases. They are also used for any species and any inherited trait. Geneticists use a standardized set of symbols to represent an individual's

sex, family relationships and phenotype. These diagrams are used to determine the mode of inheritance of a particular disease or trait, and to predict the probability of its appearance among offspring.

Pedigree analysis is, therefore, an important tool in basic research, agriculture, and genetic counselling.

Each pedigree chart represents all the available information about the inheritance of a single trait (most often a disease) within a family. The pedigree chart is therefore drawn using factual information, but there is always some possibility of errors in this information, especially when relying on family members' recollections or even clinical diagnoses. In real pedigrees, further complications can arise due to incomplete penetrance.

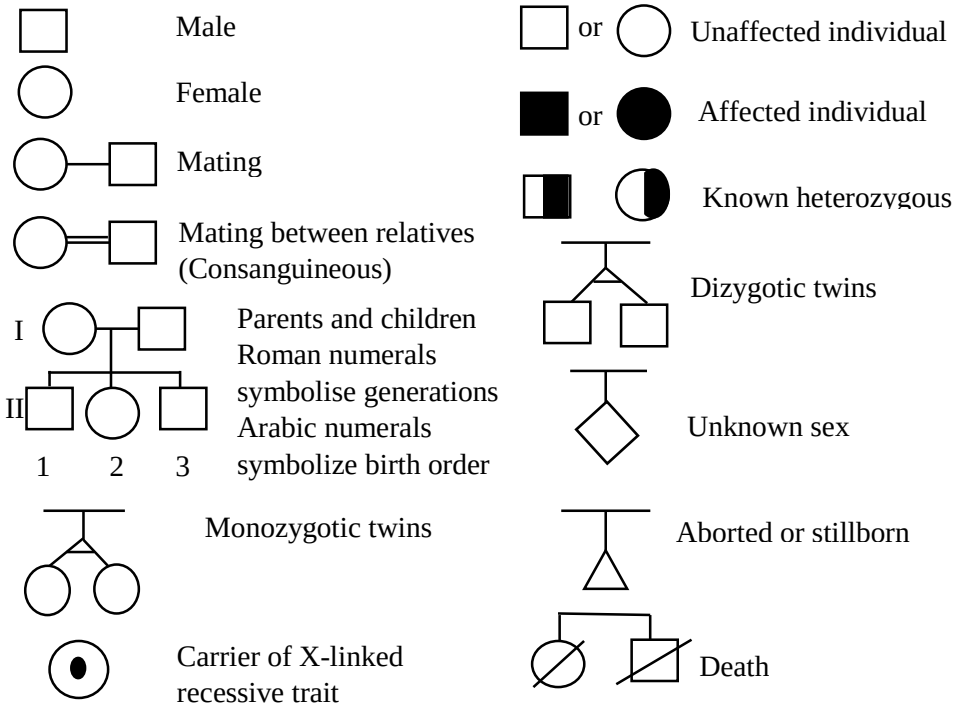
The affected individual that brings the family to the attention of a geneticist is called the **proband** (or propositus). If the individual is unaffected, they are called the **consultand**. If an individual is known to have symptoms of the disease (**affected**), the symbol is filled in. Sometimes a half-filled in symbol is used to indicate a known **carrier** of a disease; this is someone who does not have any symptoms of the disease, but who passed the disease on to subsequent generations because they are a **heterozygote**. A circle with a dot in the centre indicates female carriers of X-linked traits.

Given a pedigree of an uncharacterized disease or trait, one of the first tasks is to determine which modes of inheritance are possible and then which mode of inheritance is most likely. This information is essential in calculating the probability that the trait will be inherited in any future offspring.

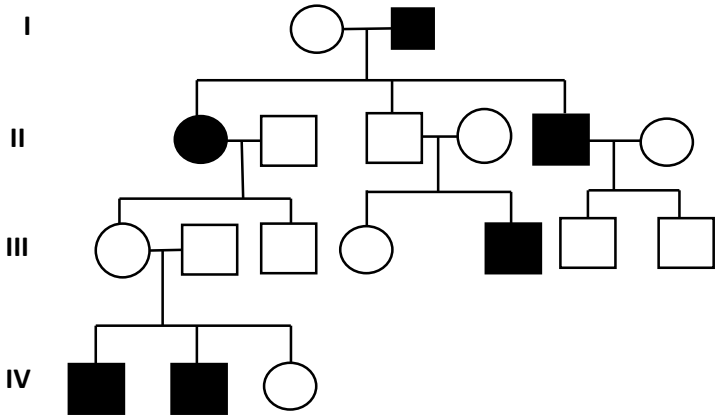
There are five major types of inheritance which are considered.

1. autosomal dominant (AD)
2. autosomal recessive (AR)
3. X-linked dominant (XD)
4. X-linked recessive (XR)
5. Y-linked (Y).

Pedigree analysis symbols

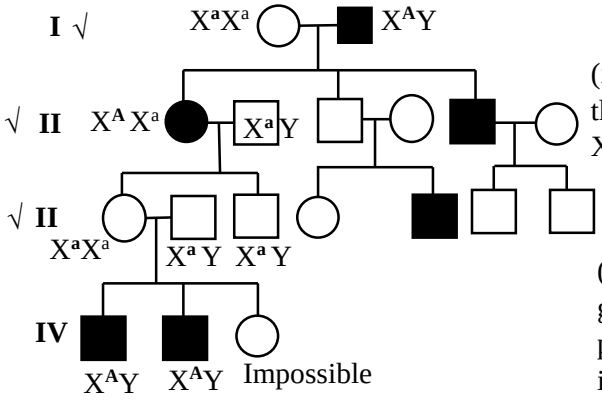


Using the pedigree below to identify the type and nature of the inherited character



Assuming the pedigree below shows a trait inherited as a sex linked dominant trait

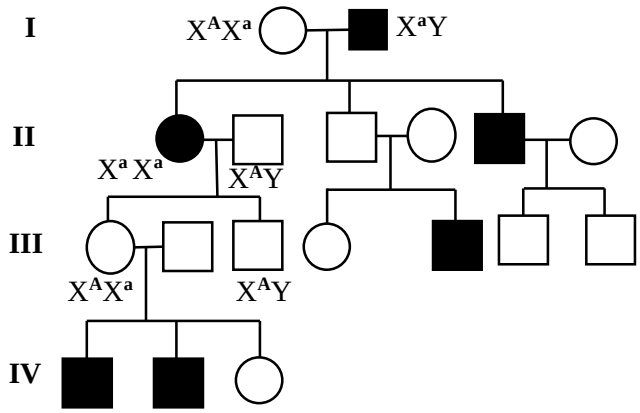
Procedure



(i) For a sex linked dominant trait the male in generation I must be X^AY

(ii) It is thus impossible to get generation **IV**, therefore the pedigree does not show a trait inherited as a sex linked dominant trait

Assuming the pedigree below shows a trait inherited as a sex linked recessive trait



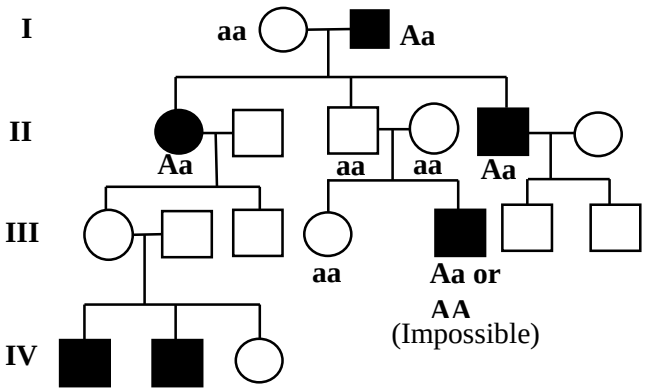
(i) For sex linked recessive trait the male in generation I must be X^aY

(ii)To get the daughter in generation II, the mother in generation I must be heterozygous

(iii) It is impossible to get the son X^AY in generation **III** because the mother (X^aX^a) in generation II is homozygous recessive

Assuming the pedigree below shows a trait inherited as an **autosomal dominant trait**

The locus for the gene must be on one of the 22 autosomes and not on the X chromosome

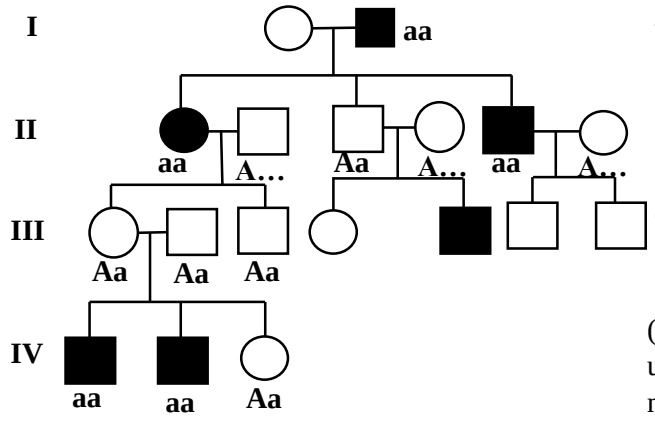


(i) For the Male in generation **I** to have the trait must have at least one dominant allele (**A**)

(ii) For the unaffected son (**aa**) in generation **II** the father in generation **I** must be heterozygous **Aa**

(iii) The son in generation **III** (**Aa or AA**) is impossible because the father (**aa**) in generation **II** married homozygous (**aa**) recessive wife

Assuming the pedigree below shows a trait inherited as an **autosomal recessive trait**



(i) For autosomal recessive trait, anyone with the trait must be homozygous (**aa**)

(ii) For the unaffected so in generation **II** the mother in generation **I** must be Heterozygous (**Aa**)

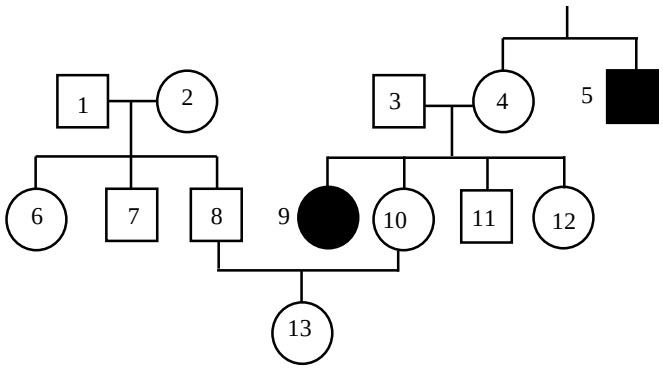
(iii) The unknown genotypes of the unaffected 1 male and two females marrying in this family must have at least one dominant allele (**A**)

(iv) To get the affected 2 sons and 1 unaffected daughter in generation IV, the mother (**Aa**) in generation III must have married heterozygous male (**Aa**)

The pedigree above shows a trait inherited by an autosomal recessive trait or gene

WORKED EXAMPLES

1. Examine the pattern of inheritance of PKU shown below



(a) What evidence is there that PKU is controlled by a recessive gene?

Couple 3 and 4 are phenotypically normal but have an affected daughter. If the gene were dominant, at least one of the parents would have been affected. The gene is unlikely to have arisen as a spontaneous mutation because it is already in the family (individual 7)

(b) What evidence is there that PKU is not sex-linked?

Individual 9 is an affected woman born to phenotypically normal parents. Given that the gene is recessive, both parents must have a copy of the gene. If it were sex-linked, the father would show the symptoms of PKU because the Y chromosome only carries genes for sex.

(c) Which individuals are definitely carriers(heterozygous) based on the evidence available.

Individuals 3 and 4

(d) Which other individuals could be carriers?

Individuals 1, 2, 6, 7, 8, 10, 11, 12 and 13 could all be carriers. It is impossible to prove a person is not a carrier on the basis of normal breeding patterns. A biochemical test would be needed.

(e) In a real situation, the individuals numbered 10, 11 and 12 may well wish to know if they are carriers since their sister suffers from PKU. What are their chances of being carriers?

50% since a ratio 1 affected: 2 carriers: 1 normal would be expected among the children of individuals 3 and 4. However, individuals 10, 11 and 12 know they are not PKU sufferers and so are either carriers or normal. In this situation, there is a 2 in 3 chance of being a carrier(66.7%).

2. Calculate the number of different combinations of chromosomes in the pollen grains of the crocus *(Crocus balansae)* which has a diploid number of six *(2n* = 6).

The number of different combinations of chromosomes in the pollen gamete cells is calculated using 2^n, where n is the haploid number of chromosomes

In Crocus, since 2n=6, n=3

Therefore, combinations $=2^3 =8$

3. A homozygous purple-flowered short-stemmed plant was crossed with a homozygous red-flowered long-stemmed plant and the F_1 phenotypes had purple flowers and short stems. When the F_1 generation was test crossed with a double homozygous recessive plant the following progeny were produced. 52 purple flower, short stem 47 purple flowers, along stem 49 red flower, the short stem 45 red flowers, long stem Explain these results fully.

The F_1 phenotypes show that purple flower and short stem are dominant and red flower and long stem are recessive. The approximate ratio of 1: 1: 1: 1 in a dihybrid cross suggest that the two genes controlling the characteristics of flower colour and stem length are not linked and the four alleles are situated on different chromosomes.

Let: **P** allele for purple flower
p represent allele for red flower
S represent allele for short stem
s represent allele for long stem

Since the parental stocks were both homozygous for both characters the F_1 genotypes must be **PpSs**

Testcross phenotypes purple flower x red flower
Short stem long stem

Testcross genotypes (2n) **PpSs** x **ppss**

Meiosis

Gametes		**PS**	**Ps**	**pS**	**Ps**
Rando fertilization Offspring genotypes (2n)	**ps**	**PS** **Ps**	**Ps** **ps**	**pS** **ps**	**Ps** **Ps**

Offspring phenotypes
1 purple flower, short stem
1 purple flower, long stem
1 red flower, short stem
1 red flower, long stem

4. If a pure strain of mice with brown- coloured fur is allowed to breed with a pure strain of mice with grey-coloured fur they produce offspring having brown-coloured fur. If the F_1 mice are allowed to interbreed they produce an F_2 generation with fur colour in the proportion of three brown-coloured to one grey.
   (a) Explain these results fully.
   (b) What would be the result of mating a brown-coloured heterozygote from the F_2 generation with the original grey-coloured parent?
   Answer

(a) Let: **B** represent allele for brown fur
b represent allele for grey fur

Parental phenotypes Brown fur x grey fur

Parental genotypes (2n) **BB** x **bb**

Meiosis
Gametes (n) **B** **B** x **b** **b**

Random fertilisation

F_1 genotypes (2n) **Bb** **Bb** **Bb** **Bb**

F_1 phenotypes all brown

F_1 phenotypes Brown fur x brown fur

F_1 genotypes **Bb** x **Bb**

Meiosis
Gametes (n) **B** **b** x **B** **b**

Random fertilisation

F_2 genotypes(2n) **BB** **Bb** **Bb** **bb**

F_2 phenotypes 3 brown fur : 1 grey fur

(b)
Experimental phenotypes Brown fur x grey fur

Experimental genotypes **Bb** x **bb**

Meiosis
Gametes (n) **B** **b** x **b** **b**

Random fertilisation
offspring genotypes(2n) **Bb** **Bb** **bb** **bb**

Offspring phenotypes brown fur grey

Offspring phenotypic ratio 1 brown fur : 1grey fur

In the case of monohybrid inheritance, the offspring from a heterozygous genotype crossed with a homozygous recessive genotype produce equal numbers of offspring showing each phenotype: in this case 50% brown fur and 50% grey fur.

5. In cats, the genes controlling the coat colour are carried on the X chromosomes and are codominant. A black-coat female mated with a ginger-coat male produced a litter consisting of black male and tortoiseshell female kittens. What is the expected F_2 phenotypic ratio? Explain the results.

Let: **B** represent black coat colour
G represent ginger coat colour
XX represent female cat
XY represent male cat

Parental phenotypes ginger-coat x black-coat
Male x female

Parental genotypes(2n) X^GY x X^BX^B

Meiosis
Gametes(n) X^G Y x X^B X^B

Random fertilisation

F_1 genotypes (2n) X^GX^B X^GX^B X^BY X^BY

F_1 phenotypes tortoiseshell Coat colour black-coat Coat colour

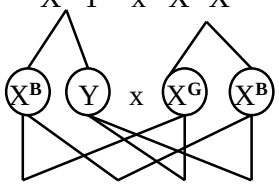
The parental female must be homozygous for black-coat colour since this is the only condition to produce a black-coat phenotype

F_1 phenotypes Black-coat Male x tortoiseshell coat female

F_1 genotypes (2n) $X^B Y$ x $X^G X^B$

meiosis

Gametes(n) X^B Y x X^G X^B



Random fertilisation

F_2 genotypes(2n) $X^B X^G$ $X^B X^B$ $X^G Y$ $X^B Y$

F_2 phenotypes

1 $X^B X^G$ tortoiseshell-coat colour female

1$X^B X^B$ black-coat colur female

1 $X^G Y$ ginger coat colour male

1 $X^B Y$ black coat colour male

6. (a) Explain, using appropriate genetic symbols, the possible blood groups of children whose parents are both heterozygous, the father being blood group A and the mother B.

Let: **I** represent the gene for blood group
A represent the allele for A(dominant)
B represent the allele for B(dominant)
o represent the allele for O (recessive)

Parental phenotypes Blood group A x Blood group B

Parental genotypes (2n) $\mathbf{I^A I^O}$ x $\mathbf{I^B I^O}$

Meiosis

Gametes(n) $\mathbf{I^A}$ $\mathbf{I^O}$ x $\mathbf{I^B}$ $\mathbf{I^O}$

Random fertilisation

Offspring genotypes $\mathbf{I^A I^B}$ $\mathbf{I^A I^O}$ $\mathbf{I^O I^B}$ $\mathbf{I^O I^O}$

Offspring phenotypes blood groups AB A B O

(b) white.

(b) If these parents have non-identical twins, what is the probability that both twins will have blood group A?

There is a probability of $\frac{1}{4}$ (25%) that each child will have blood group A. So the probability that both will have blood group A is $\frac{1}{4}$ x $\frac{1}{4}$ = $\frac{1}{16}$ (6.25%)

7. In the guinea pig (*Cavia*), there are two alleles for hair colour, black and white, and two alleles for hair length, short and long. In a breeding experiment, all the F_1 phenotypes produced from a cross between pure-breeding, short black-haired and pure-breeding, long white-haired parents had short black hair. Explain

(a) which alleles are dominant, and

(b) the expected proportions of F_2 phenotypes.

Answer

(a) If short black hair appeared in the F_1 phenotypes, then short hair must be dominant to long hair and black hair must be dominant to

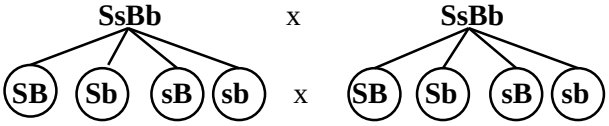
(b) Let: **B** represent allele for black hair
b represent allele for white hair
S represent allele for short hair
s represent allele for long hair

F_1 phenotypes short black hair x short black hair

F_1 genotypes (2n) **SsBb** x **SsBb**

Meiosis

Gametes (n)



Gametes		**SB**	**Sb**	**sB**	**Sb**
Random fertilisation	**SB**	**SSBB**	**SSBb**	**SsBB**	**SsBb**
	Sb	**SSBb**	**SSbb**	**SsBb**	**Ssbb**
	sB	**SsBB**	**SsBb**	**ssBB**	**ssBb**
F_2 genotypes (2n)	**Sb**	**SsBb**	**Ssbb**	**ssBb**	**Ssbb**

F_2 phenotypes 9 short black hair: 3 short white hair: 3 long black hair: 1 long white hair

8. Flower colour in sweet pea plants is determined by two allelomorphic pairs of genes **(R,r,** and **S,s).** If at least one dominant gene from each allelomorphic pair is present the flowers are purple. All other genotypes are white. If two purple plants, each having the genotype **RrSs,** are crossed, what will be the phenotypic ratio of the offspring?

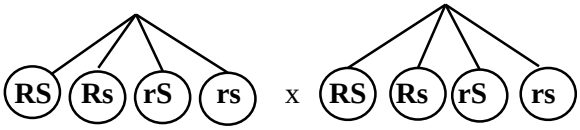
Let: **R**, **r** and **S**, **s** represent allelomorphic pairs of alleles controlling flower colour

Parental phenotypes purple x purple

Parental genotypes (2n) **RrSs** x **RrSs**

Meiosis

Gametes(n)



Gametes		**RS**	**Rs**	**rS**	**Rs**
Random fertilization	**RS**	**RRSS** Purple	**RRSs** purple	**RrSS** purple	**rRSs** purple
	Rs	**RRSs** Purple	**RRss** white	**RrSs** purple	Rrss White
	rS	**RrSS** Purple	**RrSs** purple	**rrSS** white	**rrSs** white
F_2 genotypes(2n)	**rs**	**RrSs** **Purple**	**Rrss** white	**rrSs** white	**rrss** white

Offspring phenotypic ratio: 9 purple: 7 white

9. In *Drosophila* the genes for wing length and for eye colour are sex-linked. Normal wing and red eye are dominant to miniature wing and white eye.

(a) In a cross between a miniature wing, red-eyed male and a homozygous normal wing, white-eyed female, explain fully the appearance of
   (i) the F_1 and
   (ii) the F_2 generations.

(b) Crossing a female from the F_1 generation above with a miniature wing, the 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, white-eyed males and females	18
miniature wing, red-eyed males and females	36

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

(a) Let:
N represent allele for normal wing
n represent allele for miniature wing
R represent allele for red eye
r represent allele for white eye
XX represent female fly
XY represent male fly

(i) Parental phenotypes miniature wing, red eye x normal wing, white eye

Parental genotypes (2n) $X^{nR}Y$ x $X^{Nr}X^{Nr}$

Meiosis

Gametes (n) X^{nR} Y x X^{Nr} X^{Nr}

Random fertilisation

F_1 genotypes (2n) $X^{nR}X^{Nr}$ $X^{nR}X^{Nr}$ $X^{Nr}Y$ $X^{Nr}Y$

F_1 phenotypes normal wing, red eye female; normal wing white eye male

(ii) Assuming no crossing-over between the genes for wing length and eye colour in the female, the following results are likely to appear

F_1 phenotypes normal wing, white ♂ x normal wing, red eye ♀

F_1 genotypes (2n) $X^{Nr}Y$ x $X^{nR}X^{Nr}$

Meiosis

Gametes (n) X^{Nr} Y x X^{nR} X^{Nr}

Random fertilisation

F_2 genotypes (2n) $X^{Nr}X^{nR}$ $X^{Nr}X^{Nr}$ $X^{nR}Y$ $X^{Nr}Y$

F_2 phenotypes 1 normal wing, red eye ♀; 1 normal wing, white eye ♀; 1 miniature wing, red eye♂; 1 normal wing white eye ♂

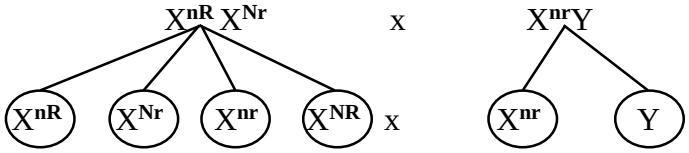
(b) The lack of a 1: 1: 1: 1 ratio of phenotypes resulting from this cross indicates crossing-over between the genes for wing length and eye colour in the female.

Testcross phenotypes normal wing, red eye ♀ x miniature wing, white eye ♂

Testcross genotypes(2n) $X^{nR}X^{Nr}$ x $X^{nr}Y$

Meiosis (crossing over)

Gametes (n)



Random fertilization	♂ / ♂	X^{nR}	X^{Nr}	X^{nr}	X^{NR}
Offspring genotypes(2n)	X^{nr}	$X^{nR}X^{nr}$ ♀	$X^{Nr}X^{nr}$ ♀	$X^{nr}X^{nr}$♀	$X^{NR}X^{nr}$♀
	Y	X^{nR}Y ♂	X^{Nr}Y ♂	X^{nr}Y ♂	X^{NR}Y ♂

Offspring phenotypes	miniature red	normal white	miniature white	normal red
	36	35	18	17

The alleles for wing length and eye colour are shown on the two F_1 female (X) chromosomes. Crossing-over between the alleles gives the recombinant genotypes. Out of 106 flies, 35 show recombination of alleles (18 + 17), therefore the crossover value is $\frac{35}{106} = 30\%$.

10. In poultry, the allele for white feather **(W)** is dominant over the allele for black feather **(w).** The alleles for pea comb, **P,** and rose comb, **R,** produce the phenotypes stated. If these alleles are present together they produce a phenotype called walnut comb and if their recessive alleles are present in the homozygous condition they produce a phenotype called a single comb. A cross between a black rose-comb cock and a white walnut-comb hen produced the following phenotypes:

3 white walnut-comb
3 black walnut-comb
3 white rose-comb
3 black rose-comb
1 white pea-comb
1 black pea-comb
1 white single-comb and 1 black single-comb.

What are the parental genotypes? Show clearly how they give rise to the phenotypes described above?

Let: **P** represent allele for pea comb
R represent allele for rose comb
a single P allele occurring together produce walnut comb
a double homozygous recessive genotype produces single comb
W represent allele for white feathers (dominant)
w represent allele for black feathers (recessive)

If eight different phenotypes are produced from the cross, each parent must possess as many heterozygous alleles as possible. Hence the genotypes are as shown below:

Parental phenotypes: Black, rose-comb cock x white, walnut-comb hen

Parental genotypes (2n) **wwRrpp x WwRrPp**

Meiosis

Gametes		WRP	WRp	WrP	Wrp	wRP	wRp	wrP	Wrp
Random fertilisation	wRp	WRP wRp white, walnut comb	WRp wRp White, rose-comb	WrP wRp white, walnut comb	Wrp wRp White, rose-comb	wRP wRp black, walnut-comb	wRp wRp black, rose-comb	wrP wRp black, walnut-comb	Wrp wRp black, rose-comb
Offspring genotypes(2n)	wrp	WRP wrp white, walnut comb	WRp wrp White, rose-comb	WrP wrp white, pea-comb	Wrp wrp white single-comb	wRP wrp black, walnut-comb	wRp wrp black, rose-comb	wrP wrp black, pea-comb	wrp wrp black, single comb

Offspring phenotypes

3 white, walnut comb: 3 black, walnut- comb: 3 White, rose-comb: 3 black, rose-comb

1 white, pea-comb: 1 white single-comb: 1 black, single comb

11. In White Leghorn fowl, plumage colour is controlled by two sets of genes, including the following:
W (white) dominant over **w** (colour) **B** (black) dominant over **b** (brown).

The heterozygous F_1 genotype **WwBb** is white. Account for this type of gene interaction and show the phenotypic ratio of the F_2 generation.

solution

Since both dominant alleles **W**, white and **B**, black, are present in the heterozygous F_1 genotype, and the phenotype is white, it may be concluded that the alleles show an epistatic interaction where the white allele represents the epistatic gene.

F_2 generation is shown in the equation

F_1 phenotypes white cock x white hen

F_1 genotypes(2n) **WwBb** x **WwBb**

Meiosis

Gametes(n) **WB** **Wb** **wB** **wb** x **WB** **Wb** **wB** **wb**

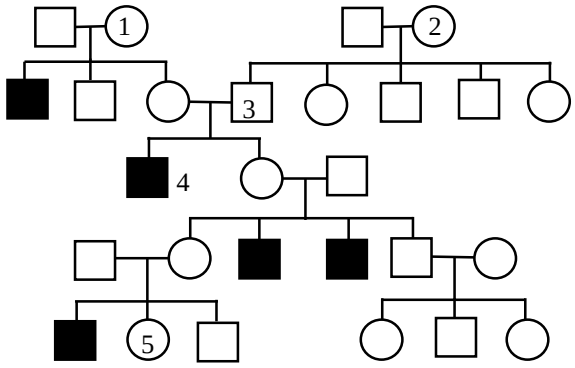
Ransdom fertilisatio		**WB**	**Wb**	**wB**	**wb**
	WB	**WWBB** white	**WWBb** white	**WwBB** white	**WwBb** white
	Wb	**WWBb** white	**WWbb** white	**WwBb** white	**Wwbb** white
F_2 genotypes	**wB**	**WwBB** white	**WwBb** white	**wwBB** **black**	**wwBb** **black**
	wb	**WwBb** white	**Wwbb** white	**wwBb** back	**Wwbb** brown

F_2 phenotypes 12 white: 3 black colour: 1 brown colour

EXERCISE

1. (a) Give two difference between the X and Y chromosomes of humans (02 marks)

   (b) The diagram below is a family tree showing the pattern of inheritance of a sex-linked genetic disorder through five generations.



   (i) Identify two features of the inheritance of this disorder that are characteristic of sex-linked inheritance. (02 marks)

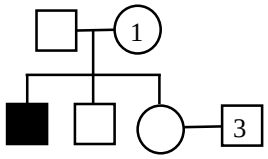
   (ii) The disorder is caused by a recessive allele of a single gene. Using the symbol A to represent the normal allele and a to represent the recessive allele, write down the most likely genotypes of individuals 1, 2, 3 and 4. (04 marks)

   (c) Individual 5 is engaged to be married. Her future partner comes from a family with no history of this genetic disorder. They plan to have several children.

   (i) If the individual 5's first child is a boy, what is the probability that he will have the disorder? (01 mark)

   (ii) If individual 5's first child is a girl, what is the probability that she will have the disorder? (01 marl)

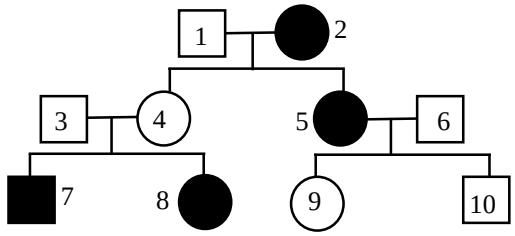
   (d) The pedigree below shows a small part of the same family tree, involving individuals 1 and 3. If the disorder had been caused by a dominant allele rather than a recessive allele, the pattern of inheritance.



Using the information in the complete tree, re-draw this part of the tree to show this different pattern of inheritance. (04 marks)

2. Cystic fibrosis is a condition in which affected people suffer from the accumulation of thick,

sticky mucus in their lungs. The diagram below shows part of a family tree in which some individuals have cystic fibrosis.

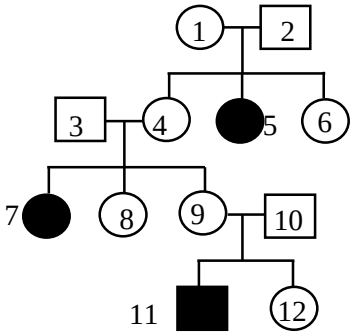


(a) Explain the evidence from this family tree that cystic fibrosis is controlled by a recessive allele. (02 marks)

(b) What is the probability that the next child born to individuals 3 and 4 would have at least one allele for cystic fibrosis? Explain. (02 marks)

(c) In Britain, 1 in 2000 people is born with cystic fibrosis. What is the frequency of the cystic fibrosis allele in the British population? (02 marks)

3. Figure below shows how sickle cell anaemia has affected a family line. Sickle cell anaemia is a recessive genetic defect which is not linked.



(a) State the numbers of all the individuals in the family line that is certain to be heterozygous for this gene.

(b) What is the probability that individual 6 is heterozygous for this gene? (Show your working)

(c) The parasite which causes malaria digests haemoglobin in the red blood cells. Suggest two reasons why an individual who is heterozygous for this gene may show resistance to malaria.

(d) State the difference between individuals who have sickle cell anaemia and those that have sickle cell trait.

4. Mary, a student, with blood group A had a baby with blood group O. Peter, a fellow Student who she named as responsible for the pregnancy, denied responsibility. The case was then taken to court. The following facts were determined.
Peter's mother was of blood group A and father, blood group B. State whether the court will find Peter guilty or innocent. Show how you reached your conclusion.

5. (a) State Mendel's first law of inheritance and explain what it means

(b) (i) State the stages of meiosis that illustrate this law

(ii) Explain what takes place in the stages you have named in a (ii) above.

(c) In human beings, brown eye are usually dominant over blue eyes. Suppose a blue-eyed man marries a brown-eyed woman whose father was blue-eye. What proportion of their children would you predict will have blue eyes?
Show your working

6. Gene R for red colour can only express itself in a Dihybrid cross in the presence of gene C which complements its action to form colour. When two white flowering genotypes CCrr and ccRR were crossed the F_1 generation were all red flowers.

(a) (i)What would be the genotypes of F_2 when the F_1 progeny are selfed? (Show your working).

(ii) What would be the phenotype ratio of the F_2 progeny?

(iii) Comment on the F_2 phenotype ratio you have obtained in (a)(ii) above.

7. (a) What is a sex-linked trait?

(b) (i) Why are sex-linked traits most common in males among humans?

(ii) Haemophilia is a condition caused by a recessive gene carried on the X

chromosome. Determine the phenotype of the children from a carrier mother and a normal father.

(c) In poultry, feather colour is controlled by two sets of alleles, W(white) dominant over w (coloured) and B (black) dominant over b(brown). A fowl heterozygous for both alleles (WwBb) is white.

8. (a) Explain why the genetic constitution of WwBb is white.
   (b) Work out to show the phenotype ratio of crossing a white cock (WwBb), with a brown hen.

   (c) State the possible genotypes of a black fowl.
   (d) In a variety of beans, yellow seed colour is dominant over the green and smooth seed coat is dominant over wrinkled. When yellow smooth beans were crossed with green wrinkled beans, all F_1 had a yellow smooth seed. The F_2 progeny yielded 556 seeds.
      (i) Assuming no linkage, state the four possible characters in the F_2 progeny and their corresponding phenotypic ratios.
      (ii) Calculate the number of individuals for each of the characters in the F_2 population. Calculate the percentage crossover in this experiment.
9. In cats, sex is determined by X and Y chromosomes in the same way as humans.
   One gene for coat colour in cats is present on the X chromosome but not on the Y chromosome. This gene has two alleles, orange (B) and black (b). An X chromosome bearing the B allele is represented by X^B and one bearing the b allele by X^b.
   Female cats that are homozygous for the X^b allele have black coats; female cats that are heterozygous have tortoiseshell coats, that is orange coats with dark patches.
   (a) Give the genotype of:
      (i) a female cat with a tortoiseshell coat.
      (ii) a male cat with an orange coat.
      (iii) a male cat with a black coat.
      (03 marks)
   (b) A black-coated male cat is mated with a tortoiseshell-coated female cat.
   Use a genetic diagram to explain what would be the expected ratios of the genotypes and the phenotypes of the kittens that could be produced by the cross.

10. In broad bean, a pure-breeding variety with green seeds black hilum was crossed with a pure-breeding variety with yellow seeds and white hilum. All the F_1 plants had yellow seeds and black hilum. When these were allowed to self-fertilise, the plants of the F_2 generation produced the following seed.
    Yellow seeds with white hilum 31
    Yellow seeds with black hilum 93
    Green seeds with white hilum 8
    Green seeds with black hilum 28
    (a) What characteristics are dominant and recessive?
    (b) Construct suitable cross diagrams to show the genotypes of the plants and their gametes in each generation. (07 marks)

11. In mice, the dominant allele (B) of a gene for coat colour gives a black coat and the recessive allele (b) of this gene gives a brown coat. A second gene determines the density of the coat colour. The dominant allele(D) of this gene allows expression of coat colour, its recessive allele(d) dilutes the colour converting black to grey and brown to cream.
    (a) A breeder crossed a male black mouse with a female brown one. The offspring produced showed four different coat colours, black, grey, brown and cream.
       (I) State the genotypes for the black parent and the brown parent giving an explanation for your answer. (05marks)

(II) Construct suitable cross diagrams to show the genotypes of the offsprinsg. (03 marks)

(b) With the aid of a genetic diagram. Explain how the breeder could determine which of the black offspring were homozygous for the full-colour allele (**D**)

(c) Explain how events taking place during gametogenesis and fertilisation lead to the production of variety in the offspring. (03 marks)

12. Maize cobs may have purple or red grains. This character is controlled by a single allele. The dominant allele **A** gives a purple colour and the recessive allele **a** gives a red colour.

(a) In an experiment, a heterozygous plant is crossed with a maize plant homozygous for allele **a**. State the genotypes of these two plants. (01 mark)

(b) Grain colour is also affected by the second pair of alleles. The presence of the dominant allele **E** allows the purple or red colour to develop, but in the homozygous recessive (**ee**) no colour will develop (despite the presence of alleles **A** or **a**). A plant of genotype AAEE is crossed with a plant of genotype **aaee**.

(i) State the genotypes and phenotypes of the offspring produced as a result of this cross. (02 marks)

(ii) The plants of the offspring are allowed to self-fertilise. Draw a genetic diagram to show the possible genotypes produced as a result of this cross. (03 marks)

(iii) Predict the phenotypic ratio that would be obtained from this cross. (03 marks)

13. (a) What is epistasis? (01 mark)

(b) How does epistasis differ from Mendelian dominance? (01 mark)

(c) In oats, the grain is enclosed by the dried remains of the outer parts of the flower, called hull. In a cross between two pure-breeding varieties of oats, one with black-hulled grains, the other with white-hulled grains, the offspring(F_1) all had black-hulled grains. Allowing the F_1 plants to self-fertilise gave an F_2 with the phenotypes below

Phenotype	Number
Black-hulled grains	418
Grey-hulled grains	106
White-hulled grains	36

Thes data show evidence of epistasis

(i) What genetic ratio is suggested from the figures given? (01 mark)

(ii) Devise suitable symbols for the alleles involved. (01 mark)

(iii) Set out the crosses, using a Punnet square, to show the gametes, genotypes and phenotypes in each generation. (05 marks)

14. In the fruit fly *Drosophila*, the vestigial wing is recessive to normal and white eye colour recessive to the normal red. These genes are on the X-chromosome and in *Drosophila* the heterogametic sex is male.

(a) Briefly explain the terms 'heterogametic' and 'sex-linkage' and describe how you would distinguish between male and female offspring. (5 marks)

(b) What phenotypes would be expected in the F_1 of a cross between a vestigial winged, red-eyed male and a homozygous normal winged white-eyed female? (5 marks)

(c) What phenotypes would be expected in the F_2 generation when Fj flies interbreed? Show clearly all your working (from (*b*)).(12 marks)

15. Typically, a 9:3:3:1 phenotypic ratio is obtained in the F_2 phenotypes in dihybrid inheritance of when independent assortment occurs.

(a) What affects does

(I) Linkage

(II) And incomplete dominance has on this ratio. (05 marks)

16. (a) Distinguish between the terms **gene** and **allele**. (04 marks)

(b) In maize plants, normal size is dominant to pygmy size, and normal leaf shape is dominant to the crinkly leaf shape. A plant heterozygous for both these genes was self-pollinated. Its seeds were collected and 320 plants subsequently grew. Assuming that the genes are not linked, what phenotypes and how many of each type would you expect to appear in these plants? Give a full explanation for your answer. (12 marks)

(c) What differences would you expect in the results if the genes had been linked? (04 marks)

17. (a) Explain the differences between the members of **each** of the following pairs of genetical terms and give **one** example of **each** term to illustrate your answer.

i. complete and incomplete dominance
ii. Continuous and discontinuous variation.
iii. chromosomal mutation and crossing-over
iv. polyploidy and haploidy. (12 marks)

(b) Crosses between ginger female cats and black male cats produce only tortoiseshell females and ginger-coloured males. A single gene controls expression of colour in cats.

i. Give a reasoned explanation of these results and show the genotypes of the parents, their gametes and the offspring produced in these crosses.
ii. Is it possible to have tortoiseshell male cats? Explain your answer. (08 marks)

18. A maize plant homozygous for smooth, coloured grain was cross-pollinated with a plant homozygous for wrinkled, colourless grain. The Fj plants all produced smooth, coloured grain. On cross-pollinating the F_1 plants, it was found that most of the F_2 generation resembled the original plants, 73% producing smooth, coloured grain and 22% producing wrinkled, colourless grain.

Using appropriate symbols, state the genotypes of F_1 and F_2. (08 marks)

19. (a) State four situations where Mendel's laws would not apply. (04 marks)

(b) In an animal species, individuals that are homozygous for gene A or its alleles die. Another independent gene B in the homozygous state blocks this lethal effect, otherwise, gene B has no other effect on the organism.

i. Workout the expected phenotypic ratio of the viable offsprings in a cross of individuals of AaBb and AaBB genotypes.
ii. State the type of gene interaction in b(i) above. (06 marks)