chapter 15

Principles of Inheritance and Variation

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Introduction

You have studied in the previous section, one of the most fundamental attributes of all living organisms is reproduction. Progeny receives the characters from parents in the form of egg and sperm. Because of this feature, the progeny resembles its parents. For example, a mango seed forms only a mango plant and not any other plant, and an elephant always gives birth only to a baby elephant and not some other animal. This has been best summed up in the phrase '**like begets like**'. The transfer of characters from parents to offspring is known as **inheritance**. Progeny produced resembles the parents closely but is not identical in all the respects. The reason behind is **variation**. Variation is the degree by which progeny differ from their parents. The branch of science which deals with **the inheritance as well as the variation of characters from parents to offspring is Genetics.**

Early agriculturists (8,000 –10,000 B.C.) knew that one of the causes of variation was hidden in sexual reproduction. Because of it, they successfully bred domesticated varieties from wild plants and animals through selective crossing and artificial selection. Indian cow (*e.g.*, Sahiwal of Punjab) is domesticated form of an ancestral wild cow. However, our ancestors had very little idea about the scientific basis of inheritance and variation.

MENDEL'S LAWS OF INHERITANCE

Gregor Johann Mendel was the first to demonstrate the scientific basis of inheritance and variation by conducting hybridisation experiment. But it should be very much clear that he was not the first to conduct these experiments, rather he was the first to consider one to three characters at one time and this was perhaps the secret of his success. His experiments were in fact the extension and development of hybridisation experiments on pea conducted by earlier workers like Knight and Goss.

Mendel was born on July 22, in 1822. He worked on *Pisum sativum* (Garden pea or Edible pea) for 7 years (1856–1863) and proposed the law of inheritance in living organisms.

Before discussing, why did Mendel select pea plant for genetics experiment, we must know what is the difference between character and trait.

Character	Trait
Feature of the individual. *e.g.*, Stem height	Distinguishable feature of a character and its detectable variant. *e.g.*, Tallness or dwarfness.

Selection of pea plant : The main reasons for adopting garden pea for experiments were as follows :

(1) Pea has many distinct alternative traits (clear contrasting traits).

(2) It produces a large number of seeds and completes its life cycle in one season.

(3) Flowers show self (bud) pollination, so are true breeding.

(4) It is easy to artificially cross-pollinate the pea flowers. The hybrids thus produced were fertile.

Mendel conducted artificial pollination or cross-pollination experiments using several ***true-breeding pea lines***. A true breeding line is one that having undergone continuous self-pollination, shows the stable trait inheritance and expression for several generations. , near homozygous & produces offspring of its own type.

Mendel selected 14 true-breeding pea plant varieties, as pairs which were similar except for one character with contrasting traits. It means, Mendel selected 7 characters in pea plant for carrying out hybridisation experiments. These characters are listed in following table.

S.No.	Character	Contrasting traits	
		Dominant trait	Recessive trait
1.	Seed shape	Round	Wrinkled
2.	Seed colour	Yellow	Green
3.	Flower colour	Violet	White
4.	Pod shape	Full/Inflated	Constricted
5.	Pod colour	Green	Yellow
6.	Flower position	Axial	Terminal
7.	Stem height	Tall	Dwarf

Did You Know?

1. Initially Mendel took 34 varieties of pea plants, then 22 but ultimately worked with only 7 pairs of varieties.
2. Term 'Pure line' was coined by Johannsen in 1900.

Reasons for Mendel's Success

(1) Mendel applied statistical method and mathematical logic for analysing his results.

(2) He kept accurate records of his experiments, giving all the details of number and type of individuals, which are a necessity in the genetic studies.

(3) Mendel experimented on a number of plants for the same trait and obtained hundreds of offspring. A large sampling size gave credibility to his results. Chances of error are little in large samples.

(4) He tried to formulate theoretical explanations for the observed results. These explanations were further tested by conducting experiments for successive generations of the test plants, that proved his results pointed to general rules of inheritance rather than being unsubstantiated ideas.

Example 1 : *State True or False.*

(1) Garden pea has seven characters only.

(2) Flowers of Pisum sativum naturally show cross pollination.

(3) A true breeding line shows the stable trait inheritance.

(4) Mendel applied statistical methods and mathematical logic for analysing the results.

Solution : (1) False

(2) False

(3) True

(4) True

Try Yourself

1. **Fill in the blanks :**
   (i) _____ is the degree by which progeny differs from their parents.
   (ii) *Pisum sativum* produces a _____ number of offspring and completes its life cycle in _____ season.
2. **State True or False :**
   (i) The transfer of characters from parents to offspring is known as inheritance.
   (ii) A true breeding line shows stable trait inheritance and expression for several generations.
   (iii) In total, Mendel selected 7 true-breeding pea plant varieties.

EXERCISE

1. The degree by which progeny differ from their parents is known as
   (1) Genetics
   (2) Variation
   (3) Heredity
   (4) Inheritance
2. Sahiwal cows of Punjab are developed by
   (1) Artificial selection
   (2) Domestication
   (3) Both (1) & (2)
   (4) Mutation
3. Which of the following genotype represents heterozygous condition?
   (1) TT
   (2) tt
   (3) Tt
   (4) RR
4. How many true breeding pea plant varieties were selected by Mendel?
   (1) 14
   (2) 7
   (3) 21
   (4) 2
5. Mendel selected *Pisum sativum* for hybridisation experiments because of
   (1) Clear contrasting characters and short life span
   (2) Long life span and non-fertile hybrids
   (3) Presence of unisexual flowers
   (4) Infertile hybrids and production of large number of seeds by each plant
6. Mark the odd one (w.r.t. dominant trait in garden pea)
   (1) Yellow pod
   (2) Inflated pod
   (3) Axial flower
   (4) Yellow seed
7. Transmission of genetic characters from parents to offspring is
   (1) Variation
   (2) Heredity
   (3) Blending
   (4) Somatoplasm
8. Who coined the term 'allele'?
   (1) Saunders
   (2) Bateson
   (3) Johannsen
   (4) Mendel
9. Which of the following trait of garden pea is present on 7^{th} chromosome?
   (1) Pod shape
   (2) Pod colour
   (3) Seed shape
   (4) Stem height
10. All traits can express themselves in heterozygous condition, except
    (1) Tall
    (2) Violet
    (3) Axial
    (4) Wrinkled seed

INHERITANCE OF ONE GENE

Study of inheritance of single pair of contrasting traits of a character at a time is called **one gene inheritance**. Mendel crossed true breeding tall variety (6–7 ft.) and true breeding dwarf variety (0.75–1 ft.) pea plants to study the inheritance of one gene. The plants used in initial cross are referred to as **P_1 and P_2 or parents**. Since pea is self-fertilising, the anthers should be removed from the female parent before maturity for the purpose of cross pollination. The method of removal of anthers from bisexual flowers of female parent plant is called **emasculation**. The pollens, then at the dehiscence stage, is brought from the male parent and is dusted on the stigma of emasculated flower. He collected the seeds produced as a result of this cross and grew them to generate plants of the first hybrid generation. This generation is also called the **filial$_1$ (offspring) progeny** or the **F_1**.

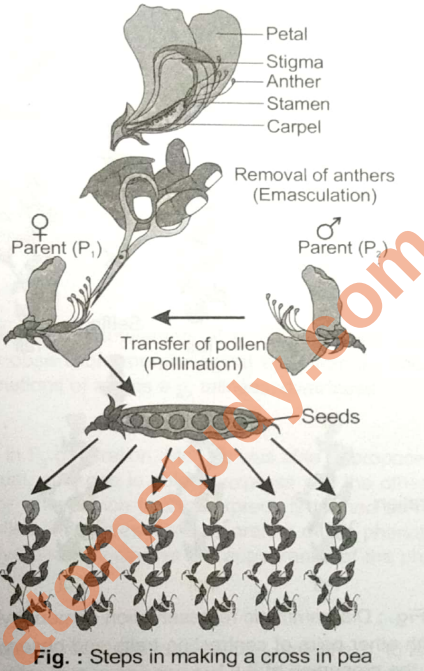


Fig. : Steps in making a cross in pea

Mendel found that all F_1 progeny plants were tall (6-7 ft), like one of its parents; none were dwarf. He made similar crosses with other pairs of contrasting traits and in every case the result was the same. In each, F_1 plants were identical to one of the parents.

S.No.	Characters	Parents		F_1 Plants
1.	Stem height	Tall	Dwarf	Tall
2.	Flower colour	Violet	White	Violet
3.	Flower position	Axial	Terminal	Axial
4.	Pod shape	Inflated	Constricted	Inflated
5.	Pod colour	Green	Yellow	Green
6.	Seed shape	Round	Wrinkled	Round
7.	Seed colour	Yellow	Green	Yellow

When Mendel self-pollinated the tall F_1 plants, both tall and dwarf plants were obtained in F_2 generation. Offspring derived from selfing of the F_1 are termed as **second filial** or **F_2 generation**. The proportion of plants that were tall were $\left(\frac{3}{4}\right)^{th}$ of the F_2 population while $\left(\frac{1}{4}\right)^{th}$ of the F_2 population were dwarf. We must note here that dwarfness which disappeared in F_1 generation, reappeared in F_2. The tall and dwarf traits in F_2 generation were identical to their parental type and **did not show any blending**, *i.e.*, all the offsprings were either tall or dwarf, **none were of in-between height**. In the example discussed here, cross is performed involving single pair of contrasting traits of a character. Such cross is known as **monohybrid cross**.

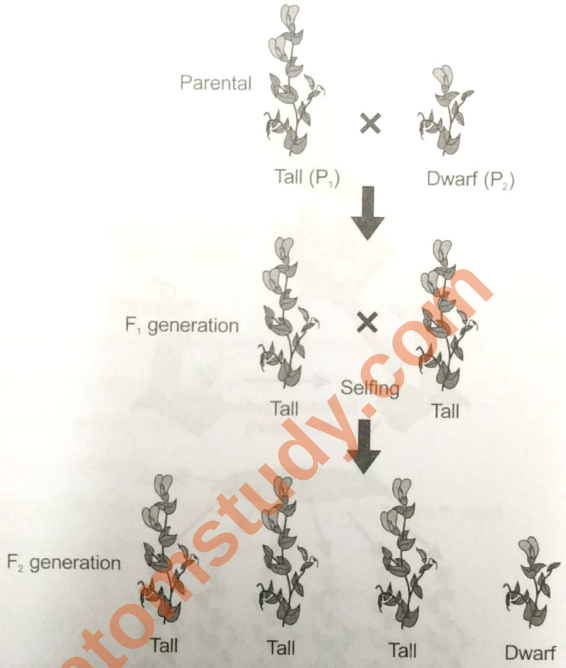


Fig. : Diagrammatic representation of monohybrid cross

He made similar crosses with other pairs of contrasting traits and observed the F_2 generation in which both the traits were expressed in the proportion of 3 : 1.

Did You Know?

1. Mendel also worked on two other plants namely, Hawkweed (*Hieracium*) and Lablab and he failed to obtain same results as found in garden pea due to parthenogenesis non-availability of pureline seeds and parthenogenesis.
2. SBE - 1 gene is responsible for the synthesis of an enzyme SBE (starch branching enzyme) essential for producing round seeds. Mutation in gene leads to failure in the production of this enzyme and hence in complex metabolic disturbances producing wrinkled seeds.

Concept of 'Factors'

Based on these observations, Mendel proposed that something was being stably passed down, unchanged, from parent to offspring through the gametes, over successive generations. He called these things as **'factors'**. We now call these factors as **"genes"**. Therefore, a gene is defined as the functional unit of inheritance. They

contain the information that is required to express a particular trait in an organism. Chemically gene is a segment of DNA that has a particular function, the common being synthesis of polypeptide.

Genes which code for a pair of contrasting traits are known as **alleles** *i.e.*, they are slightly different forms of the same gene. Therefore, term gene can be used for any factor but term allele is used with reference to another allele. We use alphabetical symbols for each gene, the capital letter is used for the trait expressed at the F_1 stage and small alphabet for the other trait. For example, if **T** is used for the 'tall' trait and **t** for 'dwarf' then **T** and **t** are alleles of each other. Therefore, in plants (Diploid) the pair of alleles for height would be **TT**, **Tt** or **tt**. We should not use **T** for tall and **d** for dwarf because we will find it difficult to remember whether **T** and **d** are alleles of the same character or not.

Did You Know?

1. Term 'gene' was given by Johannsen while term 'allele' by Bateson.
2. Alleles are the abbreviated form of the term "allelomorphs".

Homozygous and Heterozygous

Mendel proposed that in a true breeding, tall or dwarf pea variety the allelic pair of genes for height are identical, **TT** and **tt**, respectively. This condition was termed as 'homozygous' by Bateson and Saunders. An individual having two different alleles (**Tt**) will be called hybrid. Bateson and Saunders termed this condition as 'heterozygous'.

Genotype and Phenotype

Genotype is representation of genetic complement of an individual with respect to one or more characters. *e.g*, **TT**, **Tt**, **tt**. **Phenotype** is observable morphological appearance. The phenotypes of an individual is determined by different combinations of alleles *e.g*, tallness, dwarfness.

Dominant and Recessive

Based on the results obtained in F_1 generation, Mendel was able to propose that when two dissimilar factors are present in a single individual, only one is able to express and the other is not. The one that expresses itself is called **dominant** factor while which fails to express is termed as **recessive** factor. In other words we can say that a dominant allele influences the appearance of the phenotype even in the presence of an alternative allele while recessive allele influences the appearance of the phenotype only in the presence of another identical allele.

Concept of Segregation

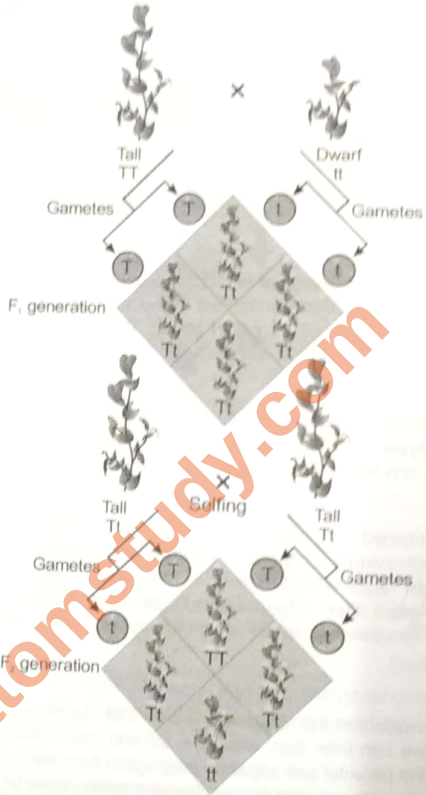
From the above observation that the recessive parental trait (dwarfness, tt) is expressed without any blending in the F_2 generation, we can infer that, when the tall and dwarf plant produce gametes by the process of meiosis, the alleles of the parental pair separate (segregate) from each other and only one allele is transmitted to a gamete. It means meiosis reduces the number of chromosomes to one half where a gamete carries only one chromosome of each type and hence only one factor of a character. The segregation of alleles is a random process and so there is a 50 percent chance of a gamete containing either allele. In this way the gametes of the tall **TT** plants have the allele **T** and the gametes of the dwarf **tt** plants have the allele **t**. During fertilisation of the two alleles, **T** from one parent through the pollen (n), and **t** from the female parent through the egg (n) are united to produce zygotes (2n) that have one **T** allele and one **t** allele *i.e.* hybrid or heterozygous **Tt** plant (2n).

Did You Know?

Type of gametes produced by a diploid individual can be calculated by using formula, 2^n. Here 'n' represents the number of heterozygotes/hybrid.

Punnett Square

The production of gametes(n) by the parents (2n), the formation of the zygotes (2n), the F_1 and F_2 plants can be understood from a diagram called **Punnett square**. Punnett square was developed by a British geneticist, Reginald C. Punnett. It is a graphical representation to calculate the probability of all possible genotypes of offspring in a genetic cross. The possible gametes are written on two sides, male in horizontal row and female in vertical column. All possible combinations are represented in boxes below in the squares, which generates a square output form.



Phenotypic ratio : tall : dwarf
3 : 1
Genotypic ratio : TT : Tt : tt
1 : 2 : 1

Fig. : A Punnett square used to understand a typical monohybrid cross conducted by Mendel between true-breeding tall plants and true-breeding dwarf plants

The Punnett square, given in above figure, shows the parental tall **TT** (male) and dwarf **tt** (female) plants, the gametes produced by them and the F_1 **Tt** progeny. The F_1 plant of the genotype **Tt** when self-pollinated, produces gametes of the genotype **T** and **t** in equal proportion. When fertilization takes place, the pollen grains of genotype **T** have a 50% chance to pollinate eggs of the genotype **T**, as well as of genotype **t**. Also pollen grains of genotype **t** have a 50% chance to pollinate eggs of genotype **T**, as well as of genotype **t**. As a result of random fertilisation, the resultant zygotes can be of the genotypes **TT**, **Tt** or **tt**. From the Punnett square it is easily seen that $1/4^{th}$ of the random fertilisations lead to **TT**, 1/2 lead to **Tt** and $1/4^{th}$ to **tt**. Due to

dominance of one trait over the other that all the F_1 are tall (though the genotype is **Tt**) and in the F_2, 3/4th of the plants are tall (though genotypically 1/2 are **Tt** and 1/4th are **TT**). This leads to a phenotypic ratio of 3/4th tall : (1/4 **TT** + 1/2 **Tt**) and 1/4th **tt**, *i.e.*, a 3 : 1 ratio, but a genotypic ratio of 1 : 2 : 1. The 1/4 : 1/2 : 1/4 ratio of **TT** : **Tt** : **tt** is mathematically condensable to the form of the binomial expression $(ax + by)^2$, that has the gametes bearing genes **T** or **t** in equal frequency of 1/2. The expression is expanded as given below :

$$(1/2\mathbf{T} + 1/2\mathbf{t})^2 = (1/2\mathbf{T} + 1/2\mathbf{t}) \times (1/2\mathbf{T} + 1/2\mathbf{t}) = 1/4\mathbf{TT} + 1/2\mathbf{Tt} + 1/4\mathbf{tt}$$

Mendel self-pollinated the F_2 plants and found that dwarf F_2 plants continued to generate dwarf plants in F_3 and F_4 generations. He concluded that the genotype of the dwarfs was homozygous *i.e.*, **tt**.

Test Cross

From the preceeding paragraphs it is clear that though the genotypic ratios can be calculated using mathematical probability, by simply looking at the phenotype of a dominant trait, it is not possible to know the genotypic composition. For example, whether a tall plant froms F_1 or F_2 has **TT** or **Tt** composition, can not be predicted. Therefore, to determine the genotype of a tall plant at F_2, Mendel crossed the tall plant from F_2 with a dwarf plant. This is called a **test cross**. In a typical test cross, an organism showing a dominant phenotype is crossed with the recessive parent instead of self-pollination. The progenies of such a cross can be easily analysed to predict the genotype of test organism.

Example : If we want to determine the genotype of a violet-flowered pea plant (test organism), then it is crossed with the recessive parent (white-flowered pea plant) instead of self crossing.

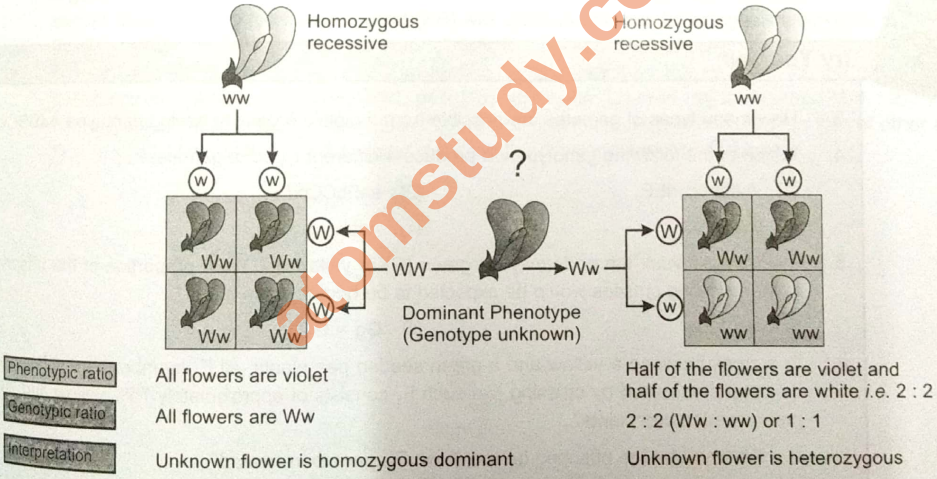


Fig. : Diagrammatic representation of a test cross

Example 2 (a) : *How many types of gametes can be produced by a diploid organism, if it is heterozygous for one locus? Also mention genotypes of gametes.*

Solution : Types of gametes = 2^n

$\because$ Genotype of organism is Aa

$\therefore$ n = 1

$\therefore$ $2^1 = 2$

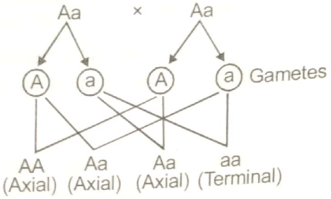
i.e., 2 type of gametes (A, a)

Example 2 (b) : *In garden pea the flowers may be axial (A) or terminal (a) in position. What proportion of the offspring in the following crosses would be expected to be axial?*

(i) $Aa \times Aa$

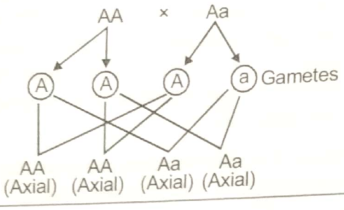
(ii) $AA \times Aa$

Solution : (i)



$\therefore$ Axial flowered plants $= \frac{3}{4}$ *i.e.* 75%

(ii)



$\therefore$ Axial flowered plants $= \frac{4}{4}$ *i.e.* 100%

Try Yourself

3. How many types of gametes are possible from a diploid organism having genotype AaBBCC?
4. Which of the following genotype will produce 4 different types of gametes?
   (1) AAbbccddEE (2) aaBbCCdd
   (3) AaBbCC (4) Aabb
5. In *Pisum sativum*, the pods may be green (G) or yellow (g). What proportion of the offspring in the following crosses would be expected to be homozygous green?
   (i) Gg × gg (ii) Gg × GG
6. In a cross between a yellow and a green seeded pea plants, all F_1 members are yellow. But F_2 generation raised by crossing two such F_1 consists of approximately 75% yellow and 25% green seeded pea plants.
   (i) What will be the offspring be like if two F_2 greens are mated?
   (ii) What will be the genotypic ratio in the population of yellow-seeded plants in F_2 generation?

Did You Know?

Mendel simply described his results and drew certain conclusions. Carl Correns gave these conclusions the shape of laws.

On the basis of his observations on monohybrid cross, Mendel proposed two general rules. Today these rules are called the principles or **Laws of Inheritance**: the first law or **Law of Dominance** and the second law or **Law of Segregation**.

1. **Law of Dominance:** Mendel experimented with garden pea for seven characters. In each case he found that:
   (i) Every character is controlled by discrete units called factors.
   (ii) The factors occur in pairs.
   (iii) In a dissimilar pair of factors (*e.g.* Tt), only one is able to express its effect that called as dominant factor. The other factor which does not show its effect is known as recessive factor.

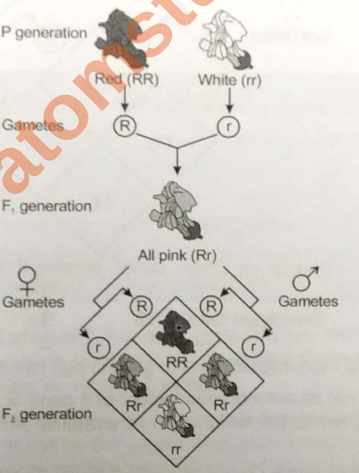
   The law of dominance is used to explain the expression of only one of the parental traits in a monohybrid cross in the F_1 and the expression of both in the F_2. It also explains the proportion of 3 : 1 obtained in F_2 generation. This law is not universally applicable.

2. **Law of Segregation** : This law is based on the fact that the two factors of a character present in an individual do not get mixed up (blending) and both the traits are recovered as such in the F_2 generation though one of these is not seen at the F_1 stage. During gamete or spore formation, factors of a pair separate or segregate from each other, so that **a gamete carries only one factor of a character**. This ensures the **purity of gametes**. Of course, a homozygous parent produces all gametes that are similar while a heterozygous will produce two type of gametes each having one factor with equal proportion. This law is universally applicable.

Exceptions to Mendelian Principles :

(1) **Incomplete Dominance:** After Mendelism, a few cases were observed where F_1 phenotype is **intermediate** between dominant and recessive phenotype, it means **F_1 did not resemble either of the two parents** and was in between the two.

A good example of incomplete dominance is that of flower colour in Snapdragon (dog flower or *Antirrhinum majus*). True-breeding red-flowered plant **(RR)** was crossed with true breeding white-flowered plant **(rr)**. F_1 offspring **(Rr)** had pink flowers. Here one allele is incompletely dominant over other so that intermediate phenotype is produced by F_1 hybrid with respect to the parents. If the F_1 is selfed, the plants of F_2 generation are of three types red **(RR)**, pink **(Rr)** and white flowered **(rr)** in the ratio of 1 : 2 : 1. In heterozygous condition **(Rr)**, phenotypic effect of one allele is more pronounced than that of other and then mixing of both colours (red & white) results in the development of pink colour.



Phenotypic ratio : red : pink : white
1 : 2 : 1

Genotypic ratio : RR : Rr : rr
1 : 2 : 1

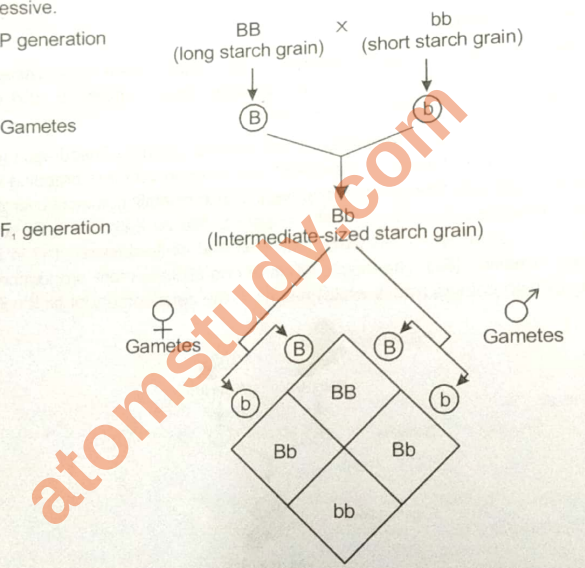
Fig. : Results of monohybrid cross in the plant Snapdragon

The Mendelian concept of a gene controlling a single character has also expanded to take into account genes which affect several characters simultaneously **(pleiotropy)**. It means in pleiotropy, **a single gene product may produce more than one effect** or control several phenotypes depending on its position. The basis of pleiotropy is the interrelationship between the metabolic pathways that may contribute towards different phenotypes. Examples :

(a) In phenylketonuria, mutation of a gene that codes for the enzyme phenylalanine hydroxylase, results in a phenotypic expression characterised by mental retardation and a reduction in hair and skin pigmentation.

(b) In *Drosophila*, white eye mutation leads to depigmentation in many other parts of the body, giving a pleiotropic effect.

(c) The gene controlling starch synthesis in garden pea. It has two alleles, **B** and **b**. Starch synthesis in **BB** homozygotes is efficient and therefore large starch grains are produced. In **bb** homozygotes, starch synthesis is less efficient, so that it produces small-sized starch grains. After maturation of seeds, **BB** seeds are round and **bb** seeds are wrinkled. Heterozygotes **Bb** form round seeds but the starch grains are of intermediate size. Now it is clear that if starch synthesis is considered, **Bb** seeds show incomplete dominance but if seed shape is considered, **B** allele is dominant and **b** is recessive.



Phenotypic ratio : Long : Intermediate : Short
1 : 2 : 1

Genotypic ratio : BB : Bb : bb
1 : 2 : 1

Fig. : Results of monohybrid cross in garden pea for size of starch grain

Therefore, dominance is not an autonomous feature of a gene or the product. It depends upon the gene product and particular phenotype we choose to examine when a gene produces more than one phenotype.

Explanation of the Concept of Dominance : Every gene contains information to express a particular trait. Diploid organisms have two copies of each gene, they are called alleles. These two alleles may be identical or non-identical. One of them may be different due to mutation (sudden change in genotype) that it has undergone which modifies the information that particular allele contains. Suppose that the normal allele produces the normal enzyme that is needed for the transformation of a substrate S. Theoretically, the modified or mutated allele could be responsible for production of

(i) The normal/less efficient enzyme, or

(ii) A non-functional enzyme, or

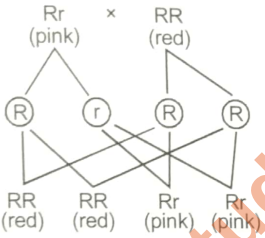
(iii) No enzyme at all

In case (i), the modified allele is equivalent to the unmodified allele, *i.e.*, it will produce the same phenotype/trait as in case of silent mutation. But, if the allele produces a non-functional enzyme or no enzyme [case (ii) & (iii)], the phenotype may be effected. The **unmodified (functioning) allele**, which represents the original phenotype is the **dominant allele/wild type** and the **modified allele** is **generally** the **recessive allele/mutant type**. Hence, the recessive trait is due to non-functional enzyme or because no enzyme is produced. Let us take the example of tallness. Plant height depends on the amount of particular plant hormone. The amount of the plant hormone made will depend on the efficiency of the process for making it. Consider now an enzyme that is important for this process. If this enzyme works efficiently, a lot of hormone will be made, and the plant will be tall. If the gene for that enzyme has an alteration that makes the enzyme non-functional or no enzyme at all, the amount of hormone will be less and the plant will be dwarf.

Example 3 : *When a cross is made between pink flowered and red flowered snapdragon plants, what proportion of phenotype in the offspring could be expected to be*

(i) Red *(ii) White*

Solution : We know that flower colour in snapdragon is an example of incomplete dominance.



(i) Red (RR) = $\frac{2}{4}$ = 50%

(ii) White (rr) = 0%

Try Yourself

7. When a cross is made between white and pink flowered *Antirrhinum* plants, what phenotypic ratio is obtained in the resulting generation?
8. State True or False:
   (i) A gamete carries only one factor of a character.
   (ii) Starch synthesis in wrinkled seeded pea plants is most efficient.
   (iii) Modified allele is always the recessive allele.

(2) **Multiple allelism:** Mendel proposed that each gene has two contrasting forms *i.e.*, alleles. But there are some genes which are having more than two alternative forms (allele). **Presence of more than two alleles for a gene is known as multiple allelism.**

A good example is different types of red blood cells that determine ABO blood grouping in human beings. ABO blood groups are controlled by the gene **I**. The plasma membrane of the red blood cells has sugar polymers that protrude from its surface and the kind of sugar is controlled by the gene. The gene **(I)** has **three alleles I^A, I^B and i/I^O** . Despite the presence of three alleles of the same gene in a population, an individual (2n) can have only two alleles. Therefore, multiple alleles can be detected **only in a population**. Since there are three different alleles, therefore six different genotypes are possible for this character (I^AI^A, I^AI^O; I^BI^B, I^BI^O; I^AI^B; I^OI^O or ii). Now to know, how many phenotype are possible, we have to see the detailed behaviour of alleles. Thus, six genotypes and four phenotypes are possible.

(3) **Co-dominance:** Besides incomplete dominance, certain alleles show co-dominance. Here in F_1 hybrid, both alleles express themselves equally and there is no mixing of the effect of the both alleles, therefore **hybrid progeny (F_1) resembles both parents**. The alleles which do not show dominance-recessive relationship and are able to express themselves independently when present together are called co-dominant alleles. The symbols used for co-dominant genes are different. One method is to show by their own capital alphabets, *e.g.*, **R** (for red hair in cattle) and **W** (for white hair in cattle). In another method, capital base symbols are employed for both the alleles with different superscripts, *e.g.*, I^A, I^B.

ABO blood group is also a good example of co-dominance. For ABO system of blood groups, allele I^A produces N-acetylgalactosamyl transferase enzyme which recognises **H** antigen present in RBC membrane and adds N-acetylgalactosamine to sugar part of **H** antigen to form **A** antigen. The allele I^B produces galactosyl transferase enzyme which adds galactose to sugar part of **H** antigen to form **B** antigen. The alleles I^A and I^B produce a slightly different form of the sugar while allele **i** does not produce any sugar or antigen. I^A and I^B are completely dominant over **i**, in other words when I^A and **i** are present only I^A expresses as **i** does not produce any sugar, and when I^B and **i** are present, only I^B expresses. When both I^A and I^B are present in a person, both enzymes or sugars thus both antigens A and B are produced. This is because of co-dominance. These antigens determines the type of blood group. Blood group A have antigen A, group B have antigen B, AB have both antigens while blood group O do not carry any antigen. Thus, six genotypes and four phenotypes are possible.

Table : Genetic Basis of Blood Groups in Human Population

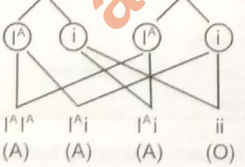
Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Antigen	Blood types of offspring
I^A	I^A	$I^A\ I^A$	A	A
I^A	I^B	$I^A\ I^B$	A, B	AB
I^A	i	I^A i	A	A
I^B	I^A	$I^A\ I^B$	A, B	AB
I^B	I^B	$I^B\ I^B$	B	B
I^B	i	I^B i	B	B
i	i	ii	Neither	O

Example 4 : *Select the set of parents that can produce child with blood group 'O'.*

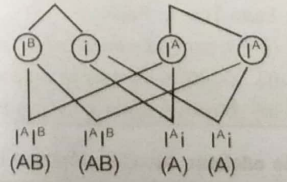
(i) $I^Ai \times I^Ai$ (ii) $I^Bi \times I^AI^A$

(iii) $ii \times I^Ai$ (iv) $I^BI^B \times I^Bi$

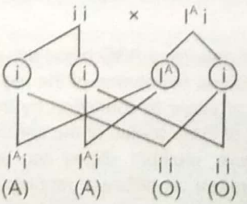
Solution : (i) $I^Ai \times I^Ai$



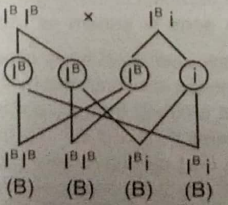
(ii) I^B i × $I^A I^A$



(iii) i i × I^A i



(iv) $I^BI^B \times I^Bi$



∴ (i) and (iii)

Try Yourself

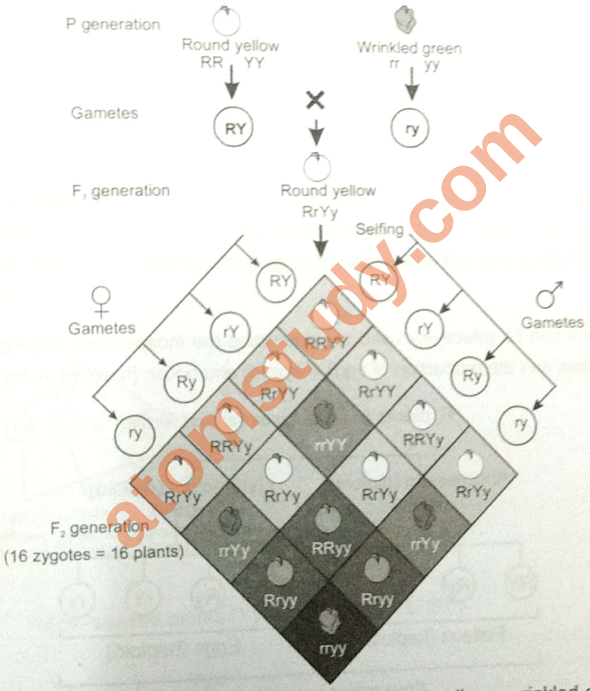
9. (i) Human beings have three alleles for ABO blood grouping: I^A, I^B and i. How many of these alleles can be present in one individual and a gamete?

(ii) A child has blood group B. If the mother has blood group AB and father blood group A, workout the genotypes of the parents and the possible genotypes of the other offsprings.

INHERITANCE OF TWO GENES

Mendel also crossed pea plants differing in two characters (dihybrid cross) to verify the results of monohybrid crosses. This helped him to understand inheritance of two pairs of factors at a time.

A cross was made between a pure round yellow-seeded pea plant **(RRYY)** with wrinkled green-seeded pea plant **(rryy)**. Yellow colour is dominant over green and round seed shape over wrinkled seed shape.



Phenotypic ratio : round yellow : round green : wrinkled yellow : wrinkled green

9/16 3/16 3/16 1/16

1 RRYY (pure round, pure yellow)	2 RRYy (pure round, hybrid yellow)	1 RRyy (pure round, green)
2 RrYY (hybrid round, pure yellow)	4 RrYy (hybrid round, hybrid yellow)	2 Rryy (hybrid round, green)
1 rrYY (wrinkled, pure yellow)	2 rrYy (wrinkled, hybrid yellow)	1 rryy (wrinkled, green)

***i.e.*, Genotypic ratio :** 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1 (9 types of genotypes)

Fig. : Results of a dihybrid cross where the two parents differed in two pairs of contrasting traits: seed colour and seed shape

Such phenotypic ratio (9 : 3 : 3 : 1) in F_2 generation was observed for several pairs of traits that Mendel studied.

Mendel found that plants of the F_1 generation have all yellow and round seeds because yellow and round traits are respectively dominant over green and wrinkled traits. These results were identical to those that he got when he made separate monohybrid crosses between yellow and green seeded plants and between round and wrinkled seeded plants.

When Mendel self hybridised the F_1 plants he found that 3/4th of F_2 plants had yellow seeds and 1/4th had green. It means, yellow and green colour segregate in a 3 : 1 ratio; just like in a monohybrid cross. Similarly 3/4th of F_2 plants had round seeded and 1/4th had wrinkled seeded condition *i.e.*, segregation of round and wrinkled shape traits in 3 : 1; just like in a monohybrid cross.

Seed colour

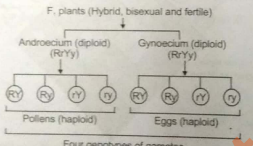
$$\left.\begin{matrix}\text{Yellow}(9+3=12)\\ \text{Green}(3+1=4)\end{matrix}\right] = 3:1$$

Seed shape

$$\left.\begin{matrix}\text{Round}(9+3=12)\\ \text{Wrinkled}(3+1=4)\end{matrix}\right] = 3:1$$

Law of Independent Assortment : Based upon the results obtained in **dihybrid crosses**, Mendel proposed a second set of generalisations that we call Mendel's Law of Independent Assortment. The law states that "when two pairs of traits are combined in a hybrid, segregation of one pair of traits is independent to the other pair of traits".

The Punnett square can be effectively used to understand the independent segregation of the two pairs of factors during meiosis and the production of eggs (haploid) and pollen (haploid) in the F_1 **(RrYy)** plant (diploid).



If we consider the segregation of one pair of factors **R** and **r**, 50% of the gametes have the factor **R** and the other 50% have **r**. Along with **R** or **r** in the gametes, it should also have the factor **Y** or **y**. Here, it is important thing to remember that segregation of 50% **R** and 50% **r** is independent from the segregation of 50% **Y** and 50% **y**. Therefore, 50% of the **r** bearing gametes has **Y** and the other 50% has **y**. Similarly 50% of the **R** bearing gametes has **Y** and the other 50% has **y**. Thus there are 4 genotypes of gametes (four types of pollen and four types of eggs). The four types are **RY**, **Ry**, **rY** and **ry each with a frequency of 25% or 1/4th of the total gametes produced**. If we write down the four types of pollens and eggs on the two sides of a Punnett square it is very easy to derive the composition of zygotes that give rise to the F_2 plants.

Now, it is clear that the segregation of one pair of factors will occur independently of the other pair or they will assort independently. Accordingly, the **gametes must carry all possible combinations of the factors in equal frequency.**

SUMMARISED ACCOUNT OF MENDEL'S EXPERIMENTS

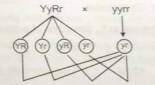
Number of traits/ hybrid (n)	Experiment	Types of gametes (2^n)	Number of zygotes/offsprings (gametes)^2	Number of Phenotype (2^n)	Number of genotype (3^n)	Phenotypic ratio	Genotypic ratio
1.	Monohybrid cross (Aa × Aa)	$2^1 = 2$	$2^2 = 4$	$2^1 = 2$	$3^1 = 3$	3 : 1	1 : 2 : 1
2.	Dihybrid cross (AaBb × AaBb)	$2^2 = 4$	$4^2 = 16$	$2^2 = 4$	$3^2 = 9$	$(3:1)^2 = 9:3:3:1$	$(1:2:1)^2 = 1:2:1:2:4:2:1:2:1$
3.	Trihybrid cross [AaBbCc × AaBbCc]	$2^3 = 8$	$8^2 = 64$	$2^3 = 8$	$3^3 = 27$	$(3:1)^3$	$(1:2:1)^3$

Example 5 : *In Pisum sativum, yellow seed colour (Y) is dominant over green (y), and round shape of seed (R) is dominant over wrinkled (y). Consider that these two pair of genes assort independently, then*

(i) What proportion of the offspring from the cross YyRr × yyrr would expected to have yellow-seeded trait?

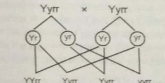
(ii) From the cross Yyrr × Yyrr, how many will be pure yellow-wrinkled plants in the resulting generation?

Solution : (i)



$\therefore$ Yellow-seeded plants (YyRr, Yyrr) = $\frac{2}{4}$ *i.e.* 50%

(ii)



$\therefore$ Pure-yellow wrinkled (YYrr) plants = $\frac{1}{4}$ *i.e.* 25%

Try Yourself

10. In a cross between two pea plants with genotypes TtYY (tall plant with yellow seeds) and Ttyy (tall plants with green seeds), what proportion of the offsprings could be expected to be:
    (i) Tall and yellow
    (ii) Dwarf and green
11. In an animal, assume that rough coat (R) is dominant over smooth coat (r) and the black (B) is dominant over white (b). Consider that these two pairs of alleles assort independently then
    (i) What proportion of the offspring from the cross RrBb × RRBB would be rough and black?
    (ii) From the cross RrBB × rrBB, how many progeny will be homozygous for both of the characters?

EXERCISE

11. The phenotype of F_1 hybrid resembles either of the two parents in
    (1) Dominance (2) Incomplete dominance
    (3) Co-dominance (4) Intermediate inheritance
12. Mendel proposed law of dominance and law of segregation based on his observations on
    (1) Monohybrid crosses (2) Dihybrid crosses
    (3) Test crosses (4) Out crosses
13. Which of the following phenotypic ratio was found by Mendel in F_2 generation of a dihybrid cross?
    (1) 3 : 1 (2) 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1
    (3) 9 : 3 : 3 : 1 (4) 12 : 4
14. Both phenotypic and genotypic ratio of F_2 are same in
    (1) Co-dominance (2) Incomplete dominance
    (3) Out cross (4) More than one option is correct
15. The ability of a gene to have multiple phenotypic effects is known as
    (1) Pleiotropy
    (2) Co-dominance
    (3) Incomplete dominance
    (4) Complete dominance
16. How many types of gametes can be produced by a diploid organism, if it is heterozygous for 3 loci?
    (1) 6 (2) 4
    (3) 8 (4) 3

17. What will be genotypic ratio in the F_2 generation of a monohybrid out cross?
    (1) 9 : 3 : 3 : 1 (2) 1 : 2 : 1
    (3) 1 : 1 (4) 3 : 1
18. A cross between F_1 hybrid and its homozygous recessive parent is called
    (1) Out cross (2) Test cross
    (3) Monohybrid cross (4) Dihybrid cross
19. Select the correct option w.r.t. law of independent assortment
    (1) It can be explained by using monohybrid cross
    (2) Inheritance of one character is dependent on another character
    (3) This law is not applicable universally
    (4) It was proposed by Bateson
20. Find the incorrect match
    (1) Gamete : Pure for a trait
    (2) Co-dominance : Flower colour in Snapdragon
    (3) Recessive gene : Expressed in homozygous
    (4) Incomplete dominance : Carl Correns

TWO GENES INTERACTION (w.r.t. Post-Mendelism)

Genes usually function or express themselves singly or individually. But, many cases are known where two genes of the same allelic pair or genes of two or more different allelic pairs influence one another. This is called gene interaction.

Non-allelic genetic interactions : These are interactions between genes located at different loci on the same chromosome or on different but non-homologous chromosomes controlling a single phenotype to produce a different expression. Each interaction is typical in itself and ratio obtained is different from those of the Mendelian dihybrid ratio. Some of these interactions of genes are explained here which fall under this category and deviate from Mendel's ratio.

1. Complementary genes

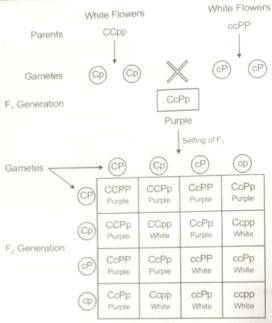
The complementary genes are two genes present on separate loci that interact together to produce dominant phenotypic character, neither of them if present alone, can express itself. It means that these genes are complementary to each other.

Bateson and Punnet have demonstrated that in sweet pea (*Lathyrus odoratus*) purple colour of flowers develop as a result of interaction of two dominant genes C and P. In the absence of dominant gene C or P or both, the flowers are white. It is believed that gene C produces an enzyme that catalyzes the formation of necessary raw material for the synthesis of pigment anthocyanin and gene P produces an enzyme which transforms the raw material into the pigment. It means the pigment anthocyanin is the product of two biochemical reactions, the end product of one reaction forms the substrate for the other.

$$\text{Substrate A} \xrightarrow{\text{Product of Gene C}} \text{Substrate B} \xrightarrow{\text{Product of Gene P}} \text{Anthocyanin}$$

Therefore, if a plant has ccPP, ccPp, CCpp or Ccpp genotypes, it bears only white flowers. Purple flowers are formed in plants having genotype CCPP or CCPp or CcPP or CcPp.

From checker board, it is clear that **9 : 7 ratio** between purple and white is a modification of 9 : 3 : 3 : 1 ratio.



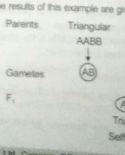
F_2 phenotypic ratio : 9 (Purple) : 7 (White)

Fig. : Results of an experiment showing inheritance of flower colour in *Lathyrus odoratus* controlled by complementary genes

2. Duplicate genes

If the dominant alleles of two gene loci produce the same phenotype, whether inherited together or separately, the 9 : 3 : 3 : 1 ratio is modified into a **15 : 1 ratio**.

Example : The capsules of shepherd's purse (Capsella) occur in two different shapes, *i.e.*, triangular and top-shaped. When a plant with triangular capsule is crossed with one having top-shaped capsule, in F_1 only triangular character appears. The F_1 offspring by self crossing produced the F_2 generation with the triangular and top-shaped capsules in the ratio of 15 : 1. Two independently segregating dominant genes (A and B) have been found to influence the shape of capsule in the same way. All genotypes having dominant alleles of both or either of these genes (A and B) would produce plants with triangular-shaped capsules. Only those with the genotype aabb would produce plants with top-shaped capsules. The results of this example are given below.



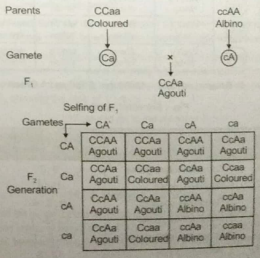
Gametes → / F_2 Generation ↓	AB	Ab	aB	ab
AB	AA BB Triangular	AA Bb Triangular	Aa BB Triangular	Aa Bb Triangular
Ab	AA Bb Triangular	AA bb Triangular	Aa Bb Triangular	Aa bb Triangular
aB	Aa BB Triangular	Aa Bb Triangular	aa BB Triangular	aa Bb Triangular
ab	Aa Bb Triangular	Aa bb Triangular	aa Bb Triangular	aa bb Top shaped

F_2 Phenotypic ratio : 15 (Triangular) : 1 (Top shaped)

Fig. : Duplicate genes interaction

3. Epistasis. A gene which masks (hides) the action of another gene (non allelic) is termed as **epistatic gene**. The process is called epistasis. The gene whose effects are masked is called **hypostatic gene**. Epistasis is of two types :

(a) **Recessive epistasis** : Here the recessive allele in homozygous condition masks the effect of dominant allele, *e.g.*, in mice, the wild body colour is known as **agouti** (greyish) and is controlled by a gene say A which is hypostatic to recessive allele c. The dominant allele C in the presence of 'a' gives coloured mice. In the presence of dominant allele C, A gives rise to agouti. So, CCaa will be **coloured** and ccAA will be **albino**. When coloured mice (CCaa) are crossed with albino (ccAA), agouti mice (CcAa) appear in F_1. cc masks the effect of AA and is therefore epistatic. Consequently, cc AA is albino. The ratio 9 : 3 : 3 : 1 is modified to **9 : 3 : 4**. The combination ccaa is also albino due to the absence of both the dominant alleles.



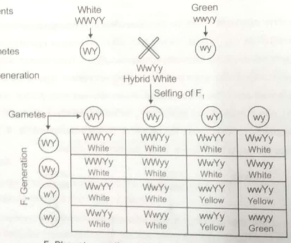
F_2 Phenotypic ratio : 9 (Agouti) : 3 (Coloured) : 4 (Albino)

Fig. : Recessive epistasis

(b) **Dominant epistasis :** In summer squash or *Cucurbita pepo*, there are three types of fruit colour – yellow, green and white. White colour is dominant over other colours, while yellow is dominant over green. Gene for white colour (W) masks the effects of yellow colour gene (Y). So, yellow colour is formed only when the dominant epistatic gene is represented by its recessive allele (w). When the hypostatic gene is also recessive (y), the colour of the fruit is green.

White Fruit – W – Y –, W – yy
Yellow Fruit – wwY–
Green Fruit – wwyy

A cross between a pure breeding white summer squash, (WWYY) with a pure breeding green summer squash, (wwyy) yields white fruits in the F_1 generation. Upon selfing of F_1, the F_2 generation comes to have **12 white fruit : 3 yellow fruit : 1 green fruit**.



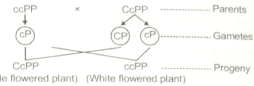
F_2 Phenotype ratio – 12 White : 3 Yellow : 1 Green or 12 : 3 : 1

Fig. : Dominant epistasis

S.No.	Types of non-allelic genetic interactions	Dihybrid phenotypic ratios in F_2 generation
1.	Complementary genes	9 : 7
2.	Duplicate genes	15 : 1
3.	Recessive epistasis	9 : 3 : 4
4.	Dominant epistasis	12 : 3 : 1
5.	Polymeric/Additive genes	9 : 6 : 1
6.	Inhibitory genes	13 : 3
7.	Supplementary genes	9 : 3 : 4
8.	Collaborative gene action	9 : 3 : 3 : 1

Example 6 : *What will be the possible phenotypic ratio if a white flowered sweet pea plant (ccPP) is crossed to a purple flowered sweet pea plant (CcPP)?*

Solution :



i.e., the phenotypic ratio = 1 : 1

Try Yourself

12. Find out the possible phenotypic ratio if a purple flowered sweet pea plant (CCPp) is crossed to a white flowered sweet pea plant (ccPp).

Polygenic inheritance or quantitative inheritance

Mendel's studies mainly described those traits that have distinct alternate forms such as flower colour which are either purple or white. But if you look around you will find that there are many traits which are not so distinct in their occurrence and are spread across a gradient. For example, in humans we do not just have tall or short people as two distinct alternatives but a whole range of possible heights. Such traits are generally controlled by two or more genes and are thus called as polygenic traits. The inheritance of polygenic traits is called polygenic or quantitative inheritance.

In quantitative inheritance, the dominant alleles have cumulative effect, with each dominant allele expressing a part of functional polypeptide and full trait is shown when all the dominant alleles are present. Genes involved in quantitative inheritance are called **polygenes**.

H. Nilsson-Ehle (1908) and East (1910) demonstrated segregation and assortment of genes controlling quantitative traits, *e.g.*, **Kernel colour in wheat and corolla length in tobacco**.

Kernel colour in wheat. Swedish geneticist, H. Nilsson-Ehle (1908) crossed red kerneled variety with white kerneled variety of wheat. Grains of F_1 were uniformly red but intermediate between the red and white of parental generation. When members of F_1 were self-crossed among themselves, five different phenotypic classes appeared in F_2 showing the ratio of 1 : 4 : 6 : 4 : 1.

(i) The extreme red – 1/16 (as red as to the parent of F_1)
(ii) Deep red (Dark red) – 4/16
(iii) Intermediate red – 6/16 (similar to F_1)
(iv) Light red – 4/16
(v) White – 1/16 (as white as to the parent of F_1)

Nilsson Ehle found that the kernel colour in wheat is determined by two pairs of genes AA and BB. Gene A and B determine the red colour of kernel and are dominant over their recessive alleles. Each gene pair shows Mendelian segregation. Heterozygotes for two pairs of genes (AaBb) segregate **into 15 red and one white kerneled plants**.

But all the red kernels do not exhibit the same shade of redness. The degree of redness was found to correspond with the number of dominant alleles.

Skin Colour in Man

The presence of melanin pigment in the skin determines the skin colour. The amount of melanin developing in the individual is determined by three (two also) pairs of genes. These genes are present at three different loci and each dominant gene is responsible for the synthesis of fixed amount of melanin. The effect of all the genes is additive and the amount of melanin produced is always proportional to the number of dominant genes.

Subsequent studies after **Davenport** have shown that as many as six genes may be involved in controlling the skin colour in human beings. As shown in table, the effect of all the genes is additive (The character is assumed to be fixed by three pairs of polygenes).

White
aabbcc
(Very light)
↓
abc

×

Negro / Black
AABBCC
(Very dark)
↓
ABC

Parents

Gametes

AaBbCc
Intermediate (Mulatto)
↓
Marriage between mulattoes

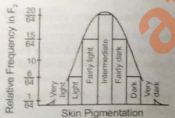
F_1 generation

Gametes → / ↓	ABC	aBC	AbC	abC	ABc	Abc	aBc	abc
ABC	AABBCC Very dark	AaBBCC Dark	AABbCC Dark	AaBbCC Fairly dark	AABBCc Dark	AABbCc Fairly dark	AaBBCc Fairly dark	AaBbCc Intermediate
aBC	AaBBCC Dark	aaBBCC Fairly dark	AaBbCC Fairly dark	aaBbCC Intermediate	AaBBCc Fairly dark	AaBbCc Intermediate	aaBBCc Intermediate	aaBbCc Fairly light
AbC	AABbCC Dark	AaBbCC Fairly dark	AAbbCC Fairly dark	AabbCC Intermediate	AABbCc Fairly dark	AAbbCc Intermediate	AaBbCc Intermediate	AabbCc Fairly light
abC	AaBbCC Fairly dark	aaBbCC Intermediate	AabbCC Intermediate	aabbCC Fairly light	AaBbCc Intermediate	AabbCc Fairly light	aaBbCc Fairly light	aabbCc Light
ABc	AaBBCc Dark	AaBBCc Fairly dark	AaBbCc Fairly dark	AaBbCc Intermediate	AABbcc Fairly dark	AABbcc Intermediate	AaBBcc Intermediate	AaBbcc Fairly light
Abc	AABbCc Fairly dark	AaBbCc Intermediate	AAbbCc Intermediate	AabbCc Fairly light	AABbcc Intermediate	AAbbcc Fairly light	AaBbcc Fairly light	Aabbcc Light
aBc	AaBBCc Fairly dark	aaBBCc Intermediate	AaBbCc Intermediate	aaBbCc Fairly light	AaBBcc Intermediate	AaBbcc Fairly light	aaBBcc Fairly light	aaBbcc Light
abc	AaBbCc Intermediate	aaBbCc Fairly light	AabbCc Fairly light	aabbCc Light	AaBbcc Fairly light	Aabbcc Light	aaBbcc Light	aabbcc Very light

Phenotypes : 1 (Very dark) : 6 (Dark) : 15 (Fairly dark) : 20 (Intermediate) : 15 (Fairly light) : 6 (Light) : 1 (Very light)

Fig. : Results of polygenic inheritance of skin colour in man

The F_1 progeny between very light and a negro individual called mulatto produces intermediate skin colour. In F_2 generation, the coloured offsprings exhibit different shades in the ratio 1 : 6 : 15 : 20 : 15 : 6 : 1.



The frequency distribution for skin colour can be represented either as a histogram or in the form of **a bell-shaped normal distribution curve**. Looking at the histogram, it can be concluded that in polygenic inheritance, the extreme phenotypes are rare and the **intermediate ones are more frequent**.

Some other example of quantitative traits are cob length in maize, human intelligence, milk and meat production, height in human and size, shape and number of seeds and fruits in plants.

(a) Number of phenotype for polygenes = 2n + 1

(b) Number of genotype for polygenes = 3^n, where n represents pair of polygenes.

Example 7 : *Calculate the sum total of phenotypes and genotypes in F_2 generation if a character is controlled by 2 pair of polygenes.*

Solution : Number of phenotype for polygenes = 2n + 1

$\therefore \quad (2 \times 2 + 1) = 5$

Number of genotype for polygenes = 3^n

$\therefore \quad 3^2 = 9$

i.e., 5 + 9 = 14

Try Yourself

13. Find out the number of phenotypes in F_2 generation if a character is controlled by 3 pair of polygenes.

EXERCISE

21. Select the odd one out w.r.t. non-allelic gene interactions
(1) Epistasis (2) Duplicate genes
(3) Incomplete dominance (4) Complementary genes

22. Fruit colour in *Cucurbita pepo* is an example of
(1) Complementary genes (2) Duplicate genes
(3) Dominant epistasis (4) Polymeric genes

23. Complementary genes were demonstrated by Bateson and Punnet in
(1) *Capsella* (2) *Lathyrus odoratus*
(3) *Antirrhinum* (4) *Mirabilis*

24. If dominant alleles of two gene loci produce the same phenotype whether inherit separately or together, it will be
(1) Recessive epistasis (2) Dominant epistasis
(3) Duplicate genes interaction (4) Inhibitory genes interaction

25. A gene which hides the action of another gene is termed as
(1) Co-dominant gene (2) Epistatic gene
(3) Hypostatic gene (4) Lethal gene

26. In polymeric gene action, the modified dihybrid phenotypic ratio in F_2 generation is
(1) 9 : 3 : 3 : 1 (2) 13 : 3
(3) 9 : 6 : 1 (4) 12 : 3 : 4

27. Which of the following genotype of sweet pea plant is related with the production of purple coloured flowers?
(1) CcPp (2) CCpp
(3) ccPP (4) Ccpp

28. Select the odd one out w.r.t. polygenic inheritance
(1) Bell-shaped curve is obtained (2) Also called quantitative inheritance
(3) Recessive alleles show cumulative effect (4) Intermediate phenotypes are more frequent

29. Select the correct match (w.r.t. dihybrid phenotypic ratio in F_2 generation)
(1) Recessive epistasis : 12 : 3 : 1
(2) Dominant epistasis : 9 : 3 : 4
(3) Collaborative gene : 9 : 3 : 3 : 1
(4) Duplicate genes : 9 : 7

30. Skin colour in man is controlled by
(1) Three pairs of polygenes (2) Duplicate genes
(3) Six pairs of polygenes (4) Supplementary genes

Chromosomal Theory of Inheritance

Mendel started his work on pea in 1856 and published it in 1865. His work did not receive any recognition, it deserved, till 1900. Mendel work remained unnoticed and unappreciated for several years due to following reasons:

(a) Communication was not easy in those days and his work could not be widely publicised. Limited circulation of the "Proceedings of Brunn Natural Science Society" in which it was published.

(b) His concept of stable, unblending, discrete units or factors for various traits did not find acceptance from the contemporaries like Charles Darwin and A.R. Wallace as an explanation for the apparently continuous variation seen in nature.

(c) His approach of using mathematical and statistical analysis to explain biological phenomena was totally new and unacceptable to many of the biologists of that time.

(d) He could not provide any physical proof for the existence of factors or the material they were made of.

(e) Non-discovery of chromosomes, mitosis and meiosis at the time of Mendel's work.

Mendel died in 1884 long before his work came to be recognised. In 1900, three scientists independently rediscovered the principles of heredity already worked out by Mendel. They were de-Vries of Holland, Carl Correns of Germany and Von Tschermak of Austria.

Did You Know?

1. de Vries found out the paper of Mendel and got it published in Flora in 1901.
2. Bateson, Punnett and other subsequent workers found Mendel's work to be universal application including animals.

Also, by this time due to advancements in microscopy that were taking place, scientists were able to carefully observe cell division. This led to the discovery of structures in the nucleus that appeared to double and divide just before each cell division. These were named chromosomes. By 1902, the chromosome movement during meiosis had been worked out. It was found that, there is a striking relationship between Mendelian factors and the chromosomes.

You have studied chapter - The cell cycle: cell division in class XI and know that

(i) DNA synthesis or replication takes place in S-phase of interphase.
(ii) Number of chromosomes remains same during the interphase.
(iii) In meiosis, each pair of synapsed homologous chromosomes is called a bivalent.

(iv) The bivalent chromosomes align on the equatorial plate in metaphase I.
(v) In anaphase-I, the homologous chromosomes separate while sister chromatids remain associated at their centromeres.
(vi) Anaphase-II involves the splitting of centromere of each chromosome and movement of chromatids towards opposite poles of the cell.
(vii) Meiosis results in 4 haploid daughter cells from one diploid parent cell.

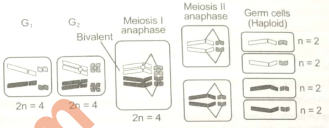


Fig. : Meiosis and germ cell formation in a cell with four chromosomes

During meiosis-I, the two chromosome pairs can align at the metaphase plate independently of each other. To understand this, we consider a heterozygous **(RrYy)** diploid cell from a plant with round-yellow seeds.

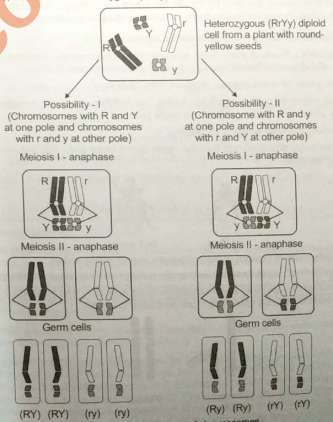


Fig. : Independent assortment of chromosomes

Chromosomal theory of inheritance was proposed independently by **Sutton and Boveri**. The two workers found a close similarity between the transmission of hereditary traits and behaviour of chromosomes while passing from one generation to the next through the agency of gametes. Sutton united the knowledge of chromosomal segregation with Mendelian principles and called it the **chromosomal theory of inheritance**. **The salient features of chromosomal theory of inheritance are as follows :**

(i) Like the hereditary traits the chromosomes retain their number, structure and individuality throughout the life of an organism and from generation to generation. The two neither get lost nor mixed up. They behave as units.

(ii) Both chromosomes as well as genes occur in pairs in the somatic or diploid cells. The two alleles of a gene pair are located on homologous sites on homologous chromosomes. Both chromosomes as well as genes segregate at the time of gamete formation such that only one of each pair is transmitted to a gamete.

(iii) A gamete contains only one chromosome of a type and only one of the two alleles of a trait.

(iv) The paired condition of both chromosomes as well as Mendelian factors is restored during fertilization.

Thus, homologous chromosomes synapse during meiosis and then separate or segregate independently into different cells which establishes the quantitative basis for segregation and independent assortment of hereditary factors.

Experimental verification of the chromosomal theory of inheritance by **Thomas Hunt Morgan** and his colleagues, led to discovering the basis for the variations that sexual reproduction produced. Morgan worked with the tiny fruit flies, *Drosophila melanogaster*, which were found very suitable for such studies.

***Drosophila melanogaster* as material for experimental Genetics**

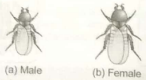
Fruit fly *Drosophila* is a tiny fly of about 2 mm size which is found over ripe fruits like mango and banana. *Drosophila* is suitable as experimental material because of following reasons :

(i) It could be grown on simple synthetic medium in the laboratory.

(ii) The fly has a short life cycle of about two weeks. The fruit fly can be bred throughout the year so that numerous generations can be obtained in a single year.

(iii) A single mating produces hundreds of offspring.

(iv) Females are easily distinguishable from the males by the larger body size and presence of ovipositor (egg-laying structure).

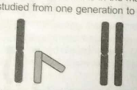


(a) Male (b) Female

(v) It has a smaller number (4 pairs) of morphologically distinct chromosomes.

(vi) It has many types of hereditary variations that can be seen with low power microscopes.

(vii) It has heteromorphic (dissimilar) sex chromosomes in the male (XY). The transmission of heteromorphic chromosomes can be easily studied from one generation to another.



Male (XY) Female (XX)

Fig. : Sex chromosomes of *Drosophila*

Example 8 : *Can you tell which of these columns A or B represent the chromosome and which represent the gene?*

Column A	Column B
Occur in pairs	Occur in pairs
Segregate at the time of gamete formation such that only one of each pair is transmitted to a gamete.	Segregate at gamete formation and only one of each pair is transmitted to a gamete.
Independent pairs segregate independently of each other.	One pair segregates independently of another pair.

Solution : Column (A) = Chromosome

Column (B) = Gene

Try Yourself

14. **State True or False** (w.r.t. Chromosomal theory of inheritance)
   (i) Both chromosomes as well as genes occur in pairs in the somatic cells.
   (ii) Both chromosomes as well as genes segregate at the time of gamete formation such that complete pair is transmitted to a gamete.
   (iii) Chromosomes are the carriers of Mendel's factors.
   (iv) The paired condition of both chromosomes as well as Mendelian factors is restored during microsporogenesis.

15. **Fill in the blanks** (w.r.t. experimental material used by Morgan)
   (i) Females are easily distinguishable from the male by the _____ body size.
   (ii) It has many types of hereditary variations that can be seen with _____ power microscope.
   (iii) Male individuals have heteromorphic _____.

Linkage and Recombination

According to Mendel's law of independent assortment, the gene controlling different characters get assorted independent to each other. It is correct if the genes are present on two different chromosomes, but if these genes are present on same chromosome they may or may not show independent assortment. If crossing over takes place between these two genes then the genes get segregated and they will assort independent to each other. But if there is no crossing over between these two genes there is no segregation, hence only parental combination will be found in gametes.

Morgan carried out several dihybrid crosses in *Drosophila* to study genes that were X-linked. The crosses were similar to the dihybrid crosses carried out by Mendel in peas.

Dihybrid crosses conducted by Morgan : At first (cross A) he crossed yellow-bodied (y) and white-eyed (w) female with brown-bodied (y⁺) red-eyed (w⁺) male and got F_1 generation in the form of brown-bodied red-eyed female and yellow-bodied white-eyed male. In F_2 generation, obtained by intercrossing of F_1 hybrids, the ratio deviated significantly from expected. He found 98.7% to be parental and 1.3% as recombinants. In a second cross (B) between white-eyed and miniature-winged female (wwmm) with wild red-eyed (w⁺) normal-winged male (m⁺) the F_1 generation included red-eyed normal-winged female and white-eyed miniature-winged male. After intercrossing the F_1, progeny was found to be 62.8% parental and 37.2% recombinant type.

Character	Dominant trait/ Wild type	Recessive trait
Body colour	Brown body (y^+)	Yellow body (y)
Eye colour	Red eye (w^+)	White eye (w)
Wings	Normal (m^+)	Miniature (m)

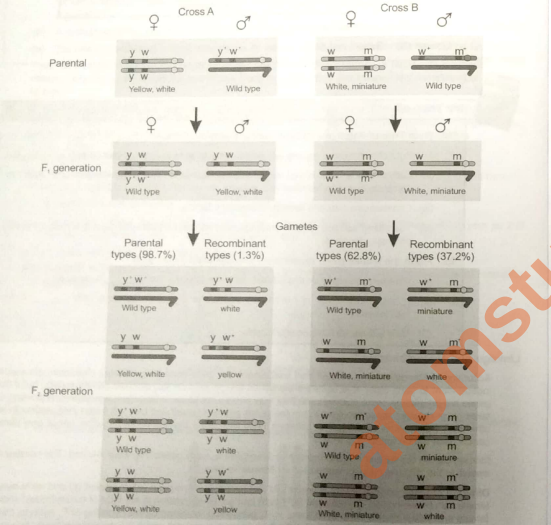


Fig. : Results of two dihybrid crosses conducted by Morgan.
Here, dominant wild type alleles are represented by (+) sign in superscript.

In both of the crosses (A and B), he observed that the two genes did not segregate independently of each other and the F_2 ratio deviated very significantly from the 9 : 3 : 3 : 1 ratio. Phenotypic ratio as 9 : 3 : 3 : 1 in F_2 generation is obtained in dihybrid cross if both genes are showing independent assortment.

Morgan and his group knew that the genes in both crosses were located on the X-chromosome (*i.e.*, same chromosome). In both crosses, Morgan found out that proportion of parental gene combination was much higher than the non-parental gene combinations.

F_2 generation	Cross A	Cross B
Parental type	98.7%	62.8%
Recombinant type (non-parental type)	1.3%	37.2%

Morgan attributed this due to the physical association of the two genes and coined the term **linkage** to describe this **physical association of genes on same chromosome** and the term **recombination** to describe the **generation of non-parental gene combinations**.

Morgan observed that recombinant types were low (1.3%) in cross A as compared to cross B, it means genes for white eye and yellow body were very tightly linked. Genes for white eye and miniature wing were loosely linked as they showed comparatively higher recombination (37.2%). Now it is clear that when genes are grouped on same chromosome, some genes are tightly linked while some are loosely linked.

Alfred Sturtevant (student of Morgan) used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and 'mapped' their position on the chromosome. Two genes show higher frequency of crossing over if the distance between them is higher and lower frequency if the distance is small. Today genetic maps are extensively used as a starting point in the sequencing of whole genomes as was done in case of the Human Genome Sequencing Project.

Chromosomal Mapping

Crossing over is important in locating the genes on chromosome. The genes are arranged linearly on the chromosome. This sequence and the relative distances between various genes is graphically represented in terms of **recombination frequencies** or cross over values (COV). This is known as linkage map of chromosome. Distance or cross over units are called centimorgan (cM) or map unit. Term **centimorgan** is used in eukaryotic genetics and **map unit** in prokaryotic genetics.

$$\text{Recombination frequency or cross over value} = \frac{\text{Number of recombinants}}{\text{Total number of offsprings}} \times 100$$

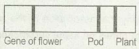
The recombination frequency depends upon the distance between the genes. If the distance between the genes is lesser the chances of crossing over is less and hence recombination frequency is also lesser and vice versa.

So, recombination frequency is directly proportional to the distance between genes. In any cross, if recombination frequency is 5%. It means the distance between the genes is 5 map unit.

A.H. Sturtevant suggested that these recombination frequencies can be utilized in predicting the sequence of genes on the chromosome. On the basis of recombination frequency, he prepared first chromosomal map or genetic map for *Drosophila*.

Knowledge Cloud

1. It was initially thought that Mendel selected seven characters and there are seven pairs of chromosome in each diploid cell in pea. Thus these genes that could have been distributed on different chromosomes, exhibited independent assortment, and their selection was a matter of luck. Detailed investigations by S. Blixt on pea led to the location of genes for Mendel's seven selected characters on four different chromosomes: two on chromosome 1, three on chromosome 4 and one each on chromosomes 5 and 7. Of the two genes on chromosome 1, they are so distantly located that no linkage is normally observed. Similarly, two out of the three on chromosome 4 are very far in relation to the third to show any linkage. This leaves only one gene pair: full vs constricted pod and tall vs dwarf plant height which ought to have shown linkage.



(Chromosome number 4 in garden pea)

If one goes through Mendel's published data, this particular combination *i.e.*, pod shape and plant height has not been mentioned at all, presumably because such a cross was never made. It thus explains as to why Mendel did not run into the problems of linkage.

2. Number of linkage group is equal to haploid set of chromosomes.

Example 9 : *Give the answer of following questions if in a test cross AaBb × aabb, 87.4% of the progeny are like parents.*

(i) Are the genes linked?

(ii) Is there any crossing over between the genes?

Solution : (i) As parental types are 87.4% (or > 50%) and non-parental types are 12.6% (or < 50%), it means genes are linked.

(ii) Yes. Crossing over leads to recombination (generation of non-parental gene combinations).

Try Yourself

16. Two heterozygous parents (AaBb) are crossed. If the two loci are completely linked (AB/ab), what would be the distribution of phenotypic features in resulting generation of test cross?
17. State True or False:
    (i) In fruit fly, genes of white eye and normal wing are X-linked recessive.
    (ii) Loosely linked genes show high recombination.

EXERCISE

31. Select the odd one out w.r.t. chromosomal theory of inheritance
    (1) It was proposed by Sutton and Boveri
    (2) Behaviour of chromosomes is parallel to behaviour of genes
    (3) Chromosomes and genes occur in pairs in diploid and haploid cells respectively
    (4) The paired condition of both chromosomes as well as Mendelian factors is restored during fertilization
32. The term gene for Mendelian factor was coined by
    (1) Sutton & Boveri (2) Morgan
    (3) Bateson (4) Johannsen
33. Morgan used *Drosophila* as experimental material because
    (1) It cannot be reared and bred under lab conditions
    (2) A single mating produces very few offsprings
    (3) It has high number of morphologically similar chromosomes
    (4) It has a short life span
34. Who carried out several dihybrid crosses in *Drosophila* to study genes that were sex-linked?
    (1) Morgan (2) Sutton
    (3) Bateson (4) Punnet
35. Female *Drosophila* is
    (1) Smaller in size than male
    (2) Larger in size than male
    (3) Larger in size with shorter life span than male
    (4) Having heteromorphic sex chromosomes
36. Find the odd one out w.r.t. complete linkage
    (1) 100% parental combinations in F_2 generation
    (2) F_2 phenotypic ratio is 3 : 1 in dihybrid cross
    (3) Dihybrid test cross ratio is 1 : 1 in F_2 generation
    (4) Linked genes tend to separate frequently
37. A condition where an individual heterozygous for two pairs of linked genes (AaBb) possesses the two dominant genes on one homologous chromosome pair and two recessive on the other, it is said to be
    (1) Cis-arrangement (2) Trans-arrangement
    (3) Partly cis partly trans (4) More than one option is correct
38. How many linkage groups are present in human male?
    (1) 24 (2) 23
    (3) 46 (4) 22
39. What is the recombination percentage between gene y and w in *Drosophila*?
    (1) 1.3% (2) 98.7%
    (3) 62.8% (4) 37.2%
40. Find the incorrect statement w.r.t. chromosomal mapping
    (1) Crossing over is important in locating genes on chromosome
    (2) Recombination frequency depends upon the distance between the genes
    (3) Recombination frequency is inversely proportional to distance between genes
    (4) The sequences and the relative distances between various genes is graphically represented in terms of recombination frequencies

SEX DETERMINATION

Establishment of sex through differential development in an individual at an early stage of life is called sex determination. Different species use very different strategies for this purpose. Some organisms like turtles rely entirely on environmental factors such as temperature for sex determination. Sex of human beings and insects like grasshopper, firefly, *Drosophila* etc. is determined genetically. The initial clue about the genetic or chromosomal mechanism of sex determination can be traced back to some of the experiments carried out in insects.

1. **Chromosomal basis of sex determination:** The foundation of this type was laid down by Henking (1891). He traced a specific nuclear structure all through spermatogenesis in a few insects. Henking also observed that only 50% of the sperm received this structure. This structure was termed '**X body**' by him, but he could not explain its significance. Further investigations by other scientists led to the conclusion that the 'X body' was actually a chromosome, therefore it was given the name X-chromosome. Stevens (1902) discovered Y-chromosome. X and Y chromosomes named as sex chromosomes by Wilson and Stevens (1905). Chromosomal basis of sex-determination is of the following types :

(a) Male heterogamety : In this type male individual produces two different types of gametes. Thus, the sperm determines the sex of the offspring. It involves two types of sex determining mechanisms; XO type and XY type.

(i) **XO type (XX - XO type) :** It is observed in large number of insects *e.g.*, Grasshopper. Number of chromosomes are different in male and female individuals.

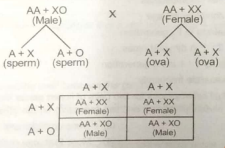


Fig.: XO type of sex determination in grasshopper

It is clear that, all eggs (ova) bear an additional X-chromosome besides the autosomes while only 50% of the sperms bear X-chromosomes. In grasshopper, eggs fertilised by (A+X) type sperm become females while those fertilised by (A+O) type sperm become males. Therefore, sperm determines the sex of the offspring. Due to the involvement of the X-chromosome in sex determination, it was designated to be the sex chromosome.

(ii) **XY type (XX - XY) type :** In a number of other insects like *Drosophila* and mammals including human beings, the males contain two types of sex chromosomes (X and Y) while females possess two similar type of sex chromosomes (XX). Both male and females have same number of chromosomes. In males, Y-chromosome is often shorter than the X-chromosome.

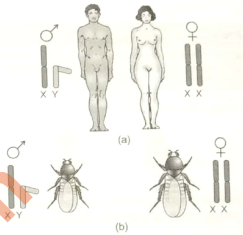


Fig.: Determination of sex by chromosomal differences (a, b); Both in humans and in *Drosophila*, the female has a pair of XX chromosomes (homogametic) and the male XY chromosomes (heterogametic) composition.

Sex Determination in Humans : Human beings have 22 pairs of autosomes and one pair of sex chromosomes. All the ova (haploid) formed by female are similar in their chromosome type (22 + X). Therefore females are homogametic. Male individual produces two types of sperms during the process of spermatogenesis. 50% of the total sperm produced possess the X-chromosome and the rest 50% has Y-chromosome besides the autosome. There is an equal probability of fertilisation of the ovum (22 + X) with the sperm carrying either X or Y chromosome. If ovum fertilises with (22 + X) type sperm, the zygote develops into a female (44 + XX) and the fertilisation of ovum with (22 + Y) type sperm results into a male individual (44 + XY). Thus, genetic makeup of the sperm determines the sex of the child. It is also clear that in each pregnancy there is always 50% or 1/2 probability of either a male or a female child.

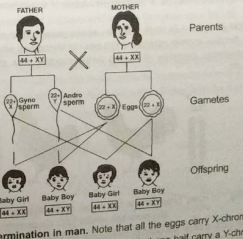


Fig.: Sex-determination in man. Note that all the eggs carry X-chromosome but one-half of the sperms carry an X-chromosome and one-half carry a Y-chromosome.

Did You Know?

In human beings, Y-chromosome carries a gene Sry (sex determining region) which codes for a product called testis-determining factor (TDF). TDF is required for the development of male sex and its absence leads to the development of female sex.

Example 10 : *State True or False (w.r.t. following diagrams)*

Male Female

(i) During gamete formation, only 50% of the sperms bear X-chromosome.
(ii) Both male and female individuals have same type of sex chromosomes.

Solution : (i) True
(ii) False

Try Yourself

18. Which of the following statement for grasshopper is **incorrect**?
(1) Male individual is heterogametic due to two heteromorphic sex chromosomes.
(2) Sperm determines the sex of offsprings.
(3) Similar number of autosomes are found in male and female individuals both.
(4) All eggs contain autosomes as well as X-chromosome.

19. In third pregnancy of a human couple, what will be probability of having a son?

(b) Female heterogamety : Female individual produces two different types of gametes. Thus, the egg determines the sex of the offspring. It involves two types of sex-determining mechanisms ZW type and ZO type.

(i) **ZW type (ZW-ZZ type) :** In birds, both the sexes possess two sex chromosomes. Unlike human beings, the females contain heteromorphic sex chromosomes while the males have homomorphic sex chromosomes. Because of having heteromorphic sex chromosomes, the females are heterogametic.

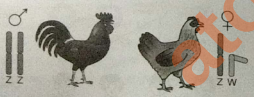


Fig. : In many birds, female has a pair of dissimilar chromosomes (ZW) and male two similar (ZZ) chromosomes.

Different symbols in birds are used to distinguish the female heterogametic in birds (ZW) from male heterogametic sex (XY) in *Drosophila* and man.

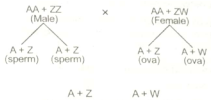


Fig. : ZW type sex determination in birds (chicks)

(ii) **ZO type (ZO-ZZ type) :** In butterflies, sex-determination is exactly opposite the condition found in grasshoppers. Here females produce two types of eggs (A+Z and A+O type).

Different systems based on chromosomal mechanism of sex-determination can be summarised as:

Type	System	Gametes		Zygotes	
		Sperms	Eggs	Males	Females
Male heterogametic	XO♂ e.g., Grasshopper, *Dioscorea*	A + X (50%) A + O (50%)	A+X (100%)	AA + XO	AA + XX
	XY♂ e.g., *Drosophila*, *Homo sapiens* (humans), *Melandrium*	A + X (50%) A + Y (50%)	A+X (100%)	AA + XY	AA + XX
Female heterogametic	ZW♀ e.g., Birds	A+Z (100%)	A + Z (50%) A + W (50%)	AA + ZZ	AA + ZW
	ZO♀ e.g., Butterflies, Moth	A+Z (100%)	A + Z (50%) A + O (50%)	AA + ZZ	AA + ZO

Sex-determination in Honey bee

The sex-determination in honey bee is based on the number of sets of chromosomes an individual receives. An offspring formed from the union of a sperm and an egg develops as a female (queen or worker), and an unfertilised egg develops as a male (drone) by means of parthenogenesis. This means that the males have half the number of chromosomes than that of a female. The females are diploid having 32 chromosomes and males are haploid, *i.e.*, having 16 chromosomes. This is called as haplodiploid sex-determination system and males are haploid, *i.e.*, having 16 chromosomes. This is called as haplodiploid sex-determination system and has special characteristic features such as the males produce sperms by mitosis shown in figure below, they do not have father and thus cannot have sons, but have a grandfather and can have grandsons.

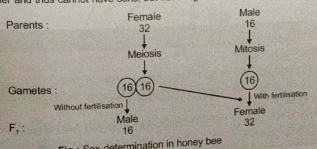


Fig.: Sex-determination in honey bee

2. **Non-Allosomic genetic sex determination :** Fertility factor of plasmid in bacteria determines sex.

3. **Genic Balance or X/A Balance Theory of Sex Determination :** Given by **C.B. Bridges**. According to him, Y chromosome plays no role in sex determination of *Drosophila* and it is the ratio between number of X-chromosome and set of autosomes which determines the sex of fly.

Chromosome Constitution	X/A ratio	Sex Index
AA + XXX	3/2 = 1.50	Super ♀
AA + XX	2/2 = 1.00	Normal ♀
AAA + XXY	2/3 = 0.67	Intersex
AA + XY	1/2 = 0.50	Normal ♂ (Fertile)
AA + XO	1/2 = 0.50	♂ (Sterile)
AAA + XY	1/3 = 0.33	Super ♂

It was concluded that **X/A ratio of > 1.0 expresses super femaleness, 1.0 femaleness, below 1.0 and above 0.5 intersexes, 0.5 maleness and < 0.5 supermaleness.**

Gynandromorphs: Gynandromorph is a **sex mosaic** (an individual with one half of the body male and the other half female). These are common in **Silk moth** and *Drosophila*. Gynandromorphism is developed due to **accidental loss of X-chromosome from a 2A + XX cell during mitosis.**

Gynander : A gynander may be male or female with patches of tissues of other sex on it.

4. **Environmental Mechanism of Sex Determination :** This mechanism is observed by **F. Baltzer** in *Bonnelia viridis* (marine worm). In this organism, the sex is undifferentiated in larva. The larva which settle down in mud, grow up into mature female while those which settle down near the proboscis of female and become parasite develop into male. It has been demonstrated that female secrete certain hormone which induces sex in larva. *Crepidula* and *Ophryotrocha* also show such mechanism.

Example 11 : *Find out the* ***Incorrect*** *match.*

(A) (B)

(1) *A - Homogametic.* (2) *B - Female chick.*
(3) *A - Sex determiner* (4) *B - Heterogametic*

Solution : (3) (B) is sex determiner as it has heteromorphic sex chromosomes.

Try Yourself

20. State True or False :
(i) In birds, both the sexes possess two sex chromosomes.
(ii) In butterflies, sex determination is exactly opposite the condition found in grasshoppers.

21. Fill in the blanks :
(i) In chicks, _____ individual produces two different types of gametes.
(ii) In butterflies, all _____ gametes contain autosomes as well as sex chromosome.

SEX LINKED INHERITANCE

Sex linkage was discovered by **Morgan**, while working on inheritance of eye colour in *Drosophila*. He made three types of crosses :

Cross - 1 : The white eyed male (w) was crossed with red eyed (w⁺) female. All the flies of F_1 generation were found to be red eyed. F_1 flies were allowed to self breed. In F_2 generation, both the traits of red eye and white eye appeared in the ratio 3 : 1 showing that white eye trait is recessive to red eye trait.

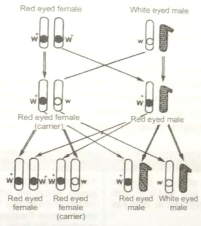


Fig. : Cross 1 of Morgan – Involving red eyed female *Drosophila* and white eyed male *Drosophila*.

F_1 generation consisted of only red eyed flies. In F_2 generation all female flies were red-eyed. 50% of the male flies were red eyed and the remaining 50% white eyed.

Cross - 2 : Red eyed females of F_1 generation were crossed with white eyed male. It is similar to test cross where hybrids are cross bred with recessive parents. Morgan obtained red and white eyed female as well as male in equal proportions– 1 red eyed female : 1 white eyed female : 1 red eyed male : 1 white eyed male. The test cross indicated that white eye colour was not restricted to the male fly.

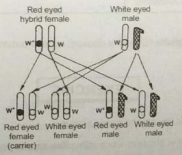


Fig. : Cross 2 of Morgan – Test cross in *Drosophila* where red and white eyed traits appear in both males and females in equal proportions

Cross - 3 : White eyed females were crossed with red eyed males. It was a reciprocal of cross 1 and should give the similar result as obtained by Mendel. However, Morgan obtained a surprising result. All the males were white eyed while all the females were red eyed.

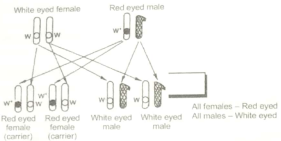


Fig. : Cross 3 of Morgan – Involving white eyed female and red eyed male showing criss-cross inheritance in *Drosophila*.

Taking all the crosses into consideration, Morgan came to the conclusion that eye colour gene is linked to sex and is present on the X-chromosome. **X-chromosome does not pass directly from one parent to the offspring of the same sex but follows a criss-cross inheritance**, *i.e.*, it is transferred from one sex to the offspring of the opposite sex. **In other words, in criss-cross inheritance a male transmits his traits to his grandson through daughter (Diagynic), while a female transmits the traits to her granddaughter through her son (Diandric).**

S.No.	Sex limited traits	Sex Influenced traits	Holandric traits
1.	The genes of these traits are **autosomal** and found in both sexes but **express in one sex only**.	These are those **autosomal genes** which are influenced by the sex of the bearer. These traits appear **more frequently in one sex** than in the other.	These are **Y-linked traits** those inherit from **male to male only**.
2.	**Examples :** (i) Milk glands in female (ii) Beard in man (iii) Deep male voice (iv) Antlers in male deer (v) Brilliant plumage in pea-cock (vi) Female or male musculature	(i) Pattern baldness (affected by male sex hormone/testosterone) (ii) Short index finger in male	(i) Porcupine skin (ii) TDF (Testes determining factor) (iii) Hypertrichosis

Sex Linkage in Human Beings

Colour blindness and **haemophilia** (Bleeder's disease) are two common examples of sex-linked diseases in human beings.

EXERCISE

41. Individuals having homomorphic sex-chromosomes produce
(1) One type of gametes (2) Two type of gametes
(3) No gametes (4) Only one gamete in complete life span

42. Holandric genes are present on
(1) X-chromosomes (2) Y-chromosomes
(3) Sex-chromosomes as well as autosomes (4) Autosomes

43. Mark the incorrect pair (w.r.t. sex determination)
(1) ZW-ZZ type – Fishes
(2) ZO-ZZ type – Birds
(3) XX-XO type – *Dioscorea*
(4) XX-XY type – *Melandrium*

44. 50% sperms are devoid of sex-chromosomes in
(1) *Melandrium* (2) Moth
(3) Grasshopper (4) Bee

45. In the XX-XO type of sex determination
(1) Females produce only one type of eggs
(2) Females have only one X-chromosome
(3) Males have two X-chromosomes
(4) Males are homogametic

46. Select the odd one out w.r.t. genic balance theory of sex-determination in *Drosophila*
(1) Y-chromosome plays no role in sex-determination
(2) Given by C.B. Bridges
(3) If X/A ratio is one, superfemales are produced
(4) If X/A ratio is less than 0.5, supermales are produced

47. Environmental mechanism of sex-determination is seen in
(1) *Bonnelia* (2) *Crepidula*
(3) Grasshopper (4) More than one option is correct

48. Select the odd one out w.r.t. haemophilia
(1) X-linked dominant disorder
(2) Bleeder's disease
(3) Criss-cross inheritance
(4) X-linked recessive disorder

49. Select the correct match
(1) Sex-limited trait – Colour blindness
(2) Sex-limited trait – Express in both sexes
(3) Sex-influenced trait – More frequent in one sex than in the other
(4) Sex-influenced trait – Porcupine skin

50. All are sex limited traits, except
(1) Beard in man (2) Porcupine skin
(3) Antlers in male deer (4) Brilliant plumage in peacock

MUTATION

Mutation is sudden, discontinuous variation in genotype and phenotype of an organism due to change in chromosomes and genes. In addition to recombination, mutation is another phenomenon that leads to variation in DNA. Depending upon the cause, mutations are of three types :

1. Gene Mutation

It is alteration of DNA due to change in nucleotide sequence. Gene mutation may occur due to change in a single base pair of DNA, known as point mutation. A classical example of point mutation is sickle cell anaemia. Change in more than one nucleotide pair is called gross mutation. Gene mutation occurs by following methods:

(a) **Frame-shift mutation**

(i) **Deletion :** Removal of one or more bases from nucleotide chain.

(ii) **Insertion or addition :** Addition of one or more bases in a nucleotide chain.

(b) **Substitution.** The replacement of one base by another. It is of two types:

(i) **Transition :** When a purine base (A or G) is substituted by another purine base or pyrimidine base (T or C) is substituted by another pyrimidine base.

(ii) **Transversion :** Substitution of a purine base with a pyrimidine base or vice versa.

Gene mutation may occur naturally and automatically due to internal reason. They are named as spontaneous mutations. However, they are produced by external physical or chemical factors. These factors are named as mutagens that are used to create induced mutations.

2. Chromosomal Aberrations

Chromosomes are made up of proteins, DNA and RNA. Each chromatid possesses one DNA helix that runs continuously from one end to the other. In chromatids, DNA is present in a highly supercoiled form. Therefore, loss (deletions) or gain (insertion/duplication) of a segment of DNA, results in alternation in chromosomes. We know that genes are located on chromosomes, so that alteration in chromosomes results in abnormalities or aberrations. These are commonly observed in cancer cells. The important aberrations are as follows :

(a) **Deletion :** Occurs when a part of the chromosome is lost. It can be divided into two types-terminal and intercalary. Terminal deletion is the loss of a terminal segment of a chromosome and is produced by a single break in the chromosome. During intercalary deletion there is the loss of an intercalary segment of a chromosome due to double break.

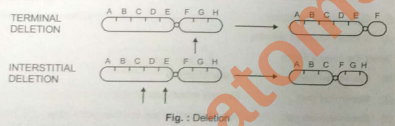


Fig. : Deletion

(b) **Duplication :** Occurs due to addition of a part of chromosome so that a gene or set of genes is represented twice.

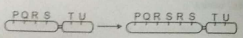


Fig. : Duplication

(c) **Translocation :** It involves shifting of a part of one chromosome to another non-homologous chromosome. So new recombinant chromosomes are formed, **as this induces faulty pairing of chromosomes during meiosis**. An important class of translocation having evolutionary significance is known as **reciprocal translocation** or **segmental interchanges**, which involves mutual exchange of chromosome segments between non-homologous chromosome, *i.e.*, **illegitimate crossing over**. Chronic myelogenous leukemia (CML) occurs due to translocation of segment of long arm from chromosome 22 to chromosome 9. **Chromosome 22 is called Philadelphia chromosome**.

(d) **Inversion :** Change in linear order of genes by rotation of a section of chromosome by 180°. Inversion occurs frequently in *Drosophila* as a result of X-ray irradiation. They may be of two types :

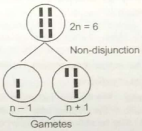
(i) Paracentric : Inversion without involving centromere (Inverted segment does not carry centromere).

(ii) Pericentric : Inversion involving centromere.

3. Genomatic Mutation :

It is change in chromosome number that bring about visible effects on the phenotype. It is of two types:

(a) **Aneuploidy :** In aneuploidy, any change in number of chromosomes in an organism would be different than the multiple of basic set of chromosomes. It commonly arises due to non-disjunction (absence of separation of two homologous chromosomes during cell division) of the two chromosomes of homologous pair so that one gamete comes to have an extra chromosome (n +1) while the other becomes deficient in one chromosome (n – 1).



Fusion of these gametes (n – 1 or n + 1) with normal gametes (n) gives rise to different types of aneuploids.

S. No.	Gametic fusion	Type of aneuploid
(i)	n × (n–1)	2n – 1 (Monosomic condition)
(ii)	(n–1) × (n–1)	2n – 2 (Nullisomic condition)
(iii)	n × (n+1)	2n + 1 (Trisomic condition)
(iv)	(n+1) × (n+1)	2n + 2 (Tetrasomic condition)

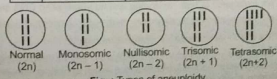


Fig. : Types of aneuploidy

Examples of aneuploidy :

(i) Trisomy: Down's syndrome, Klinefelter's syndrome

(ii) Monosomy: Turner's syndrome

(b) Euploidy : In euploidy, any change in the number of chromosomes is the multiple of the number of chromosomes in a basic set or it occurs due to variation in one or more haploid sets of chromosomes.

Accordingly, these may be haploidy and polyploidy.

(i) Haploidy : One set of chromosomes. Haploids are better for mutation experimental studies, because all mutations either dominant or recessive can express immediately in them, as there is only one allele of each gene present in each cell.

(ii) Polyploidy : More than two sets of chromosomes. Failure of cytokinesis after telophase stage of cell division results in an increase in a whole set of chromosomes in an organism and this phenomenon is called as polyploidy. It is often seen in plants. In case of animals, polyploidy usually results in sterility. Therefore polyploidy is rare in animals. Polyploidy fall into two major categories- autopolyploidy and allopolyploidy.

Mutagens

Mutations can be artificially produced by certain agents called mutagens or mutagenic agents. Following are two major types of mutagens :

(1) Physical mutagens :

Radiations are the most important physical mutagens. **H.J. Muller** who used X-rays, for the first time, to increase the rate of mutation in *Drosophila*, opened an entirely new field in inducing mutations. So, Muller is considered as **father of Actinobiology**.

The main source of spontaneous mutations are the natural radiations coming from cosmic rays of the sun.

The spectrum of wavelengths that are shorter (*i.e.*, of higher energy) than the visible light can be subdivided into following two groups :

(a) **Ionizing radiations**

(b) **Non-ionizing radiations**

They occur in small amounts in the environment and are known as background radiations.

The following are biological effects of radiations :

(a) Effects of ionizing radiations : The ionizing radiations include X-rays, γ-rays, α-rays and β-rays. Ionizing radiations cause breaks in the chromosome. These cells then show abnormal cell divisions. If these include gametes, they may be abnormal and even die prematurely. Different types of cancers may result due to radiations. The frequency of induced mutations is directly proportional to the doses of radiations.

(b) Effects of non-ionising radiations : The non-ionizing radiations have longer wavelengths but carry lower energy. This energy is insufficient to induce ionization. Therefore, non-ionizing radiations such as UV light do not penetrate beyond the human skin. **Thymine (pyrimidine) dimer** formation is a major mutagenic effect of UV rays that disturbs DNA double helix and thus, DNA replication.

(2) Chemical mutagens : Large number of chemical mutagens are now known. These are more injurious than radiations. The first chemical mutagen used was mustard gas by **C. Auerbach *et. al.*** during world war II.

Chemical mutagens are placed into two groups :

(a) Those which are mutagenic to both replicating and non-replicating DNA such as nitrous acid,

(b) Those which are mutagenic only to replicating DNA, such as acridine dyes and base analogues.

Following are the effects of some of the chemical mutagens :

(i) **Nitrous acid :** It is mutagenic to both replicating and non-replicating DNA. It acts directly by oxidative deamination on A, G and C bases which contain amino groups. A is deaminated to **hypoxanthine** which is complementary to cytosine. G is converted to **xanthine** which pairs with C. Cytosine is converted to U which pairs with A.

(ii) **Acridines :** Acridines and proflavins are very powerful mutagens. These can intercalate between DNA bases and interfere with DNA replication, producing insertion or deletion or both of single bases respectively. This induces frame shift-mutations or gibberish mutation, *e.g.*, **Thalassaemia**.

(iii) **Base analogues :** These have structures similar to the normal bases and are incorporated into DNA only during DNA replication. Base analogues cause mispairing and eventually give rise to mutations. The base analogues may either be natural or artificial. Natural base analogues include 5-methyl cytosine, 5-hydroxymethyl cytosine, 6-methyl purine etc.

The most commonly used artificial base analogues are 5-bromouracil and 2-aminopurine. 5-bromouracil is a structural analogue of thymine. It gets incorporated into the newly replicated DNA in place of thymine (T). 2-aminopurine is an artificial base analogue of adenine. It acts as a substitute of adenine (A) and can also pair with cytosine (C).

Example 12 : *Give one word for the following :*

(i) Phenomenon which results in alteration of DNA sequences and consequently results in changes in the genotype and the phenotype of an organism.

(ii) Type of mutation that arise due to change in a single base pair of DNA.

Solution : (i) Mutation

(ii) Point mutation

Knowledge Cloud

Types of Mutations : Different classifications of mutations are known, each based on a definite criterion or character.

I. Spontaneous and induced mutations : These are based upon the agency involved.

Spontaneous mutations. These are natural mutations. They have also been called background mutations. Such mutations occur at a frequency of 1×10^{-5} in nature. **Induced mutations.** These have been observed in organisms due to specific factors such as radiations, ultra violet light or variety of chemicals. The agents which induce mutations on their application, are called **mutagens** or mutagenic agents.

II. On the basis of the type of cells in which mutations occur, there are other two types of mutations.

(a) Somatic mutations. These mutations occur in somatic cells, *i.e.*, body cells or the cells other than germinal cells. The somatic mutations do not have any genetic or evolutionary importance. This is because only the derivatives or the daughter cells formed from the mutated cell will show mutation and not the whole organism.

(b) Germinal mutations. These mutations occur in the gametes or germ cells and are also known as gametic mutations. Such mutations are heritable, and, therefore, are of great evolutionary significance. If the mutations are dominant, these are expressed in the next generation and if recessive, their phenotypic expressions remain suppressed.

III. Forward and backward mutations : The commonest type of mutation, is the change from normal or wild type to new genotype (recessive or dominant). Such mutations are called **forward mutations.**

An organism which has undergone forward mutation, may again develop mutation which restores the original wild-type phenotype. Such reversions are known as backward mutations or **reverse mutations.**

Try Yourself

22. State True of False :
    (i) Chromosomal aberrations are commonly observed in cancer cells.
    (ii) Mutation is the only phenomenon that leads to variation in DNA.
23. Fill in the blanks :
    (i) Deletions and insertions of base pair of DNA, causes_____.
    (ii) A classical example of point mutation is _____.

GENETIC DISORDERS

1. Pedigree Analysis :

(Method of study of human genetic disorders)

Human beings, like other living organisms, also follow the principles of inheritance but common Mendelian experiments cannot be carried out over us due to following reasons.

(i) Controlled crosses are not possible in human beings.

(ii) Number of offspring per couple is small.

Because of the reasons described above, human geneticist has to resort to slightly different methods of genetic analysis. Such an analysis of traits in a several of generations of a family is called the pedigree analysis. In the pedigree analysis the inheritance of a particular trait is represented in the family tree over generations. It is useful for the genetic counsellors to advice intending couples about the possibility of having children with genetic defects like haemophilia, colour blindness, phenylketonuria, thalassemia, sickle cell anaemia (recessive traits), myotonic dystrophy and polydactyly (dominant traits).

A family tree or pedigree is drawn up using certain standard symbols. Some of the important symbols are as follows

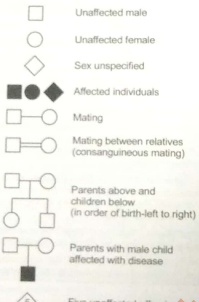
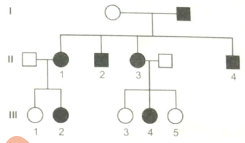


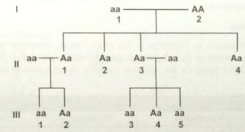
Fig. : Symbols used in the human pedigree analysis

By pedigree analysis one can easily understand whether the trait in question is autosomal dominant or recessive. Similarly, the trait may also be linked to the sex chromosome as in case of haemophilia. For carrying out simple analysis involving recessive or dominant allele, certain clues or simple rules are sought from the pedigree. For example, in the case of recessive allele, characteristic condition can appear in the progeny of apparently unaffected parents. Moreover, two affected individuals cannot have unaffected child. Let us try to understand pedigree analysis with the help of following example.

Example 13 : *In the pedigree given below, indicate whether the shaded symbols indicate dominant or recessive allele. Also give genotype of the whole pedigree.*

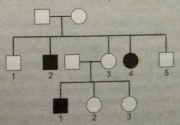


Solution : Since the shaded symbol appears in all the offsprings, father must be homozygous dominant while the mother homozygous recessive (AA × aa = all Aa) because in all other cases this possibility is absent (Aa × aa = 2Aa + 2aa; aa × AA = all Aa; aa × Aa = 2aA + 2aa). All the members of II generation will, therefore, be heterozygous (Aa). This is further confirmed by marriage of II – 1 with homozygous recessive (Aa × aa = 2 Aa, 2 aa) and bearing children of both the parental types. Marriage of II – 3 with the homozygous recessive can produce both recessive and heterozygotes as are present here.



Try Yourself

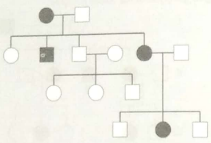
24. Study the given pedigree chart and answer the questions that follow :



(i) Trait is autosomal recessive (True/False).

(ii) Give the genotype of the members 3 and 4.

25. In the following pedigree chart, the mutant trait is shaded black. The gene responsible for the trait is



(1) Dominant and X-linked
(2) Dominant and autosomal or dominant and X-linked
(3) Recessive and X-linked
(4) Recessive and Y-linked

Types of Human Genetic Disorders :

We know that each and every feature in any organism is controlled by one or the other gene located on the DNA present in the chromosome. DNA is the carrier of genetic information. It is hence transmitted from one generation to the other without any change or alteration. However, changes do take place occasionally. A number of disorder in human beings have been found to be associated with the inheritance of changed or altered genes or chromosomes.

2. Mendelian Disorders

These are mainly determined by mutation in the single gene, therefore also called gene related human disorders. They are transmitted to the offspring as per Mendelian principles. The pattern of inheritance of such disorders can be traced in a family by the pedigree analysis. Some common and prevalent Mendelian disorders are as follows:

S. No.	Disorder	Dominant / Recessive	Autosomal / Sex linked
(1)	Haemophilia	Recessive	X-linked
(2)	Colour blindness	Recessive	X-linked
(3)	Sickle cell anaemia	Recessive	Autosomal
(4)	Phenylketonuria	Recessive	Autosomal
(5)	Cystic fibrosis	Recessive	Autosomal
(6)	Thalassemia	Recessive	Autosomal
(7)	Myotonic dystrophy	Dominant	Autosomal

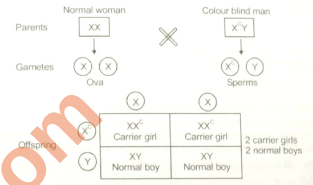
(a) Colour blindness: Colour blindness is a recessive sex-linked trait in which the eye fails to distinguish red and green colours. The gene for normal vision is dominant. The normal gene and its recessive allele are carried by X-chromosome. In female, colour blindness appears only when both the sex chromosomes carry the recessive gene (X^cX^c). The females have normal vision but function as carrier if a single recessive gene for colour blindness is present (XX^c). However, in human males the defect appears in the presence of a single recessive gene (X^cY) because Y-chromosomes of males do not carry any gene for colour vision. Colour blindness, like any other sex-linked trait, shows **criss-cross inheritance** (*i.e.*, a male transmits his trait to his grandson through daughter, while a female transmits the traits to her grand-daughter through her son or it is transfer of trait from one sex to the offspring of the opposite sex).

It should be very much clear, colour blindness does not mean not seeing any colour at all, it means that those who are colourblind have trouble in seeing the differences between certain colours.

Most colourblind people cannot tell the difference between red or green. That does not mean that they cannot do their normal work. In fact, they can also drive - they learn to respond to the way the traffic signal lights up the red light is generally on the top and green is on the bottom.

If a colourblind man (X^cY) marries a girl with normal vision (XX), the daughters would have normal vision but would be carrier, while sons would also be normal (**shown in cross(a)**).

Cross (a)



If the carrier girl (heterozygous for colour blindness, X^cX) now marries a colour blind man X^cY, the offspring would show 50% females and 50% males. Of the females, 50% would be carrier for colour blindness and the rest 50% would be colour blind. Of the males, 50% would have normal vision and the 50% would be colour blind (**shown in cross (b)**).

Cross (b)

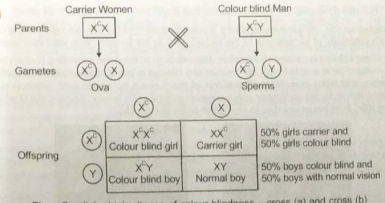


Fig. : Sex-linked inheritance of colour blindness – cross (a) and cross (b)

(b) Haemophilia : It is X-linked recessive trait therefore shows its transmission from normal carrier female (heterozygous) to male progeny. Due to presence of defective form of blood clotting factor (protein), exposed blood of affected individuals fails to coagulate.

The possibility of a female becoming a haemophilic is extremely rare because mother of such a female has to be at least carrier and the father should be haemophilic (unviable in the later stage of life). Haemophilic female dies before birth. The family pedigree of Queen Victoria shows a number of haemophilic descendents as she was a carrier of the disease.

The person suffering from this disease cannot synthesize a normal blood protein called antihaemophilic globulin (AHG) required for normal blood clotting (**Haemophilia A** - more severe). Therefore, even a very small cut may lead to continuous bleeding for a long time. This gene is located on X chromosome and is recessive. **It remains latent in carrier females.**

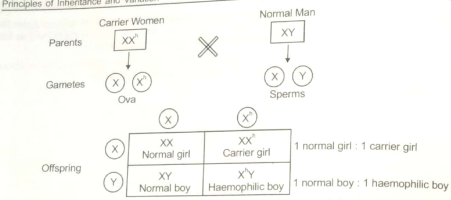


Fig.: Inheritance of haemophilia when the mother is carrier and the father is normal

If a normal man marries a girl who is carrier for haemophilia, the progeny would consist of 50% females and 50% males. Of the females, 50% would be normal and the rest 50% would be hemophilia carrier. Of the males, 50% would be normal and the rest would be haemophiliacs.

Haemophilia - B (Christmas disease) – plasma thromboplastin is absent, Inheritance is just like Haemophilia A.

(c) **Sickle-cell anaemia :** As it is autosomal recessive disease therefore it can be transmitted from parents to the offspring when both male and female individuals are carrier (heterozygous) for the gene. The disease is controlled by a single pair of allele, Hb^A and Hb^S. Thus three genotypes are possible in population.

(i) $Hb^A Hb^A$ (Normal, homozygous)

(ii) $Hb^A Hb^S$ (Normal, carrier)

(iii) $Hb^S Hb^S$ (Diseased, die before attaining maturity)

Heterozygous ($Hb^A Hb^S$) individuals appear apparently unaffected but they are carrier of the disease as there is 50% probability of transmission of the mutant gene to the progeny, thus exhibiting sickle-cell trait.

The disease/defect is caused by mutation (transversion) of the gene controlling β-chain of haemoglobin. The mutated gene is called Hb^S. Hb^S causes one change in amino acid sequence of β-chain. It replaces glutamic acid (Glu) present at 6th position of the β-chain by amino acid valine (Val). The mutant haemoglobin molecule undergoes polymerisation under low O_2 tension causing the change in the shape of the RBC from biconcave disc to elongated sickle-like structure.

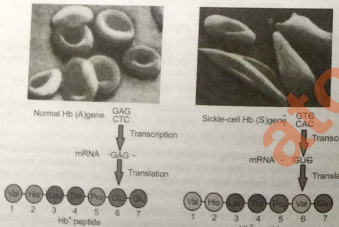


Fig. : Micrograph of the red blood cells and the amino acid composition of the relevant portion of β-chain of haemoglobin: (a) From a normal individual; (b) From an individual with sickle-cell anaemia

(d) **Phenylketonuria :** This inborn error of metabolism is also inherited as the autosomal recessive trait. The affected individual lacks a liver enzyme called phenylalanine hydroxylase that converts the amino acid phenylalanine into tyrosine. As a result of this phenylalanine is accumulated and converted into phenylpyruvic acid and other derivatives. Accumulation of these in brain results in mental retardation. These are also excreted through urine because of its poor absorption by kidney.

(e) **Thalassemia :** Thalassemia is a recessive autosomal genetic defect, originated in Mediterranean region – by their mutation or deletion recessive autosomal. Thalassemias are a group of disorders caused by defects in the synthesis of globin polypeptide in RBC. Absence or reduced synthesis of one of the globin chains results in an excess of the other. In this situation free globin chains, which are insoluble, accumulate inside the red cells and form precipitates which damage the cell, causing cell lysis and resulting in anaemia. There are two main types of thalassemias in which synthesis of α or β globin is defective. It is common in Mediterranean, Middle East, Indian subcontinent and in south-east Africa.

(i) **Alpha (α) thalassemia :** The α-thalassemias involve the genes HBA1 and HBA2, inherited in a Mendelian recessive fashion. There are two gene loci and so four alleles. It is also connected to the deletion of the 16p (short-arm) chromosome. α-Thalassemias result in decreased α-globin production, therefore, fewer alpha-globin chains are produced, resulting in an excess of β chains in adults and excess γ chains in newborns. The excess β chains form unstable tetramers (called Hemoglobin H or HbH of 4 beta chains) which have abnormal oxygen dissociation curves.

(ii) **Beta (β) thalassemia :** β-Thalassemias are due to mutations in the HBB gene on chromosome 11, also inherited in an autosomal-recessive fashion. The severity of the disease depends on the nature of the mutation. Mutations are characterised as ($β^0$ or β thalassemia major) if they prevent any formation of β chains (which is the most severe form of β thalassemia); they are characterised as ($β^+$ or β thalassemia intermedia) if they allow some β chain formation to occur. In either case, there is a relative excess of α chains, but these do not form tetramers; rather, they bind to the red blood cell membranes, producing membrane damage, and at high concentrations they form toxic aggregates.

(iii) **Delta (δ) thalassemia :** Just like β thalassemia, mutations can occur which affect the ability of this gene to produce δ chains. α and β chains are present in hemoglobin but about 3% of adult hemoglobin is made of alpha and delta chains. Earlier you have studied SCA where there is a synthesis of incorrectly functioning globin but here in thalassemia too few globins are synthesised.

3. Chromosomal Disorders :

Mendelian disorders like haemophilia, sickle-cell anaemia and phenylketonuria are due to the mutant allele and their defective products. However, disorders can also be created by imbalance in chromosome number and chromosomal rearrangement. These are called as chromosomal disorders. Down's syndrome, Klinefelter's syndrome and Turner's syndrome are common examples of chromosomal disorders.

(a) **Down's syndrome :** It was first described in 1866 by Langdon Down. The disorder develops due to trisomy of chromosome number 21. Trisomic condition arises due to the formation of n+1 male or female gamete by non-disjunction and the subsequent fertilisation by a normal (n) gamete. It is characterised by :

(i) Short stature

(ii) Small round head

(iii) Furrowed tongue

(iv) Partially open mouth

(v) Broad palm with characteristic palm crease

(vi) Many 'loops' on finger tips

(vii) Big and wrinkled tongue

(viii) Physical (underdeveloped gonads and genitals, loose jointedness), psychomotor and mental development is retarted.

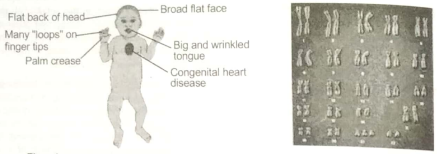


Fig. : A representative figure showing an individual inflicted with Down's syndrome

(b) Klinefelter's syndrome : It is caused due to the presence of an additional copy of X-chromosome resulting into 44 + XXY type chromosome complement. The defect appears due to union of an abnormal egg (22 + XX) and a normal sperm (22 + Y) or normal egg (22 + X) and abnormal sperm (22 + XY). Such persons are sterile males with overall masculine development and some female characteristics (*e.g.*, Feminine pitched voice, development of breast or gynaecomastia).

(c) Turner's syndrome : The disorder is due to monosomy. It appears due to fusion of abnormal egg (22 + 0) and a normal sperm (22 + X) or a normal egg (22 + X) and abnormal sperm (22 + 0). Such females are sterile as ovaries are rudimentary besides other features including lack of other secondary sexual characters.

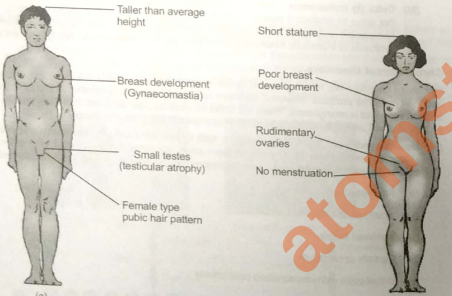
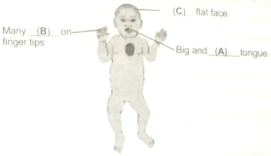


Fig. : Representative figures showing individuals inflicted with (a) Klinefelter syndrome and (b) Turner's syndrome

Example 14 : *Given below is the figure showing an individual inflicted with Down's syndrome.*



Write down the correct words for all the three blanks (A), (B) and (C) indicated in the figure.

Solution : (A) Wrinkled
(B) Loops
(C) Broad

Try Yourself

26. Read the following paragraph carefully and find out the correct words for all the three blanks indicated as (A), (B) and (C).

"The substitution of amino acid in the globin protein results due to the single base substitution at the 6th codon of the β-globin gene from __(A)__ to __(B)__. The mutant haemoglobin molecule undergoes polymerisation under __(C)__ tension causing the change in the shape of the RBC from biconcave disc to elongated sickle-like structure.

27. State True or False :

(i) Heterozygous female for haemophilia may transmit the disease to sons.

(ii) Affected individuals with phenylketonuria lack an enzyme that converts the amino acid phenylalanine into phenylpyruvic acid.

(iii) Klinefelter's syndrome is caused due to the presence of an additional copy of X-chromosome resulting into a karyotype of 47, XXX.

(iv) Failure of segregation of homologous pair of chromosomes during cell division cycle results in Turner's syndrome.

Representative Recessive and Dominant Human Traits

Recessive Traits	Dominant Traits
Albinism	Achondroplasia
Alkaptonuria	Brachydactyly
Blue eyes	Brown eyes
Cystic fibrosis	Huntington disease

Representative Recessive and Dominant Human Traits	
Recessive Traits	**Dominant Traits**
Tongue Nonroller's	Tongue roller's
Duchenne muscular dystrophy	Morphan syndrome
Lesch-Nyhan syndrome	Phenylthiocarbamide **(PTC)** tasting
Fused ear lobes	Free ear lobes
Tay-Sach's disease	Widow's peak

CYTOPLASMIC INHERITANCE

Some self replicating genes (DNA) are present in the cytoplasm (mitochondrial DNA and chloroplast DNA) also. These are called **plasmagenes** and all the plasmagenes together constitute plasmon (like genome). The inheritance of characters by plasmagenes is called **extranuclear** or **extrachromosomal inheritance**.

Certain most important examples of extranuclear inheritance in eukaryotes are following :

Maternal inheritance : The amount of nuclear hereditary material contributed by the two sexes is almost equal but the cytoplasm in egg is always much more than that of the sperm. So, in extranuclear inheritance, contribution of female parent is more. This is called maternal inheritance. The evidence of maternal inheritance is the coiling of shell in snails.

Organelle inheritance : The DNA is present in mitochondria and chloroplast which controls the inheritance of some characters. A well known example of the characters controlled by chloroplasts is plastid inheritance in *Mirabilis jalapa* (4 O'clock plant), discovered by **Correns**. Other examples of organellar inheritance are **iojap inheritance in maize**, **inheritance of poky** (imbalance in the mitochondrial physiology) in the fungus *Neurospora crassa* and **Petite** in yeast, a mitochondrial character. **Cytoplasmic male sterility in maize**, is also a function of defective mitochondria.

EXERCISE

51. Mark the odd one (w.r.t. genomatic mutation)
    (1) Hypoploidy
    (2) Tetrasomy
    (3) Duplication
    (4) Allopolyploidy
52. Find the incorrect match
    (1) Somatic mutation – No evolutionary importance
    (2) Germinal mutation – Gametic mutation
    (3) Frame shift mutation – Gibberish mutation
    (4) Chromosomal mutation – Transversion

53. Substitution of a purine with another type of purine is called

(1) Transversion (2) Transition

(3) Inversion (4) Translocation

54. Inversion without involving the centromere is called

(1) Paracentric

(2) Monosomy

(3) Pericentric

(4) Tautomerization

55. Aneuploidy which results in loss of a complete homologous pair of chromosome is

(1) Trisomy (2) Tetrasomy

(3) Nullisomy (4) Euploidy

56. Which of the following chemical is a base analogue?

(1) 5-bromouracil (2) Acridines

(3) Nitrous acid (4) Hypoxanthine

57. Cytoplasmic male sterility in maize is due to defective

(1) Mitochondria (2) Lysosome

(3) Golgi body (4) Leucoplast

58. Select the incorrect statement w.r.t. pedigree analysis

(1) Solid symbol shows the unaffected individual

(2) It is useful for genetic counsellors

(3) Proband is the person from which case history starts

(4) It is an analysis of traits in a several generations of a family

59. Which of the following abnormalities is due to X-linked recessive mutation?

(1) Cystic fibrosis

(2) Thalassaemia

(3) Klinefelter's syndrome

(4) Lesch-Nyhan syndrome

60. Find odd one (w.r.t. dominant traits in humans)

(1) Blue eyes

(2) Brown eyes

(3) Free ear lobes

(4) Myotonic dystrophy

Some Important Definitions

- **Genetics :** Subject that deals with the inheritance as well as the variation of characters from parents to offspring.
- **Inheritance :** Process by which characters are passed on from parent to progeny.
- **Variation :** Degree by which progeny differ from their parents.
- **True breeding line :** A true breeding line is one that, having undergone continuous self-pollination, shows the stable trait inheritance and expression for several generations.
- **Alleles :** Genes which code for a pair of contrasting traits.
- **Homozygous :** Two alleles of a gene are identical (TT or tt).
- **Heterozygous :** An individual having two different alleles (Tt).
- **Genotype :** Representation of genetic complement of an individual with respect to one or more characters.
- **Phenotype :** It is observable morphological appearance.
- **Dominant allele :** Influences the appearance of the phenotype even in the presence of an alternative allele.
- **Recessive allele :** Influences the appearance of the phenotype only in the presence of another identical allele.
- **Punnett square :** Graphical representation to calculate the probability of all possible genotypes of offspring in a genetic cross.
- **Test cross :** A cross between hybrid (Tt) and homozygous recessive individual (tt).
- **Incomplete dominance :** F_1 phenotype does not resemble either of the two parents and is in between the two.
- **Co-dominance :** F_1 phenotype resembles both of the parents.
- **Multiple allelism :** Presence of more than two alleles for the same character.
- **Pleiotropy :** A single gene product may produce more than one effects.
- **Linkage :** It is physical association of the two genes on similar chromosome.
- **Recombination :** It describes the generation of non-parental gene combinations.
- **Sex determination :** Establishment of sex through differential development in an individual at an early stage of life.
- **Gene mutation :** Alteration of DNA due to change in nucleotide sequence consequently resulting in changes in the genotype and the phenotype of an organism.
- **Pedigree analysis :** Analysis of inheritance of traits in a several of generations of a family.
- **Aneuploidy :** Non-disjunction of two homologous chromosomes during cell division cycle results in the gain or loss of a chromosome(s).
- **Polyploidy :** Failure of cytokinesis after telophase stage of cell division results in an increase in a whole set of chromosomes in an organism.

Formulae Chart

1.	Type of gametes	2^n
2.	Number of zygotes/offsprings	$(\text{Gametes})^2$
3.	Number of phenotype	2^n
4.	Number of genotype	3^n
5.	Number of genotypes for multiple allelism	$\frac{n}{2}(n+1)$ Here, n = Number of alleles
6.	Recombination frequency or cross over value	$\frac{\text{Number of recombinants}}{\text{Total number of offsprings}} \times 100$

n = Number of traits/hybrid (n)

Quick Recap

1. **Genetics** is a branch of biology which deals with principles of inheritance and variation.
2. **Mendelian Inheritance** (Mendelism)
   - (i) Mendel proposed that something was being stably passed down, unchanged, from parent to offspring through the gametes, over successive generations. He called these things as 'factors'.
   - (ii) The dominant characters are expressed when factors are in heterozygous condition (Law of Dominance).
   - (iii) The characters never blend in heterozygous condition.
   - (iv) The recessive characters are only expressed in homozygous condition.
   - (v) A recessive trait that was not expressed in heterozygous condition may expressed again when it become homozygous. Hence, characters segregate while formation of gametes (Law of Segregation).
   - (vi) Mendel also studied the inheritance of two characters together and he found that the factors independently assort and combine in all permutations and combinations (Law of Independent Assortment).
3. The factors on chromosomes regulating the characters are called the **genotype** and the physical expression of the characters is called **phenotype**.
4. Walter Sutton and Theodore Boveri noted that the behaviour of chromosomes was parallel to the behaviour of genes and used chromosome movement to explain Mendel's laws.
5. Mendel's law of independent assortment is not true for the genes that were located on the same chromosomes (*i.e.*, **linked genes**).
6. Closely located genes assorted together, and distantly located genes, due to recombination, assorted independently.

7. **Frequency of recombination** between gene pairs on the same chromosome is a measure of the distance between genes.
8. **Mutation** is defined as change in the genetic material. A **point mutation** is a change of a single base pair in DNA. Some mutations involve changes in whole set of chromosomes (polyploidy) or change in a subset of chromosome number (aneuploidy).
9. Sickle-cell anaemia is caused due to change of one base in the gene coding for β-chain of haemoglobin.
10. Inheritable mutations can be studied by generating a **pedigree** of a family.
11. **Down's syndrome** is due to trisomy of chromosome 21. In **Turner's syndrome**, one X-chromosome is missing and the sex chromosome is as XO.
12. In **Klinefelter's syndrome**, the condition is XXY.

❑ ❑ ❑