GENETICS

Genetics is a branch of biology concerned with the study of genes, genetic variation, and heredity in organisms. **Gregor Mendel**, a scientist discovered genetics in the late 19th-century. Mendel studied trait inheritance, patterns in the way traits are handed down from parents to offspring. He observed that organisms (pea plants) inherit traits by way of discrete units of inheritance.

Mendel's success was due, in part, to his careful choice of experimental organism, the garden pea, *Pisum sativum.*

There were several varieties available with distinct characteristics.

- The plants were easy to cultivate.
- The reproductive structures were completely enclosed by the petals so that the plant was normally self- pollinating.
- Artificial cross-breeding between varieties was possible and resulting hybrids were completely fertile

Important terms used in genetics

Gene: The basic unit of inheritance for a given characteristic

Allele: One of the alternative forms of the same gene responsible for determining contrasting characteristics.

Locus: Position of allele within a DNA molecule

Homozygous: The diploid condition in which the allele at a given locus are identical.

Heterozygous: The diploid condition in which the allele at a given locus are different

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

Genotype: The genetic constitution of an organism with respect to the alleles under consideration.

Dominant allele: The allele which influences the appearance of the phenotype even in the presence of an alternative allele.

Recessive allele: The allele which influences the appearance of the phenotype only in the presence of another identical allele.

F_1 generation: The generation produced by crossing homozygous parental stocks.

F_2 generation: The generation produced by crossing two F_1 organisms.

Monohybrid inheritance and the principle of segregation

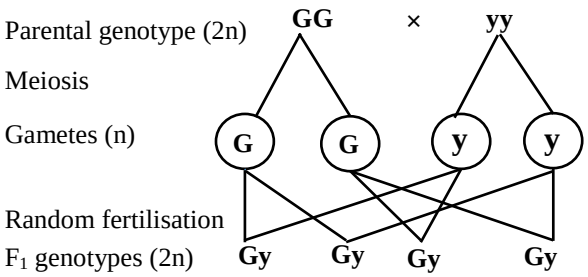
Monohybrid inheritance is the inheritance of a single gene.

Inheritance of pod colour in peas

If pea plants with **green pods** are bred repeatedly with each other so that they consistently give rise to plants with green pods, they are said to be **pure breeding** for the character of green pods. Pure breeding strains can be bred for almost any character. Organisms are homozygous (i.e. they have two alleles that are the same) for that particular gene.

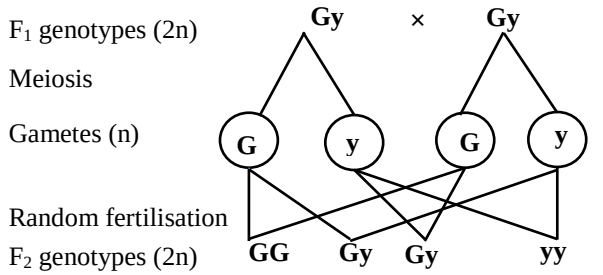
If these pure breeding green pod plants are then crossed with pure breeding yellow pod plants, all the offspring, known as the **first filial** or **F_1 generation,** turn out to produce green pods. This means that the allele for green pods is **dominant** to the allele for yellow pods, which is therefore **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 3plants with green pods to each 1 plant with yellow pods

Let: **G** represent allele for green colour (dominant)
y represent allele for yellow colour (recessive)
Parental phenotypes: Green plants X yellow plants



F_1 phenotypes all heterozygous green

The F_1 generation were self-pollinated



F_2 Phenotypes: 3 green: 1 yellow

The ratio of dominant phenotypes to recessive phenotypes of 3:1 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 were 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, became 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.*

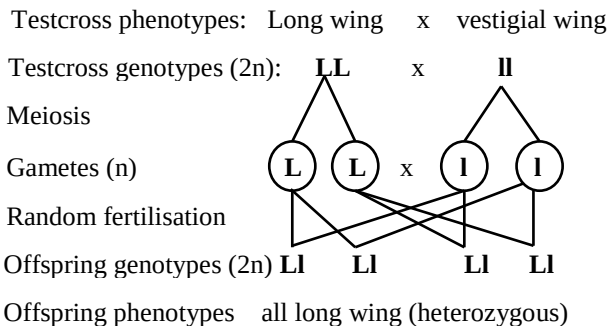
These factors determining characteristics, such as flower position, are regions of the chromosome known as **genes.** By convention, the initial letter of the dominant characteristic is used as the symbol for the gene and its capital form (e.g. A) represents the dominant form of the gene (the dominant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

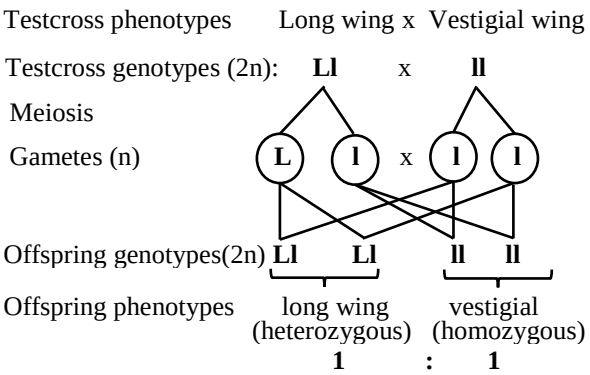
Example in the fruit fly, *Drosophila*, long wing is dominant to vestigial wing. The genotype of a long wing *Drosophila* may be homozygous **(LL)** or heterozygous **(LI).** In order to establish which is the correct genotype the fly is test crossed with a double recessive (**II**) vestigial wing fly.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

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

Homozygous long wing parent



Heterozygous long wing parent



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

Mendel's breeding experiment with tall and dwarf plant is an example of monohybrid inheritance. Monohybrid inheritance is inheritance of a single characteristics determined by one gene. Examples of monohybrid inheritance in humans include the following,

►Albinism. ►Cystic fibrosis. ►Haemophilia.
►Huntington's disease. ►Lactose intolerance. ►Phenylketonuria.
►Rhesus blood group.

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 variety of characteristics. He collected a total of 556 F2 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.** Two deductions were made from the above observations.

(a) Let
R represent round seed (dominant) **Y** represent yellow seed (dominant)
r represent 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

(b) F_1 phenotypes round and yellow seed X round and yellow seed
F_1 genotypes (2n) **RrYy** X **RrYy**
Meiosis

Gametes		**RY**	**Ry**	**rY**	**ry**
Random fertilization	**RY**	**RRYY**	**RRYy**	**rRYY**	
	Ry	**RRyY**	**RRyy**	**rRYy**	**rRrr**
F_2 genotypes(2n)	**rY**	**RrYY**	**RrYy**	**rrYY**	**rryY**
	ry	**RrYy**	**Rryy**	**rrYy**	**rryy**

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

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.***

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 of a pair 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.

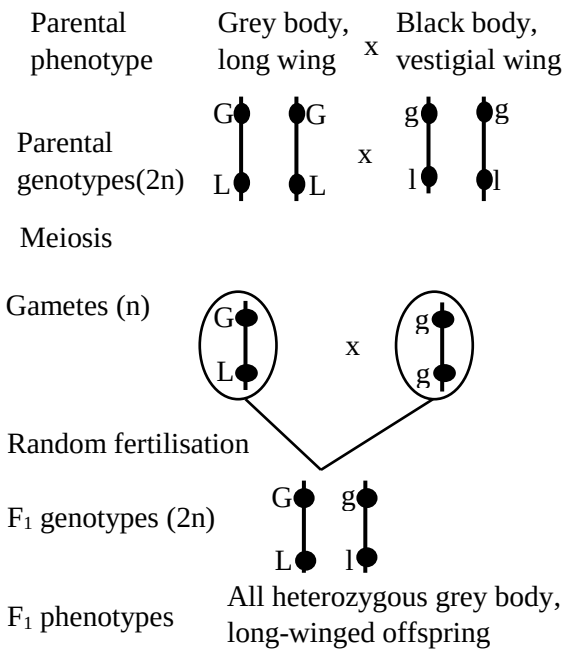
Meiosis and fertilization	**Mendel's hypotheses**
Diploid cells contain pairs of chromosomes(homologous chromosomes)	Characteristics controlled by pairs of factors
Homologous chromj0bmosomes separate during meiosis	Pairs of factors separate during gamete formation
One homologous chromosomes passes into 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

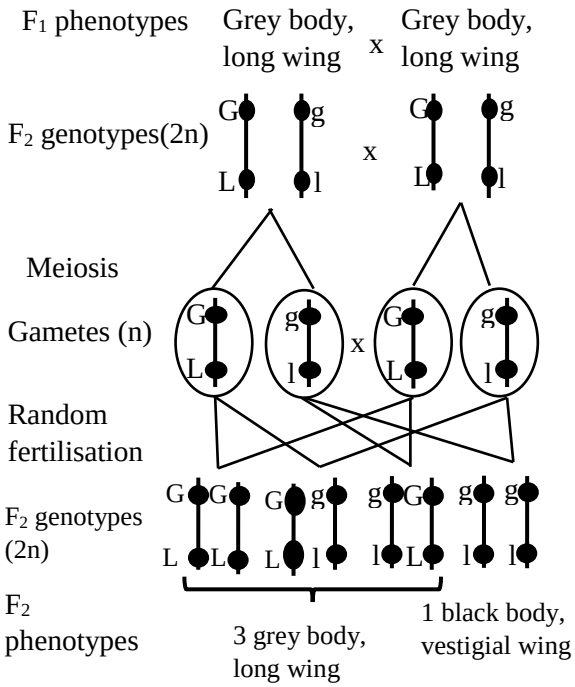
Genes situated on the same chromosome are said to be **linked.** All genes on a single chromosome form a **linkage group** and usually pass into the same gamete and are inherited together. As a result of this, genes belonging to the same linkage group usually do not show independent assortment. Since these genes do not conform to Mendel's principle of independent assortment they fail to produce the expected 9 : 3 : 3 : 1 ratio in a breeding situation involving the inheritance of two pairs of contrasted characteristics (dihybrid inheritance).

In *Drosophila* the genes for body colour and wing length have the following **allelomorphs** (phenotypic characteristics determined by different alleles): grey and black body, and long and vestigial (short) wings. Grey body and long wing are dominant. When pure-breeding grey-bodied long-winged *Drosophila* are crossed with black-bodied vestigial-winged *Drosophila*, 3:1 F_2 phenotypic ratio was produced not the 9:3:3:1as expected. This is because the genes for body colour and wing length are found on the same chromosome that is they are linked.

Let
G represents grey body (dominant)
g represent black body (recessive)
L represents long wing (dominant)
l represent vestigial wing (recessive)



Intercrossing F_1 offspring



In practice, though, this 3 : 1 ratio is never achieved and four phenotypes are invariably produced. This is because **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 and crossover values

Crossing over is the exchange of genetic material between non-sister chromatids of two homologous chromosomes that results in recombinant chromosomes during sexual reproduction. This occurs during meiosis. The alleles of parental linkage groups separate and new associations of alleles are formed in the gamete cells, a process known as **genetic recombination.** Offspring formed from these gametes showing new combinations of characteristics are known as **recombinants.** Thus crossing- over is a major source of observable genetic variation within populations.

The behaviour of a pair of homologous chromosomes in *Drosophila*, carrying the alleles grey body and long wing (both dominant) and black body and vestigial wing (both recessive), during formation of chiasmata is used to illustrate the principle of crossing-over. A cross between a male heterozygous grey-bodied long-winged *Drosophila* and a female homozygous black-bodied vestigial-winged *Drosophila* produced heterozygous F_1 offspring with grey bodies and long wings. Test crossing the F_1 generation flies with homozygous double recessive flies produced the following results.

Parental phenotypes	Grey body, long wing	965
	Black body, vestigial wing	944
Recombinants phenotypes	Black body, vestigial wing	206
	Grey body, vestigial wing	185

These results indicate that the genes for body colour and wing length are linked. (a hybrid cross between an F_1 heterozygote and a double homozygous recessive would have produced a 1 : 1 : 1 : 1 phenotypic ratio if the genes had been situated on different chromosomes and therefore had undergone random assortment.) Using the figures obtained from the above cross it is possible to calculate the recombination frequency of the genes for body colour and wing length.

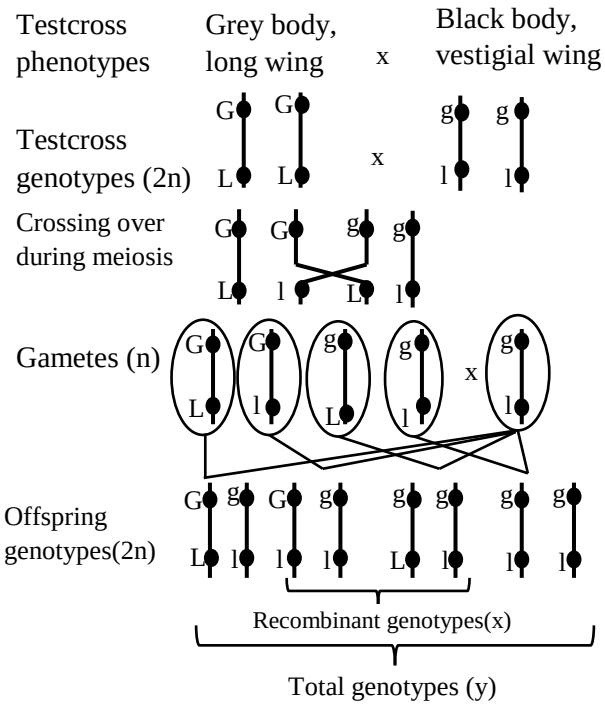
The **recombination frequency** is calculated using the formula:

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

From the example above the recombination frequency (%) is

$$\frac{(206+185)}{(965+944)+(206+185)}\ \text{X}\ 100: \frac{391}{2300}\ X\ 100 = 17\%$$

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.



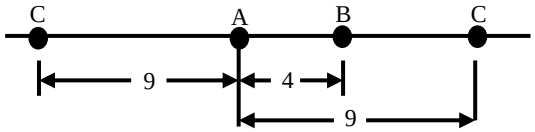
Genetic explanation of crossing-over and the reappearance of recombinant genotypes. The recombination frequency can be calculated by counting the number of individuals showing recombination and the total number of individual and applying the formula.

Recombination frequency (%) = $\frac{x}{y}\ x\ 100$

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

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

P-Q = 24%

R-P = 14%

R-S = 8%

S-P = 6

Sex determination

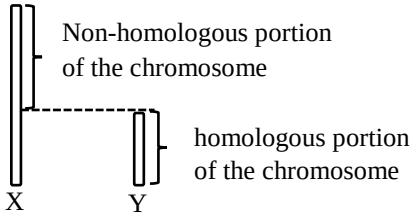
Examination of the chromosome structure of a range of animals revealed that males and females showed certain chromosomal differences. Pairs of chromosomes (homologous chromosomes) are found in all cells, but one pair of chromosomes always shows differences between the sexes. These are the **sex chromosomes** or **heterosomes.** All other chromosomes are known as **autosomal chromosomes** or **autosomes.** The chromosomes are known as **X** and **Y** chromosomes, and the genotype of the female is **XX** and that of the male is **XY**. These characteristic sex genotypes are found in most animals, including humans; but in the case of birds (including poultry), moths and butterflies the sex genotypes are reversed: the females are **XY** and the males are **XX**. In some insects, such as the grasshopper,

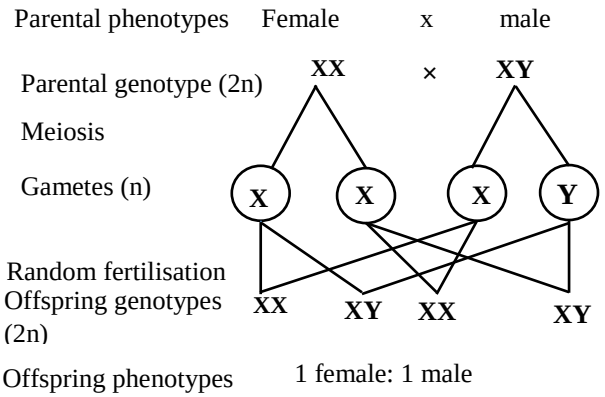
the Y chromosome may be absent entirely and so the male has the genotype **XO**.

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 contain 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. The sex having the **XX** genotype is described as **homogametic** as it produces gamete cells containing only **X** chromosomes. Organisms with the **XY** genotype are described as **heterogametic** since half their gametes contain the **X** chromosome and half the Y chromosome.

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.





Sex linkage

Genes carried on the sex chromosomes are said to be **sex-linked**. In the case of the heterogametic sex there is a portion of the X chromosome for which there is no homologous region of the Y chromosome. Characteristics determined by genes carried on the non-homologous portion of the X chromosome therefore appear in males even if they are recessive. This special form of linkage explains the inheritance of **sex-linked traits** such as red-green colour blindness, premature balding and haemophilia. Haemophilia or 'bleeder's disease' is a sex- linked recessive condition which prevents the formation of factor VIII, an important factor in increasing the rate of blood clotting. 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

Parental phenotypes:	Normal female(carrier)	x	Normal male
Parental genotypes (2n):	X^HX^h	x	X^HY
Meiosis Gametes (n) Random fertilisation	X^H X^h	x	X^H Y
Offspring genotypes (2n)	$X^H X^H$ $X^H Y$		$X^h X^H$ X^hY
Offspring phenotypes:	normal Female	normal male	normal female (carrier) haemophiliac male

Mechanism for the inheritances of sex-linked allele for Haemophilia

Examples of sex linked (X linked) conditions in humans include,

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

Red and white eyes in Drosophila is another sex linked (X linked) trait.
Inheritance of eye colour. 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) $X^R X^R$ 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) $X^r X^r$ 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 a phenotype somewhere **between those of the two** parental varieties. This phenomenon is seen 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. The alleles involved in sickle cell condition and coat colour in some breeds of cattle are all examples of characteristics involving incomplete dominance

In genetics, genes responsible for codominance are represented by capital letters but 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.

As there are no accepted genotypic symbols for alleles showing codominance, the importance of specifying symbols in genetic explanations is apparent. For example, in the case of the Andalusian fowl numerous genotypic symbols may be used to illustrate the alleles. The results of a cross between black and splashed white homozygous fowl.

If the F_1 generation are 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 have the F_1 genotype. This ratio of 1:2:1 is characteristic of examples of codominance.

Let:
B represent the black allele
B^W represent the splashed white allele

Parental phenotypes Black (homozygous) x splashed white (homozygous)

Parental genotypes (2n) **BB** x **B^WB^W**

Meiosis

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

Random fertilisation

F_1 genotypes (2n) **BB^W** **BB^W** **BB^W** **BB^W**

F_1 phenotypes all 'blue' heterozygotes

Production of F_1 hybrids of Andalusian fowl

F_1 phenotypes Blue x Blue

F_1 genotypes (2n) **BB^W** x **BB^W**

Meiosis

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

Random fertilisation

F_2 genotypes (2n) **BB** **BB^W** **BB^W** **B^WB^W**

F_2 phenotypes 1 black 2 black 1 splashed white

Production of F_2 hybrids of Andalusian fowl.

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 generation

F_1 phenotypes Pink flower 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

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 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 characteristic 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 allelomorph)** condition and it controls such characteristics as coat colour in mice, eye colour in mice and blood group in humans.

1. 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

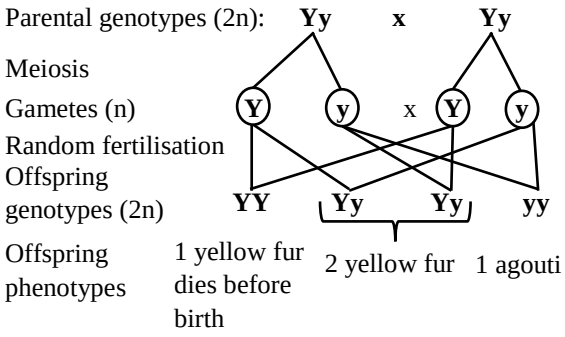
These are genes that may prevent development or cause the death of an organism or its germ cells. They are usually a result of mutations in genes that are essential to growth or development. **Lethal alleles** may be recessive, dominant, or conditional depending on the gene or genes involved

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. Using Mendelian ratio of 3: 1, this explains the fetal death of *homozygous* yellow coat mice. 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)
y represent allele for agouti fur(recessive)
Parental phenotypes: yellow fur x yellow fur



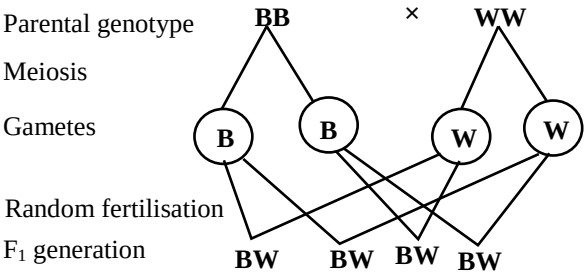
Genetic explanation of fur colour inheritance in mice showing the lethal genotype YY

Codominance

In this variation, the two alleles both 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 do not show intermediate characteristic but instead both characteristics of the two pure line parents express itself independent and both characteristics will 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**.

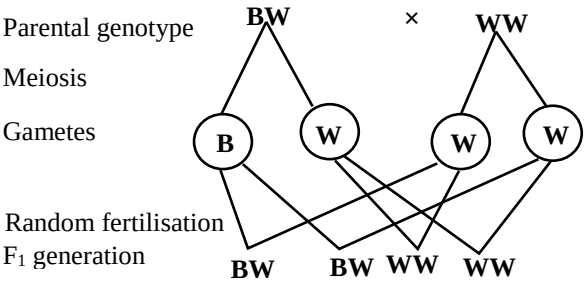
Thus: a person with the genotype **AA** or **AO** belongs to blood group A,
A person with the genotype **BB** or **BO** belongs to blood group B.
A person with the genotype **AB** belongs to blood group AB.
A person with the genotype **OO** belongs to blood group O.
The genetic crosses of codominance condition is done like for incomplete codominance, where the two genes are represented with capital letters of different kinds.
In a monohybrid cross, a male rabbit homozygous for brown hairs was mated with a female rabbit homozygous for white hairs. Each one of the F_1 offspring had a mixture of brown and white hairs. Show the nature of the offspring between F_1 generations and a white parent.

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
F_1 genotype: 2 BW, 2WW

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- nothing manner but show 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)

Characteristics of some organisms are determined by the interaction of several genes which form a **gene complex.** 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 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**

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**

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 color 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 color), 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 said to be epistatic to the gene that codes for black or brown pigment.

Mating black mice that are heterozygous for both genes (***BbCc***) follow 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 balck 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

Most genes, however, have multiple phenotypic effects, a property called **pleiotropy**. 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 color also affects the color of the coating on the outer surface of the seed, which can be gray 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 extreme are continuum of types depending on the relative proportions of dominant and recessive alleles. Poly genes give rise to continuous variation.

PEDIGREE ANALYSIS

Pedigree charts are diagrams that show the phenotypes and/or genotypes for a particular organism, its ancestors, and descendants. While commonly used in human families to track genetic diseases, they can be 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

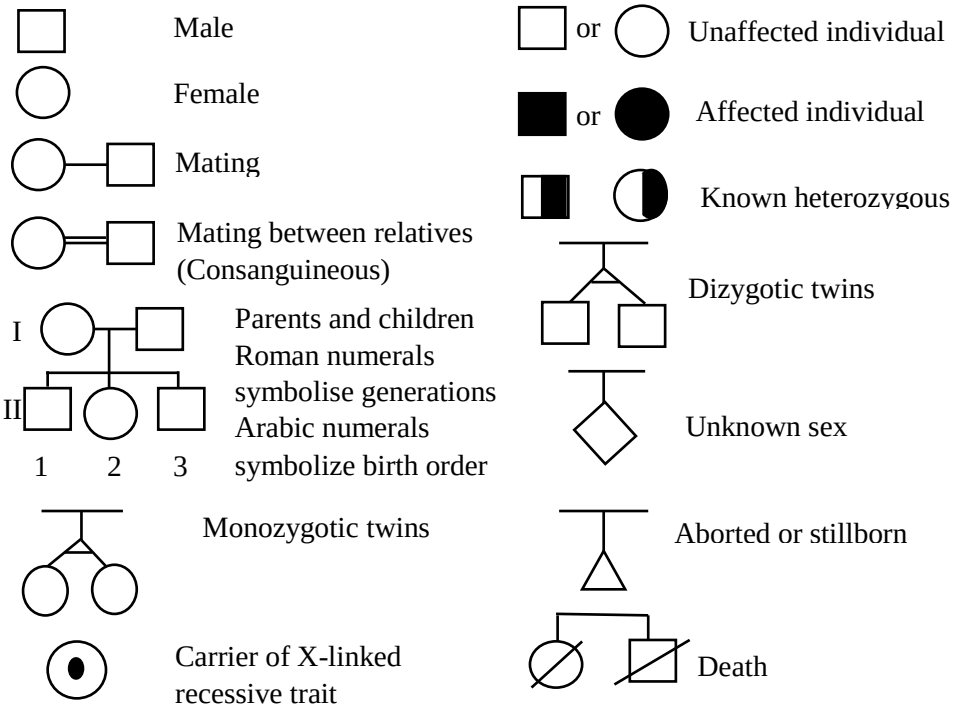
Pedigree analysis is therefore an important tool in basic research, agriculture, and genetic counseling.

Each pedigree chart represents all of 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

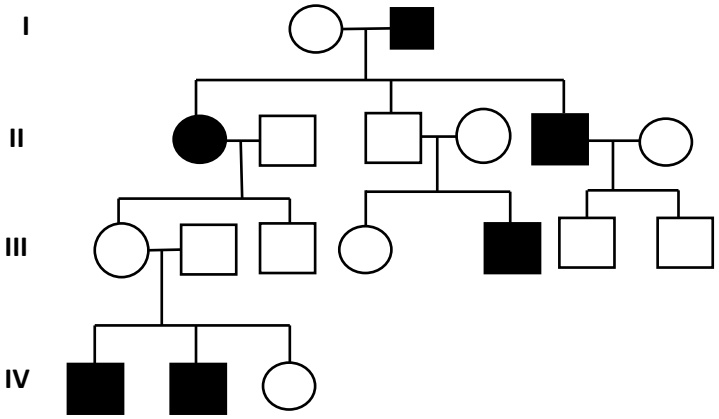
The affected individual that brings the family to the attention of a geneticist is called the **proband** (or propositus). If the individual is unaffected, they are called the **consultand**. If an individual is known to have symptoms of the disease (**affected**), the symbol is filled in. Sometimes 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 in heritance is most likely. This information is essential in calculating the probability that the trait will be inherited in any future offspring. We mostly consider five major types of inheritance: autosomal dominant (AD), autosomal recessive (AR), X-linked dominant (XD), X-linked recessive (XR), and Y-linked (Y).

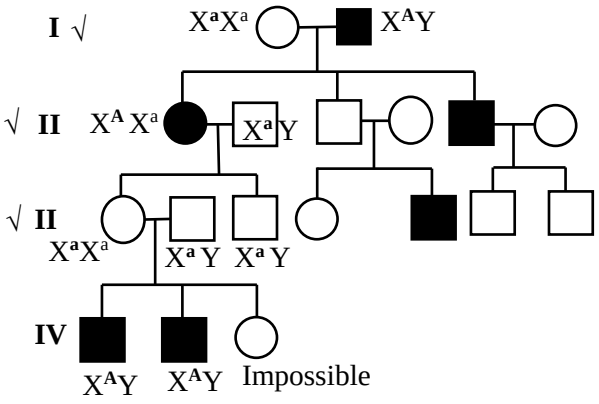
Pedigree analysis symbols



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



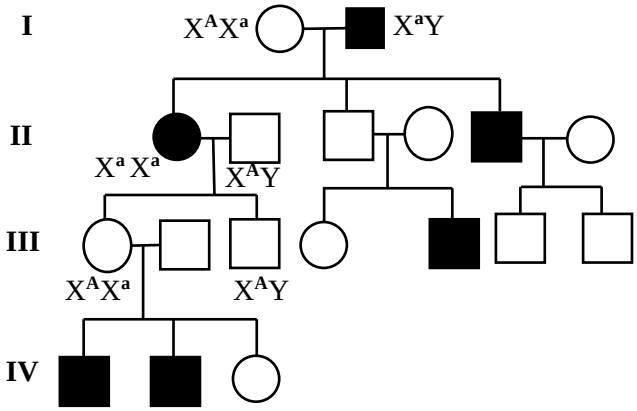
Assuming the pedigree below shows a trait inherited as a sex linked dominant trait



(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



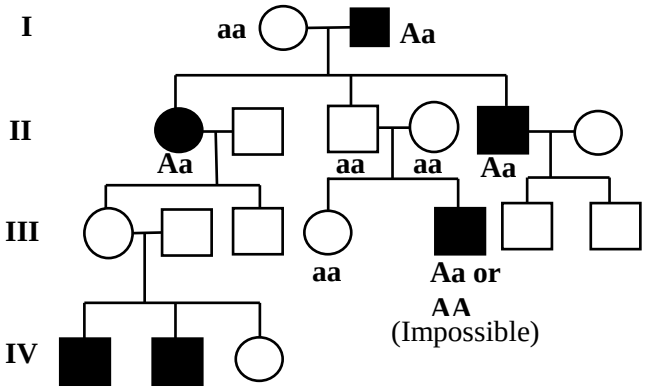
(i) For sex linked recessive trait the male in generation I must be X^aY

(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

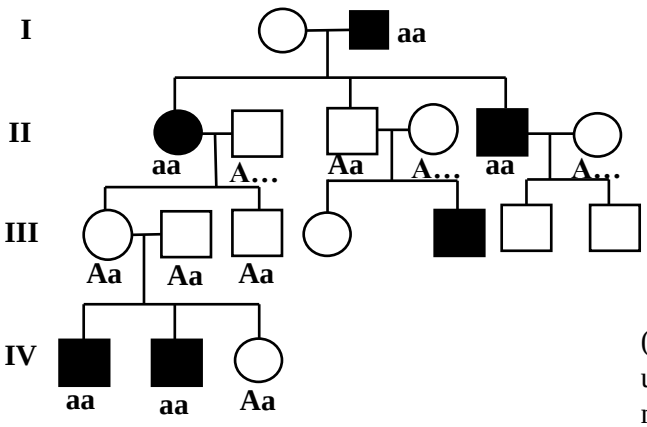


(i) For the Male in generation **I** to have the trait must have at least one dominant allele (**A**)

(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

VARIATION

This is the difference in characteristics shown by organisms belonging to the same natural population or species. Whilst the phenotypic appearance of any characteristic is ultimately determined by the genes controlling that characteristic, the extent to which certain characteristics develop may be more influenced by the environment**.**

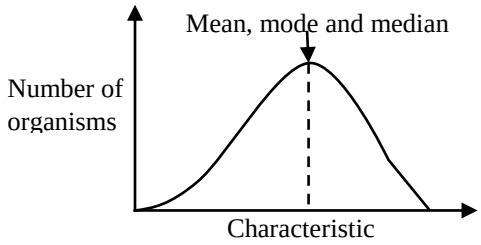
Types of variations

1. Discontinuous
2. Continuous variation

Discontinuous variation: There are certain characteristics within a population which exhibit a limited form of variation. Variation in this case produces individuals showing clear- cut differences with no intermediates between them, such as blood groups in humans, wing lengths in *Drosophila*, melanic and light forms in *Biston betularia*, style length in *Primula* and sex in animals and plants. Characteristics showing discontinuous variation are usually controlled by one or two major genes which may have two or more allelic forms and their phenotypic expression is relatively unaffected by environmental conditions.

Since the phenotypic variation is restricted to certain clear-cut characteristics, this form of variation is alternatively known as **qualitative inheritance,** as opposed to **quantitative inheritance** which is characteristic of continuous variation.

Continuous variation: Characteristics in a population shows a complete gradation from one extreme to the other without any break. There is no clear cut difference between the characters. Examples include mass, linear dimension, shape and colour of organs and organisms. The frequency distribution for a characteristic exhibiting continuous variation is a **normal distribution curve**. Most of the organisms in the population fall in the middle of the range with approximately equal numbers showing the two extreme forms of the characteristic. Characteristics exhibiting continuous variation are produced by the combined effects of many genes (polygenes) and environmental factors. Individually each of these genes has little effect on the phenotype but their combined effect is significant



Normal distribution curve

Differences between discontinous and continous variation

Discontinuous variation	Continuous variation
Phenotype differences are distinct and separate	Phenotype differences are very slight
The differences that exist in their characteristics are qualitative and cannot be measured	The differences in their characteristics are quantitative and can be measured
Controlled by different allele of a single gene	Controlled by combined effect of many genes
The phenotypes are not affected or influenced by the environmental conditions	The phenotypes can be influenced by the environmental factors
The population shows no intermediate characteristics	The population shows intermediate characteristics

Influence of the environment

The ultimate factor determining a phenotypic characteristic is the genotype. At the moment of fertilisation the genotype of the organism is determined, but the subsequent degree of expression allowed to this genetic potential is influenced greatly by the action of environmental factors during the development of the organism. For example, Mendel's tall variety of garden pea normally attained a height of six feet. However, it would only do so if provided with adequate light, water and soil conditions. A reduction in the supply of any of these factors **(limiting factors)** would prevent the gene for height exerting its full effect. It is the genetic and environmental differences which act to produce phenotypic differences between individuals.

Sources of variation

1. **Crossing over:** This takes between chromatids of homologous chromosomes during prophase I of meiosis. This produces new linkage groups and so provides a major source of genetic recombination of alleles.
2. **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 the 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 calculable number of different chromosome combinations in the gametes.
3. **Random fusion of gametes:** Fusion of male and female gametes is completely random. Thus, any male gamete is potentially capable of fusing with any female gamete.

These sources of genetic variation account for the routine **'gene reshuffling'** which is the basis of continuous variation. The environment acts on the range of phenotypes produced and those best suited to

it thrive. This leads to changes in allele and genotypic frequencies. 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.

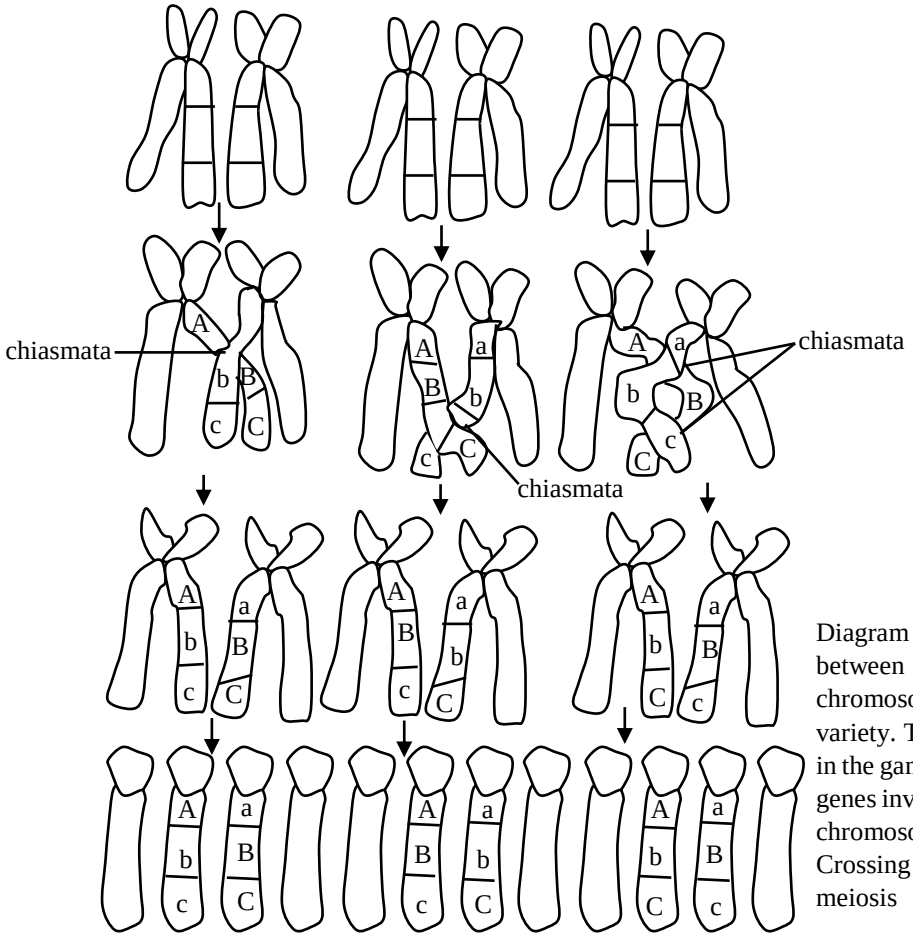


Diagram showing crossing over between the chromatids of homologous chromosomes can lead to genetic variety. The new combinations possible in the gametes depends on the number of genes involved and their positions on the chromosomes relative to the chiasmata. Crossing over takes during prophase I of meiosis

Mutation

A mutation is a change in the amount, arrangement or structure of the DNA of an organism. This leads to a change in the genotype which may be inherited by cells derived by mitosis or meiosis from the mutant cell. Mutations occurring in gamete cells are inherited, whereas those occurring in somatic cells can only be inherited by daughter cells produced by mitosis. The latter are known as **somatic mutations.**

A mutation resulting from a change in the amount or arrangement of DNA is known as **chromosomal mutation** or **chromosomal aberration.**

A change in the structure of the DNA at a single locus is known as a **gene mutation** or **point mutation.**

Causes of mutation

- X-rays and gamma they cause gene and chromosomal aberrations.
- High energy electromagnetic radiation such as ultra-violet light, they distort the structure of DNA.
- High-energy particles, such alpha and beta particles, neutrons and cosmic radiation, are also **mutagenic.**
- **C**hemical substances, including caffeine, formaldehyde, certain constituents of tobacco.
- Mustard gas, guanine in DNA is replaced by other bases.
- Colchicine, prevents spindle fibre formation during mitosis doubling the number of chromosomes.

- Nitrous acid, causes deamination of adenine in DNA to behave like guanine.
- some drugs
- Food preservatives and pesticides, have been shown to be mutagenic.
- Acridine orange, causes addition or removal of bases in DNA

Chromosome mutations

Chromosomal mutations may be the result of changes in the number or structure of chromosomes. They may affect several genes and have a more profound effect on the phenotype than gene mutations.

Changes in the number of chromosomes are usually the result of errors occurring during meiosis but they can also occur during mitosis. These changes may involve the loss or gain of single chromosomes, a condition called **aneuploidy,** or the increase in entire haploid sets of chromosomes, a condition called **euploidy (polyploidy).**

Aneuploidy

This is a condition where half the daughter cells produced have an extra chromosome $(n+1)$, $(2n+1)$ and so on, whilst the other half have a chromosome missing (n-1), $(2n-1)$ and so on. Aneuploidy can arise from the failure of a pair, or pairs, of homologous chromosomes to separate during anaphase I of meiosis. If this occurs, both sets of chromosomes pass to the same pole of the cell and separation of the homologous chromosomes during anaphase II may lead to the formation of gamete cells containing either one or more chromosomes too many or too few. This is known as **nondisjunction.** Fusion of either of these gametes with a normal haploid gamete produces a zygote with an odd number of chromosomes.

Zygotes containing less than the diploid number of chromosomes usually fail to develop, but those with extra chromosomes may develop. In most cases where this occurs in animals it produces severe abnormalities. One of the commonest forms of chromosomal mutation in humans resulting from non-disjunction is a form of trisomy called Down's syndrome (2n = 47).

Non-disjunction of the male and female sex chromosomes may also occur and produce aneuploidy affecting secondary sexual characteristics, fertility and, in some cases, intelligence.

Euploidy (polyploidy)

This is a condition where gamete and somatic cells containing multiples of the haploid number of chromosomes are called **polyploids,** and the prefixes tri-, tetra-, and so on, indicate the extent of polyploidy, for example 3nis triploid, 4n is tetraploid, 5n is pentaploid and so on.

Polyploidy is much more common in plants than in animals. For example, approximately half the 300000 known species of angiosperms are polyploid. The relatively low occurrence in animals is explained by the fact that the increased number of chromosomes in polyploids makes normal gamete formation during meiosis much more prone to error.

Since most plants are capable of propagating themselves vegetatively they are able to reproduce despite being polyploid. Polyploidy is often associated with advantageous features such as increased size, hardiness and resistance to disease.

This is called **hybrid vigour**. Most of our domestic plants are polyploids producing large fruits, storage organs, flowers or leaves.

There are two forms of polyploidy, autopolyploidy and allopolyploidy.

Autopolyploidy: This condition may arise naturally or artificially as a result of an increase in number of chromosomes within the same species. For example, if chromosomes undergo replication (during interphase) and the chromatids separate normally (during anaphase) but the cytoplasm fails to cleave (during cytokinesis), a **tetraploid** $(4n)$ cell with a large nucleus is produced. This cell will undergo division and produce tetraploid cells. The amount of cytoplasm in these cells increases to preserve the ratio of the volumes of nucleus: cytoplasm and leads to an

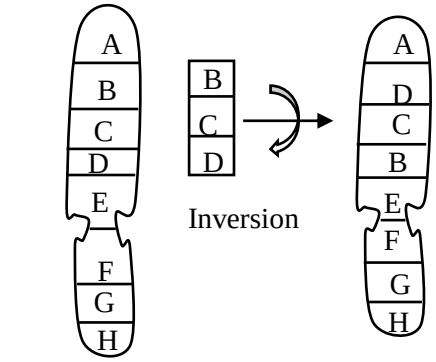
increase in the size of the whole plant or some part of it. Autopolyploidy can be induced by the use of a drug called **colchicine** which is extracted from the corm of the autumn crocus (Colchicum). Colchicine and related drugs have been used in the breeding of certain varieties of economically important crops such as tobacco, tomatoes and sugarbeet. Autopolyploids can be as fertile as diploids if they have an even number of chromosomes sets.

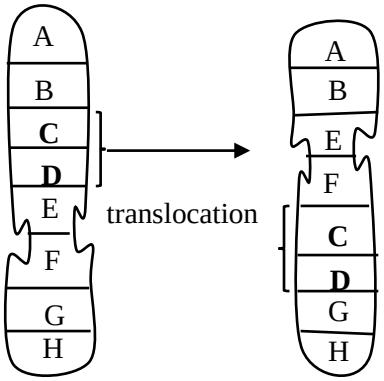
Allopolyploidy: This condition arises when the chromosome number in a sterile hybrid becomes doubled and produces fertile hybrids. F_1 hybrids produced from different species are usually sterile since their chromosomes cannot form homologous pairs during meiosis. This is called **hybrid sterility.** However, if multiples of the original haploid number of chromosomes, for example $2(n_1 + n_2)$, $3(n_1 + n_2)$ and so on (where n_1 and n_2 are the haploid numbers of the parent species) occur, a new species is produced which is fertile with polyploids like itself but infertile with both parental species.

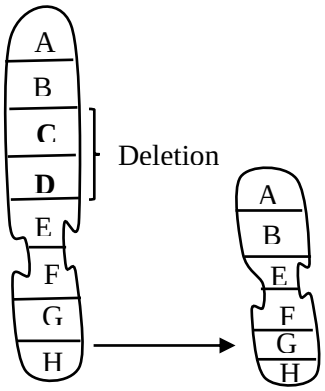
Structural changes in chromosomes

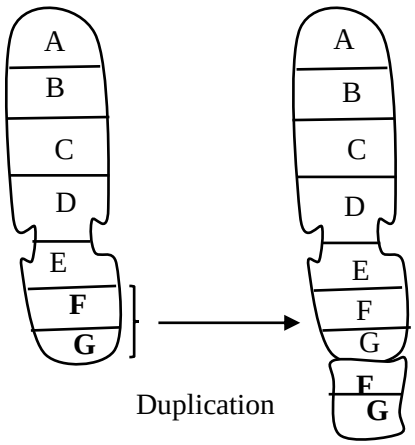
1. **Inversion** occurs when a region of a chromosome breaks off and rotates through 180° before rejoining the chromosome. No change in genotype occurs as a result of inversion but phenotypic changes may be seen. This suggests that the order of gene loci on the chromosome is important, a phenomenon known as the **position effect.**
2. **Translocation** involves a region of a chromosome breaking off and rejoining either the other end of the same chromosome or another non-homologous chromosome. The position effect may again be seen in the phenotype. Reciprocal translocation between non-homologous chromosomes can produce two new homologous pairs of chromosomes.
3. **Deletion:** This involves the loss of a region of a chromosome, either from the ends or internally. This results in a chromosome becoming deficient in certain genes. Deletion can affect one of a homologous pair of chromosomes, in which case the alleles present on the non-deficient chromosome will be expressed even if recessive. If deletion affects the same gene loci on both homologous chromosomes the effect is usually lethal.
4. **Duplication**. A region of a chromosome becomes duplicated so that an additional set of genes exists. The additional region of genes may be incorporated within the chromosome or at one end of the chromosome, or become attached to another chromosome.

Let: A, B, C…..represent loci of different genes









Gene mutation

This is a change in the nucleotide sequence of the DNA molecule in a particular region of the chromosome. Such a change in the base sequence of the gene is transmitted to mRNA during transcription and may result in a change in the amino acid sequence of the polypeptide chain produced from it during translation at the ribosomes.

Types of gene mutation

Duplication, insertion, deletion, inversion or substitution of bases. In all cases they change the nucleotide sequence and result in the formation of a modified polypeptide.

Effects of gene mutations

Gene mutations occurring during gamete formation are transmitted to all the cells of the offspring and may be significant for the future of the species. Somatic gene mutations which arise in the organism are inherited only by those cells derived from the mutant cells by mitosis.

Whilst they may affect that organism, they are lost on the death of the organism. Somatic mutations are probably very common and go unnoticed, but in some cases they may produce cells with an increased rate of growth and division. These cells may give rise to a tumour which may be **benign** and not affect other tissues, or **malignant,** which live parasitically on healthy cells, a condition known as **cancer.**

The effects of gene mutation are extremely variable. Most minor gene mutations pass unnoticed in the phenotype since they are recessive, but there are several cases where a change in a single base in the genetic code can have a profound effect on the phenotype. **Sickle cell anaemia** in humans is an example of **base substitution** mutation affecting a base in one of the genes involved in the production of haemoglobin.

Sickle cell anaemia

The major characteristics of the disease are anaemia and a tendency of the red blood cells to change shape (sickle) at low oxygen concentrations. The sickle cells tend to jam in capillaries and small blood vessels and prevent normal blood flow.

- Affects kidneys and joints.
- blocking of blood vessels causes pain in the arms, legs, back and stomach
- Joints may become stiff and painful and hands and feet may swell.
- Poor growth and development and are more prone to infections.

Cause

Haemoglobin from sickle cell anaemia sufferers, **HbS**, is different from that of normal adult haemoglobin, **HbA**. Charge on **HbS** is positive whereas it is negative on **HbA**. Difference is due to a single amino acid and since then the entire amino acid sequence of **HbA** and **HbS** has been determined. Haemoglobin is made of four polypeptide chains, two a-chains which are 141 amino acids long and two beta-chains which are 146 amino acids long. The fault occurs at the sixth amino acid in the beta-chain. The amino acid should be glutamic acid. In **HbS** however it is replaced by valine. Glutamic acid carries a negative charge and is polar whereas valine is non-polar and hydrophobic. The presence of valine makes deoxygenated **HbS** less soluble. Therefore when **HbS** loses its oxygen the molecules come out of solution and crystallise into rigid rod-like fibres. These change the shape of the red cell, which is normally a flat circular disc. The reason

for the changed amino acid is a change, or mutation, in the **DNA** coding for the amino acid.

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

If two people suffering from sickle cell trait (carriers of sickle cell anaemia) have children, there is a 1 in 4 chance of any given child being a sufferer of sickle cell anaemia.

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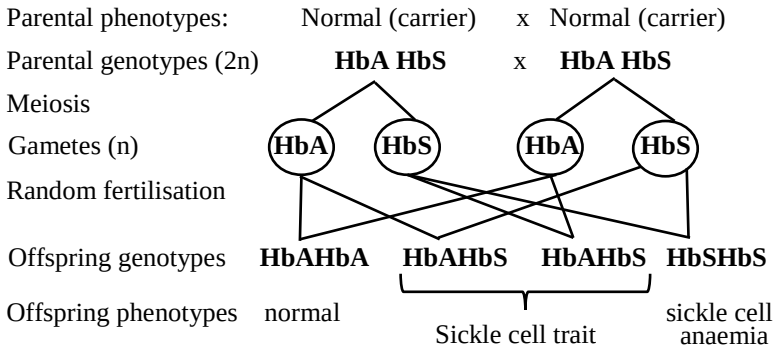
Someone carrying the faulty gene is far less susceptible to malaria (the malaria parasite multiplies inside normal red blood cells). Although homozygous sufferers often die before reproductive age, heterozygous carriers have a **selective advantage** over non- carriers and so are more likely to survive and pass on their genes to the next generation.



Genetic diagram showing the possible children of two people suffering from sickle cell trait

Cystic fibrosis (CF)

This is a genetic disorder that affects mostly the lungs, but also the pancreas, liver, kidneys, and intestine.

The cause is a recessive mutation in a gene located on chromosome 7. The gene codes for a chloride channel which is a protein, 1480 amino acids long, and known as **CFTR (cystic fibrosis transmembrane regulator).** It allows diffusion of chloride ions into and out of epithelial cells and is located in the cell surface membranes of these cells. In CF sufferers it does not function. Since the gene is recessive, CF sufferers are homozygous and have two copies of the faulty gene.

The cause of the problem is the deletion of three base pairs from the gene; codon number 508 in the mRNA is therefore missing. As a result the amino acid phenylalanine (F) is missing at position 508 in the protein. The mutation is therefore called ΔF508.

Symptoms

Mucus becomes abnormally thick and sticky because the normal outward flow of chloride ions from the cells is prevented. Chloride ions are negatively charged, so in order to balance the negative charge which builds up in the cells more sodium ions enter. The high ion concentration inside the cell in turn prevents water from leaving the cell. The parts of the body most affected are the lungs, pancreas and liver.

- In the pancreas fibrous patches, called cysts, develop which give the disease its name.
- The thick mucus clogs up the airways of the lungs, and the branches of the pancreatic duct and the bile duct from the liver into the gut.
- Repeated lung infections are caused, as well as digestive problems, including poor release of pancreatic enzymes and poor absorption of digested food.

- The intestine may also become obstructed.
- Males are almost always infertile and females are frequently infertile.
- Sweat is saltier than usual because the sweat duct is relatively impermeable to chloride ions and once again sodium follows the chloride.

Phenylketonuria (PKU)

This is a condition in which the body can't break down an amino acid called phenylalanine. Amino acids are used to build protein in the body. Without treatment, phenylalanine builds up in the blood and causes health problems.

Cause

PKU is a recessive, autosomal condition. It is a very distressing condition, but fortunately early diagnosis and treatment can prevent damage to health.

The disease is due to an inability to convert the amino acid phenylalanine to another amino acid, tyrosine:

The enzyme **phenylalanine hydroxlase** (PAH) is normally present in the liver, but is faulty in sufferers from PKU. The gene for this enzyme is on chromosome 12. As a result of faulty PAH, phenylalanine builds up in the body. The excess is converted to toxins which affect mental development. Affected children appear normal at birth because, while in their mother's uterus during pregnancy, excess phenylalanine moves across the placenta and is removed by the mother's liver

- severe mental retardation
- Patients have IQs of less than 20.
- hyperactive and irritable behaviour in children; awkward posture and walk
- lighter skin pigmentation and fair hair (because tyrosine is normally used in the synthesis of the brown skin pigment melanin); dry, rough skin (eczema)
- repetitive movements of the fingers, hands or entire body
- convulsions due to abnormal brain activity.

Huntington's chorea (HC)

This is an inherited disorder that results in death of brain cells. It is caused by an autosomal mutation which is *dominant* the gene was located on chromosome 4

The function of the protein it codes for is unknown, although it has been given a name, 'huntingtin'.

The disease causes progressive deterioration of brain cells and gradual loss of motor control (control of voluntary muscle by motor nerves) resulting in uncontrollable shaking and dance-like movements. This accounts for the use of the term 'chorea', [r] meaning dance, to describe the disease. Intellectual ability is lost, hallucinations, slurring of speech, mood changes, personality changes and memory loss (temporary or permanent) may all occur. The brain shrinks between 20-30% in size.

Down's syndrome

Also known as trisomy 21, is a genetic **disorder** caused by the presence of all or part of a third copy of chromosome 21. It is typically associated with physical growth delays, mild to moderate intellectual disability, and characteristic facial features.

Down's syndrome is named after a physician John Langdon Down who worked at an asylum in Surrey, England and who in 1866 was the first to describe the condition. The presence of three copies of a chromosome is known as **trisomy,** hence Down's syndrome is also known as **trisomy 21.**

Symptoms

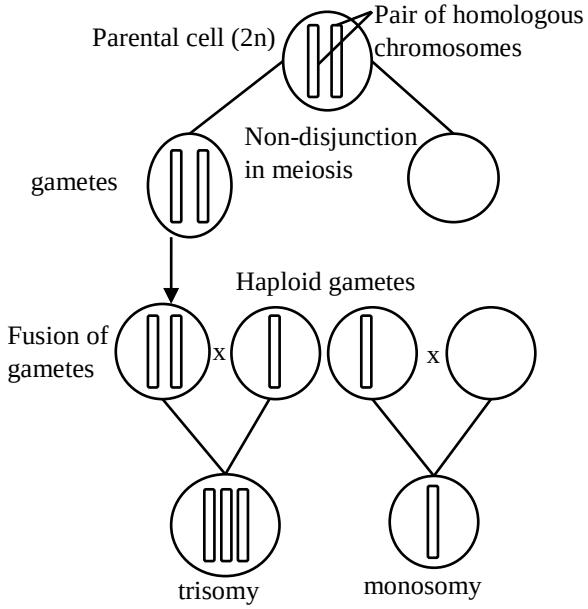
- Eyelids which apparently slant upwards clue to a fold of skin over the inner corner of the eye.
- The face is typically flat and rounded.
- Mental retardation, often severe.
- Short stature and relatively small skull due to poor skeletal development.
- Heart defects occur in about one-quarter of Down's children.
- Increased risk of infection, particularly respiratory and ear infections; coarse, straight hair.

- squat hands with a characteristic crease which runs all the way across the palm;
- Intestinal problems and leukaemia arc slightly more common than normal.

During meiosis II chromatids fail to separate, two chromosomes or two chromatids enters one daughter cell and none enters the other.

About 3-4% of Down's syndrome cases are due to a type of mutation known as a translocation

Chromosome 21 is translocated (moved) to chromosome 14 or, less commonly, to chromosome 22. An even less common cause is a 21 to 21 translocation.



Klinefelter's syndrome

Also known as 47, XXY or XXY, is the set of symptoms that result from two or more X chromosomes in males. The primary features are infertility and small testicles. Often, symptoms may be subtle and many people do not realize they are affected.

This is due to an extra X chromosome. The genotype is therefore XXY instead of the normal XY and the sufferer has 47 chromosomes instead of 46. It is an example of trisomy. It may occur during spermatogenesis (sperm production) in the male parent or during oogenesis (egg production) in the female parent. As a result of non-disjunction in the male sex chromosomes, equal numbers of zygotes will contain only one X chromosome and no Y chromosome (represented as XO). In the female, XXX and YO zygotes are also created.

XXX women tend to be slightly taller. YO zygotes do not develop because many vital genes are missing completely.

Typical symptoms of these and similar patients are as follows:

1. infertility - sperm are never produced, although erection and ejaculation are possible;
2. usually taller than average;
3. some breast development, although not necessarily very obvious;
4. smaller testes than normal, although this is not necessarily obvious;
5. higher than usual FSH secretion for males (FSH is follicle stimulating hormone and is produced by the pituitary gland in both men and women;
6. trunk may show signs of obesity (eunuch-like appearance);
7. little facial hair;
8. voice pitched higher than normal;
9. Educational difficulties and behavioural problems are fairly common.

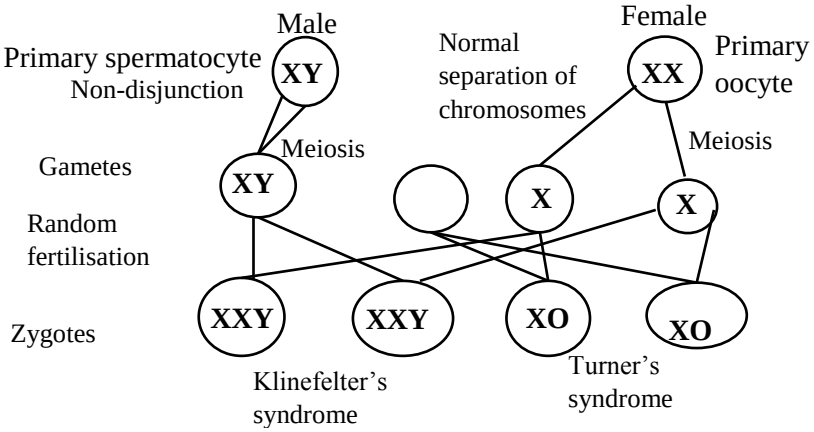
Turner's syndrome

This is a chromosomal condition that affects development in females

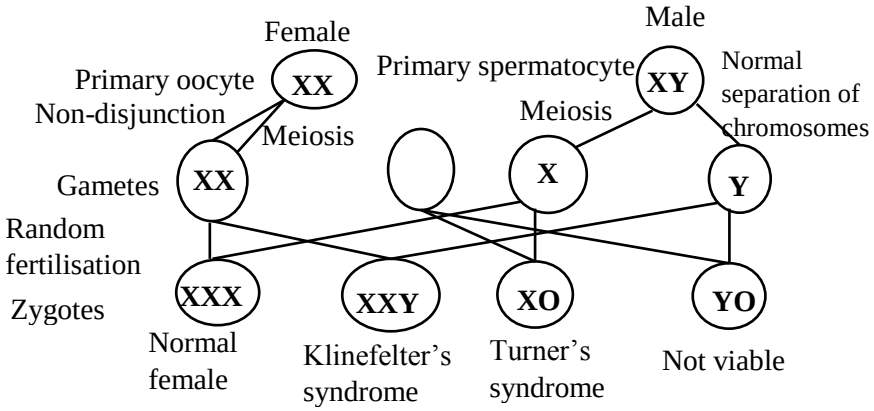
Patients can best be described as incompletely developed females, although there are often no obvious external differences compared with normal females. Typical symptoms are as follows

- Infertility - ovaries are absent (represented only as connective tissue)
- Shortness of stature, averaging 1.5 m (less than 5 feet)
- Small uterus
- Webbed neck may occur
- Puffy fingers with deep set finger nails which arc more convex than normal
- The hair line (line at which hair starts to grow) at the back of the head is lower than normal.

It is as a result of missing X chromosome. The genotype is therefore XO instead of the normal XX and the sufferer has 45 chromosomes instead of 46. This is an example of monosomy as a result of non-disjunction during meiosis



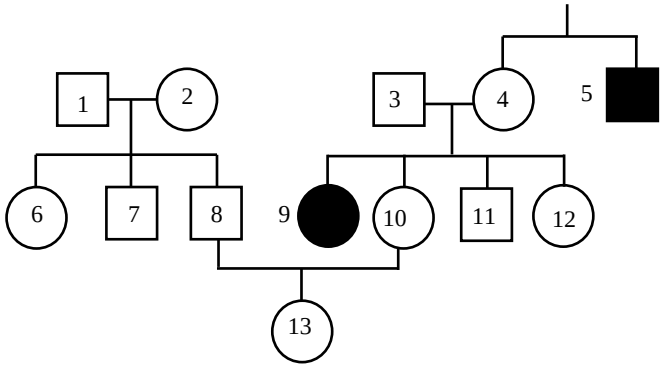
Non-disjunction of Father's sex hormones



Non-disjunction of the mother's sex chromosomes

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 carrires?
Individuals 1, 2, 6, 7, 8, 10, 11, 12 and 13 could all be carrires. 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 carrires?
50% since a ratio 1 affected: 2 carrier: 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 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).

Answer

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 flower, long stem 49 red flower, short stem 45 red flower, 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 are 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 x tortoiseshell
Male coat female

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

meiosis

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

Random fertilisation

F_2 genotypes(2n) X^BX^G X^BX^B X^GY X^BY

F_2 phenotypes

1 X^BX^G tortoiseshell-coat colour female
1X^BX^B black-coat colur female
1 X^GY ginger coat colour male
1 X^BY 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)	$\mathbf{I^AI^O}$	x	$\mathbf{I^BI^O}$
Meiosis			
Gametes(n)	$\mathbf{I^A}$ $\mathbf{I^O}$	x	$\mathbf{I^B}$ $\mathbf{I^O}$
Random fertilisation			
Offspring genotypes	$\mathbf{I^AI^B}$ $\mathbf{I^AI^O}$		$\mathbf{I^OI^B}$ $\mathbf{I^OI^O}$
Offspring phenotypes blood groups	AB A		B O

(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 white.

(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)	**SB** **Sb** **sB** **sb**	x	**SB** **Sb** **sB** **sb**

Gametes		**SB**	**Sb**	**sB**	**sb**
Random fertilistion	**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 hiar: 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) **RS** **Rs** **rS** **rs** x **RS** **Rs** **rS** **rs**

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, 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) X^{nR} X^{Nr} X^{nr} X^{NR} x X^{nr} Y

Random fertilisation	♂ / ♂	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 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 com

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**)

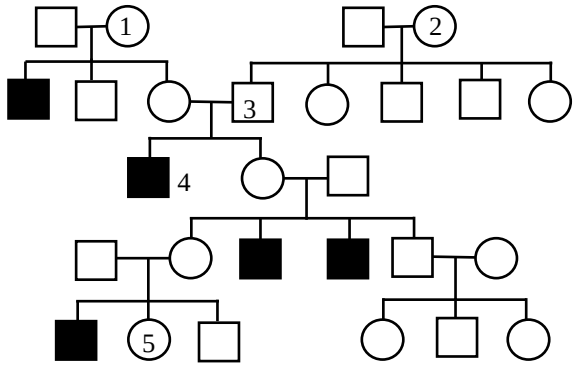
Ransdom fertilisation		**WB**	**Wb**	**wB**	**wb**
	WB	**WWBB** white	**WWBb** white	**WwBB** white	**WwBb** white
	Wb	**WWBb** white	**WWbb** white	**WwBb** white	**Wwbb** white
F_2 genotypes	**wB**	**WwBB** white	**WwBb** white	**wwBB black**	**wwBb black**
	wb	**WwBb** white	**Wwbb** white	**wwBb** 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 throughfive generations.



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

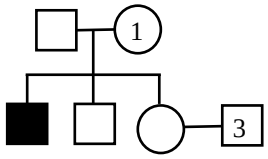
(ii) The disorder is caused bya recessive allele of a single gene. Using the symbol A to represent the normal allele and a to represent the recessive allele, write down the most likely genotypes of individuals 1, 2, 3 and 4 (04 marks)

(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 probality that he will have the disorder? (01 mark)

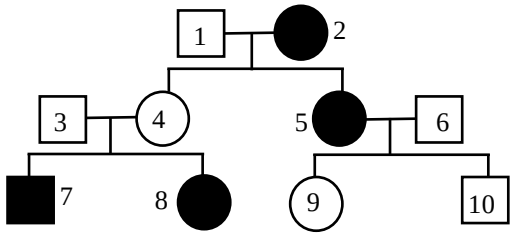
(ii) If individual 5's first child is a girl,what is the probability that she will have the disorder? (01 mark)

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



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



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

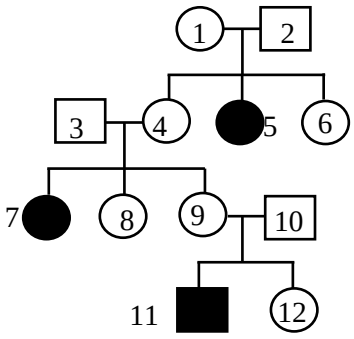
(b) What is the probability that the next child born to inviduals 3 and 4 would have at least one allele for cystic fibrosis? Explain.

(02 marks)

(c) In britain, 1 in 2000 people are 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 defeat which is not linked.



(a) State the numbers of all the individuals in the family line that are 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 anemia 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 green and smooth seed coat is dominant over wrinkled. When yellow smooth beans were crossed with green wrinkled beans, all F_1 had 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 allele, 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 woild 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 hilums was crossed with a pure-breeding variety with yellow seeds and white hilums. All the F_1 plants had yellow seeds and black hilums. When these were

allowed to self-fertilise, the plants of the F_2 generation produced the following seed.
Yellow seeds with white hilums 31
Yellow seeds with black hilums 93
Green seeds with white hilums 8
Green seeds with black hilums 28

(a) What charactsristcs are dominat 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) Abreeder crossed a male black mouse with a female brown one. The offsrping produced showed four different coat colurs, 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 colur 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 alleles. The dominant allele **A** gives a purple colur 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 isalso affected by a 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) allhad black-hulled grains. Allowing the F_1 plants to self-fertilse 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 ratiois suggested from the figures given? (01 mark)

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

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

14. In the fruit fly *Drosophila*, 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 have 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 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 lethel 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