# KRISHNAGAR ACADEMY

# BIOLOGY -

# PRINCIPLE OF INHERITANCE &VARIATION

Class -12

# THE CHROMOSOMAL THEORY OF INHERITANCE

Soon after the rediscovery of Mendel's laws in 1900, **Walters S. Sutton** (an American graduate student) and **Theodor Boveri** (a German biologist), in 1903, have independently observed a parallelism between the behaviour of chromosomes and Mendelian factors (genes). They noticed several similarities between the two.

## A Comparison between the Behaviour of Chromosomes and Mendelian Factors (Genes)

(1) Chromosomes occur in pairs like the alleles of a Mendelian factor (now known as genes).
(2) The homologous chromosomes segregate during meiosis like the pair of similar or dissimilar alleles of a Mendelian factor segregate at the time of gamete formation.
(3) Different chromosomes orient and separate independently during meiosis like Mendelian factors.
(4) The paired condition of both chromosomes and Mendelian factors is restored during fertilization.
(5) Both, Mendelian factors and chromosomes maintain their individuality from generation to generation.

These similarities led **Sutton** and **Boveri** to postulate the **Chromosomal Theory of Inheritance**. They noted that the behaviour of chromosomes was parallel to the behaviour of genes

and used chromosome movement to explain Mendel's laws. This theory states that **Mendelian factors (genes) are located on chromosomes, and it is the chromosome that segregates and assorts independently during meiotic cell division.** During meiosis (gamete formation) the two chromosomes of a homologous pair get separated from each other. Since factors (genes) are located on chromosomes, they also get separated. A simplified version of the chromosomal theory is presented in figure 16 taking only two pairs of chromosomes and an equal number of genes. The figure shows the movement of alleles of two genes (seed shape and seed colour) located on two different chromosomes during a dihybrid cross of round-yellow and wrinkled-green pea seed varieties. The two duplicated and paired homologous chromosomes (a tetrad of four chromatids) assembled at the metaphase stage of meiosis I can separate in two possible ways. This leads to independent assortment of the chromosomes and the genes they carry in four possible ways. After meiosis II, four types of gametes are formed, each with half the number of chromosomes and genes. The two parental types and the two new combinations (recombinants) are formed in equal ratio. Two genes located on separate chromosomes produce 50 per cent recombinants during meiosis. Thus the $F_2$ progeny exhibits phenotypic dihybrid ratio of 9 : 3 : 3 : 1. In a test cross, the two parental types (round-yellow and wrinkled-green) and the two recombinants (round-green and wrinkled-yellow) are formed in equal ratio. These results confirm the ratio of each type of gamete formed by the dihybrid following independent assortment of chromosomes carrying the alleles of two genes.

**NOTE**

The term 'factor' (used by Mendel) was replaced with the term 'Gene' by Johannsen in 1909.

## LINKAGE

**Walters S. Sutton** (1903), commenting on the chromosomal theory of inheritance, indicated that since the number of genes in an organism is much larger than the number of pairs of chromosomes, each chromosome pair should contain several genes. This means that during meiosis chromosomes move as a unit and all the genes of a chromosome go together and do not assort independently as expected according to Mendel's law of independent assortment. Only the genes located on different chromosome pairs assort independently. This also implies that genes situated on the same chromosome should be transferred *en bloc* from one generation to another, i.e. they do not assort independently. The group of genes which behaves as a unit is said to be **linked** and the phenomenon is known as **linkage**. All the genes on a pair of homologous chromosomes collectively form a group, known as **linkage group**. The number of linkage groups in an organism is the same as the haploid number of chromosomes or the number of pairs of chromosomes. This indicates that the genes linked together are situated on the same chromosome. For instance, *Drosophila melanogaster* has four linkage groups which correspond to $n = 4$ (number of homologous pair of chromosomes). The genes which constitute a linkage group are inherited together and not independently. Thus, linkage may be defined as the **tendency of two or more genes of the same chromosome to remain together during the process of inheritance**. According to **T.H. Morgan**, the degree or intensity with which two genes are linked together is known as **linkage value**. The linkage value depends upon the distance between the linked genes on the same chromosome.

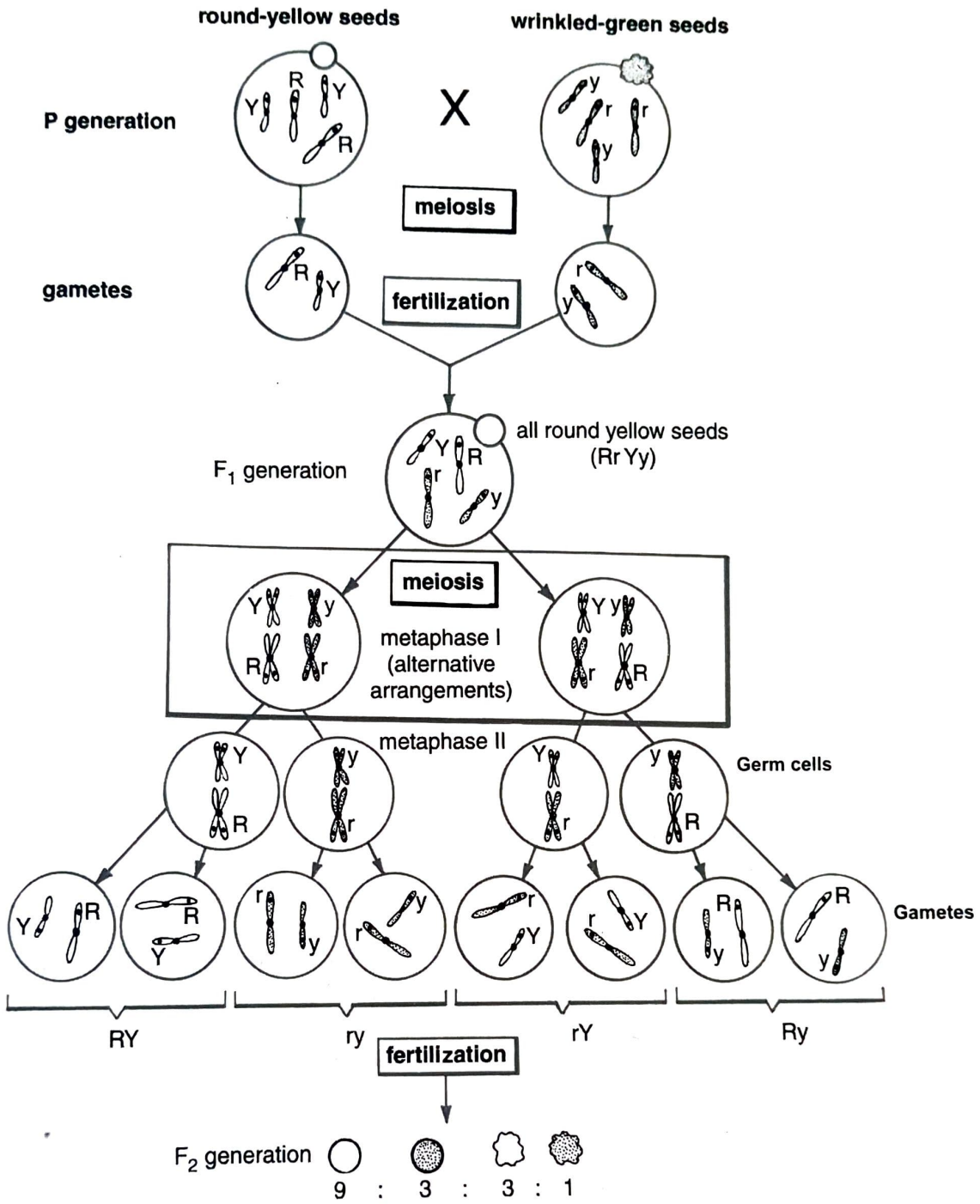


Fig. 16. Mendel's law of independent assortment of factors (alleles, R, r, Y, y), described in terms of the separation of homologous chromosomes which occurs during meiosis.

## Types of Linkage

There are two types of linkage, **complete** and **incomplete**.

**Complete linkage. Complete linkage** is exhibited when the genes for a particular character are present very close to one another. The genes located on the same chromosome do not separate and are inherited together over the generations due to the absence of crossing over. It allows combination of parental traits to be inherited as such. It is due to non-break in the gene combination situated on a chromosome. It is very rare in nature. The best example of complete linkage is male *Drosophila* for grey body colour and long wings. **Morgan** (1920) demonstrated through his experiment on *Drosophila* that a cross between the male with grey body and long wings and the female with two recessive characters, black body and vestigial wings, results in

$F_1$ hybrids which are like the male parent. If a male from $F_1$ generation is test crossed with a double recessive black vestigial female, four kinds of offsprings in equal numbers as the result of independent assortment are expected. There are however, only two types of offsprings which resemble the two grand parents *(Fig. 17)*. This suggests that the genes for grey body and long wings are linked together and thus show complete linkage.

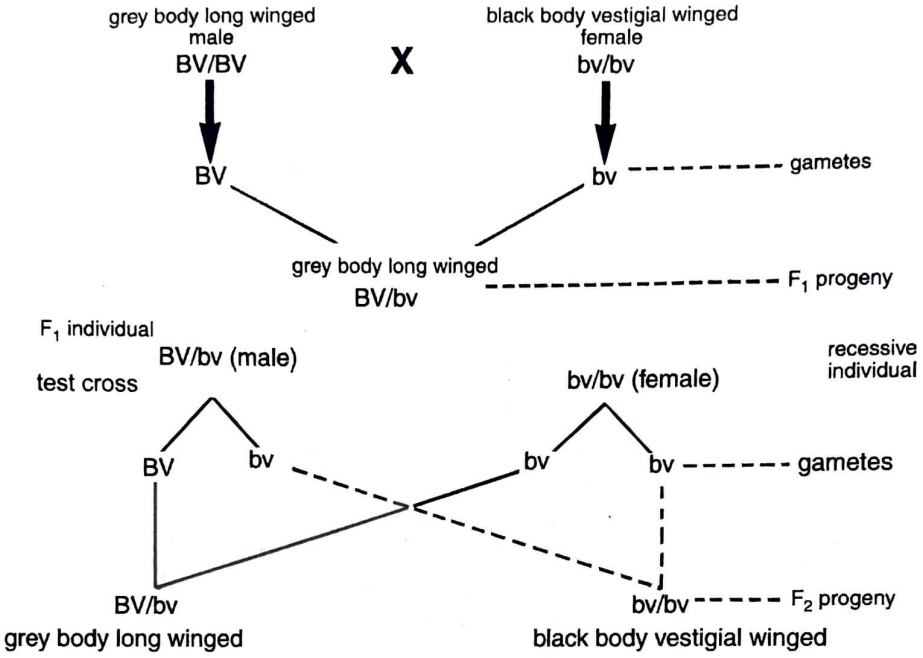


**Fig. 17.** Complete linkage in *Drosophila* fly.

**Incomplete linkage. Incomplete linkage** occurs when the genes for different characters are separated at the time of gamete formation due to breaking and exchange of chromosome pieces during meiosis. Genes present in the same chromosome have a tendency to separate due to crossing over and hence produce recombinant progeny. In sweet pea, blue flower colour and long pollen character exhibit incomplete linkage. When a sweet pea variety with blue flowers (B) and long pollen (L) is crossed with another variety having red flowers (b) and round pollen (l), $F_1$ individuals (Bb Ll) produced blue flowers and long pollen *(Fig. 18)*. These $F_1$ individuals when crossed with plants having red flowers and round pollen (bb ll), we find the test cross frequency as shown below.

| Phenotype | Gene combination | Test cross frequency | | |
|---|---|---|---|---|
| | | Observation | Expected if independently assorting | Expected if fully linked |
| Blue-long | Parental | 43.7% | 25% | 50% |
| Blue- round | Recombinant | 6.3% | 25% | 0% |
| Red-long | Recombinant | 6.3% | 25% | 0% |
| Red-round | Parental | 43.7% | 25% | 50% |

In independent assortment, 1 : 1 : 1 : 1 ratio is expected in a test cross. But instead 7 : 1 : 1 : 7 ratio was actually obtained, indicating that there was a tendency in dominant alleles to remain together. The above results show that two new combinations (blue-round and red-long) also occur in $F_2$ generation, but their frequency is only 12.6 per cent. The fully linked genes, however, should exhibit zero per cent recombination. The answer lies in the crossing over, i.e. there was physical exchange of parts of non-sister chromatids of homologous chromosomes following synapsis at meiosis.

## Factors Affecting Linkage

The linkage is affected by several factors. Some important ones are as follows :

1. **Distance.** Strength of linkage is inversely proportional to the distance between the linked genes.
2. **Age of organism.** With increasing age, the chances of crossing over are reduced and hence the linkage increases.
3. **X-rays.** Exposure to X-rays reduces the strength of linkage.
4. **Temperature.** Increase in temperature decreases the strength of linkage as chances of recombination increases.

## Significance of Linkage

The phenomenon of linkage has great significance as it reduces the possibility of variability in gametes unless crossing over separates the linked genes.

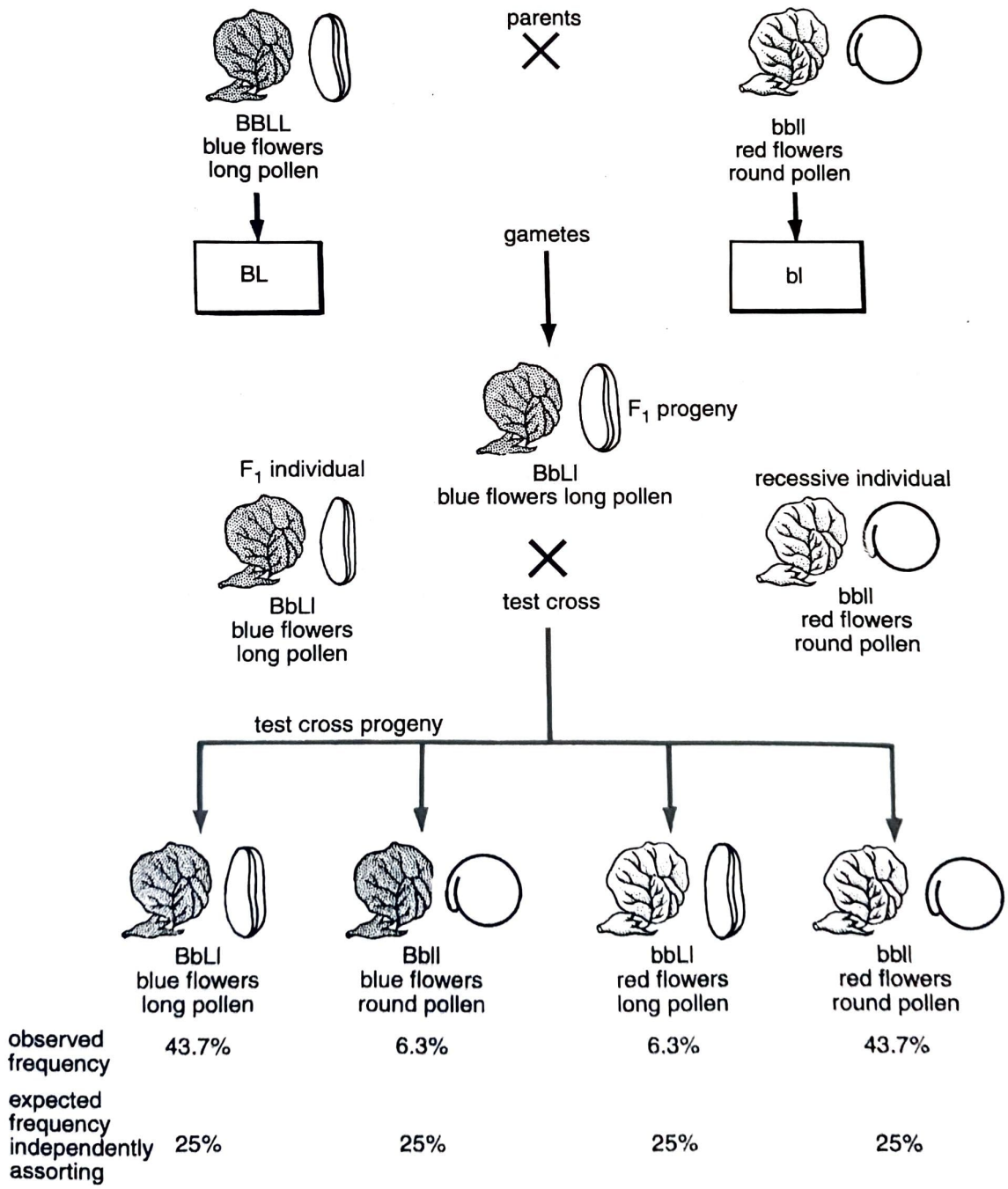


Fig. 18. Incomplete linkage in sweet pea.

## Linked and Unlinked Genes

Genes which are present on the same chromosome are called **linked genes** and they together form a linkage group.

The genes which are present on a chromosome together form a distinct group called **linkage group**. The number of linkage groups in an individual is similar to the haploid number of the organism. For example, 23 in man, 7 in garden pea, 4 in *Dorsophila*, 2 in *Mucor* and 10 in maize.

Two genes located on different chromosomes are called **unlinked genes**.

Unlinked genes follow the principle of independent assortment and give 9 : 3 : 3 : 1 in a $F_2$ phenotypic ratio. In test cross, they always give 1 : 1 : 1 : 1 ratios.

The linked genes do not follow principle of independent assortment. For the same reason Mendel was not able to observe the linkage, because the genes of all the seven characters he considered, were having no linkage among them and thus he always got 9 : 3 : 3 : 1 ratio.

# CROSSING OVER

**Mutual exchange of blocks of homologous genes between a pair of homologous chromosomes is known as crossing over.** A cross-over between linked genes allows their recombination during meiosis.

## Mechanism of Crossing Over

Crossing over involves breaking and rejoining of chromosomes in the synaptonemal complex. During zygotene and pachytene stages of the prophase I of meiosis, pairing takes place in such a way that similar parts of chromosomes lie side by side, probably by mutual attraction between allelic genes. During diplotene, each chromosome splits longitudinally into two chromatids so that each bivalent is now composed of four chromatids. The chromatids originating from the same chromosome are called **sister chromatids**. During the process of crossing over, the two non-sister chromatids come in contact at certain points. This is the region where crossing over takes place by breakage and reciprocal fusion of two non-sister chromatids. The points or places where homologues are held together and exchange bits of chromatids are known as **chiasmata** *(Fig. 19 A-G)*. The number of chiasmata in a chromosome depends on its length.

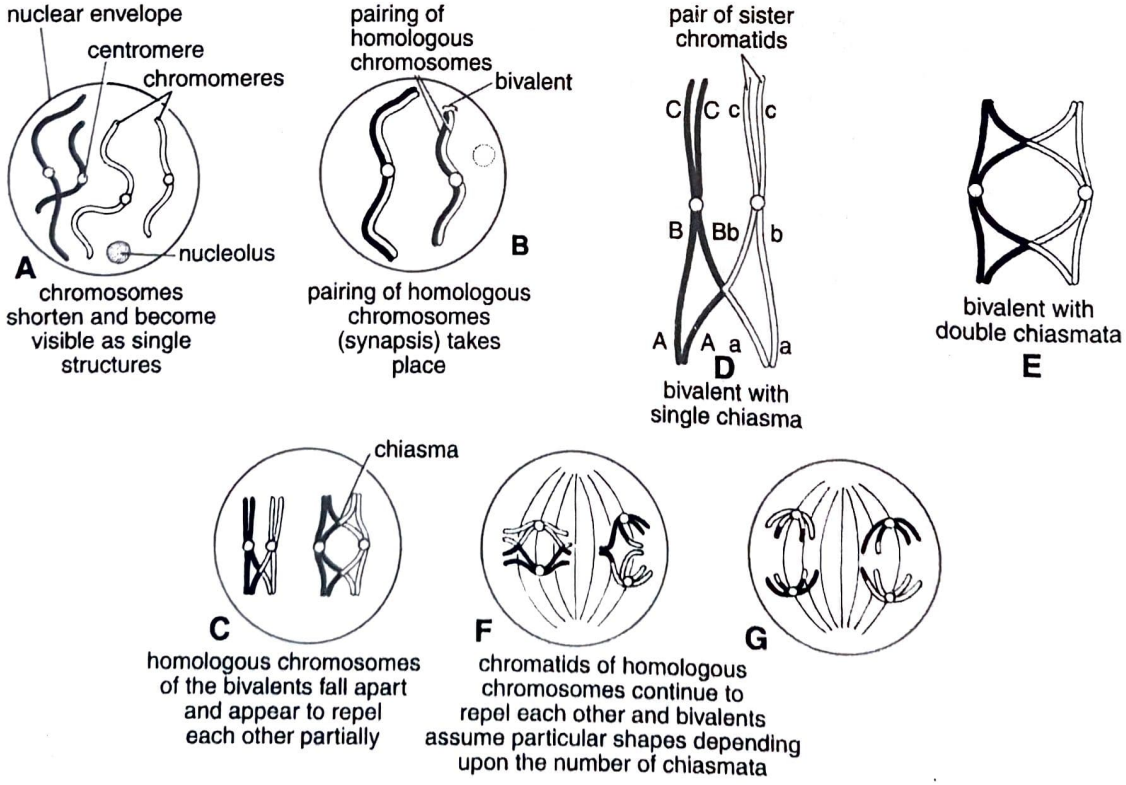


Fig. 19 A-G. Process of crossing over during meiosis I.

Crossing over is a highly precise process as the two chromatids exchange exactly equivalent segments and except in very rare instances neither they loss nor gain any gene. At each chiasma, the two non-sister chromatids exchange their section so that the chromosomes carried in the gametes are new as they carry genes which were originally not located in them.

## Types of Crossing Over

The type of crossing over depends upon the number of chiasmata present in chromosomes. The following three types of crossing over have been recognised depending on the number of chiasmata.

1. **Single crossing over.** In **single crossing over**, there is only one chiasma where chromatids of homologous chromosomes contact. The chromosomes break only at one point along their entire length. Bivalents having a single chiasma appear as open crosses *(Fig. 20 A)*.
2. **Double crossing over.** In **double crossing over**, the crossing over occurs at two points in the same chromosome pair, i.e. the chromatids break and rejoin at two points. Here two chiasmata produce a ring shape *(Fig. 20 B)*.
3. **Multiple crossing over.** In **multiple crossing over**, the number of chiasmata formed is more than two in the same chromosome. The chiasmata produce loops lying at right angles to each other *(Fig. 20 C)*.

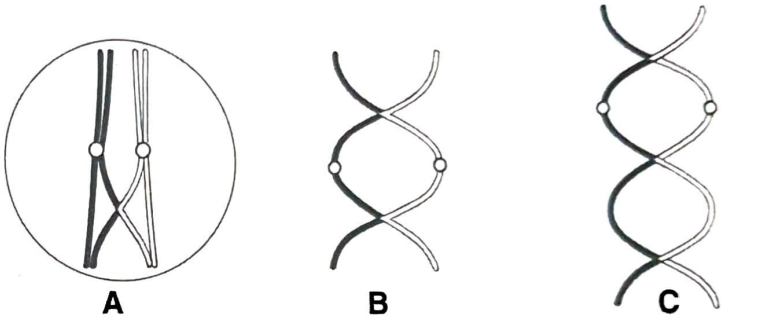

**Fig. 20 A-C.** Types of crossing over : **A.** single; **B.** double; **C.** multiple.

## Factors Affecting Crossing Over

Several factors affect crossing over. Some important ones are as follows :

1. **Distance.** The frequency of crossing over is directly proportional to the distance between them in the chromosome. In other words, the more distant two genes are in a chromosome, the greater opportunity they have for crossing over.
2. **Age.** With advancing age, crossing over frequency generally decreases.
3. **X-rays.** Exposure to X-rays increases the frequency of crossing over.
4. **Temperature.** Variations in temperature increase crossing over frequency.

## Significance of Crossing Over

The phenomenon of crossing over is of great significance. It provides an inexhaustible store of genetic variability in sexually reproducing organisms. Since, crossing over helps in the development of new characters, it is of paramount important in plant breeding. As a result of crossing over new gene combinations are produced which play an important role in micro-evolution. The frequency of crossing over is helpful in the mapping of chromosomes. Crossing over also affords a proof for the linear arrangement of genes.

# AUTOSOMES AND SEX CHROMOSOMES

In organisms where two sexes are distinct, certain chromosomes (usually one or two) in a diploid cell differ from the rest in staining reaction and behaviour during cell division. These chromosomes determine the sex of an individual and are thus called **sex chromosomes**. The rest of the chromosomes are said to be **autosomes**. In a diploid individual, there are $2n-2$ autosomes and two sex chromosomes. In certain insects like grasshoppers and roundworms females have two sex or X-chromosomes while males have only one, and thus females are designated XX and males XO. In humans and other mammals, most insects (e.g., *Drosophila*, fruit fly) and many plants (e.g., *Coccinia*, *Melandrium*), females have two X-chromosomes (XX), whereas males have one X and a morphologically distinct Y-chromosome (XY) *(Fig. 21 A-B)*. Y-chromosome, though different in size and shape, pairs with X during meiosis. Thus females are XX and males XY. Since males produce two types of gametes, X or O in XO type and X or Y in XY type, they are said to be **heterogametic**. Females are homogametic, producing only one type of gamete with an X-chromosome.

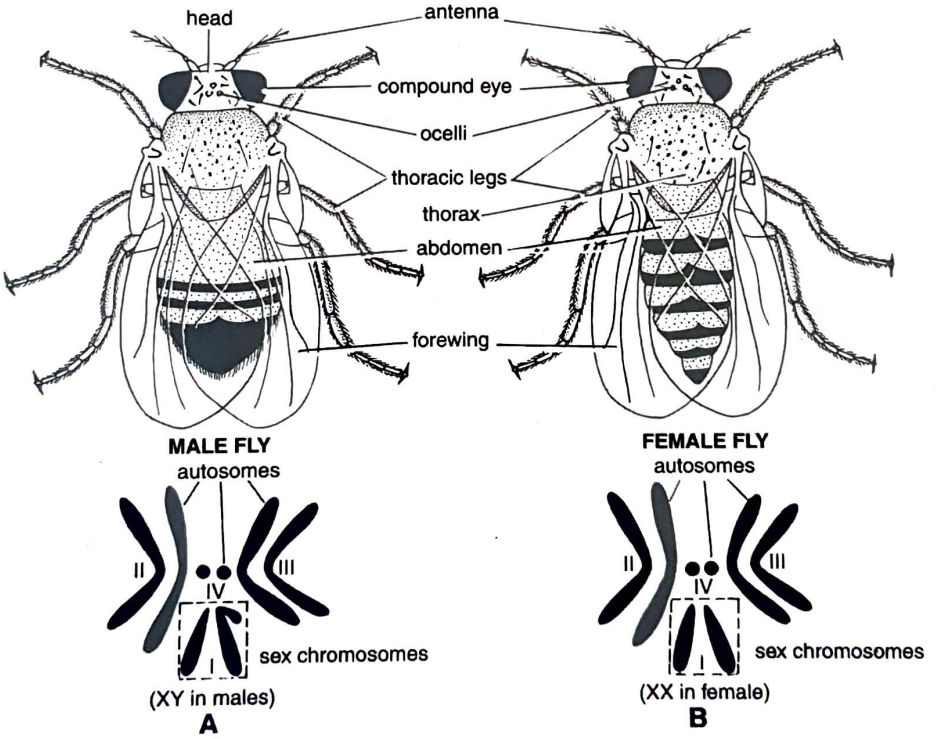


**Fig. 21 A-B.** Male and female fruit flies (*Drosophila melanogaster*) and their diploid chromosomes complement (Note that female is larger and has an ovipositor at the tip of the abdomen; chromosome pair numbered I is sex chromosomes which is XY in male and XX in females and rest numbered II, III and IV are autosomes).

However, in birds and some reptiles, females are heterogametic while males are homogametic. To avoid confusion with the male heterogametic XY system, this is referred to WZ system; with males WW and females WZ.

# SEX DETERMINATION

**Sex determination deals with the study of factors which are responsible for making an individual male, female or a hermaphrodite.** Both biologists and non- biologists were puzzled for long by the riddle that what determines the sex of an offspring. Numerous mistaken hypothesis and wild guesses were put forward but a valid solution became possible only with the discovery of sex chromosomes in the early years of the 20th century.

In 1891, **Henking** traced a specific nuclear structure all through spermatogenesis in some insects and he observed that 50 per cent of the sperm received this structure after spermatogenesis, whereas the other 50 per cent sperm did not receive it. **Henking** called this structure as **'X-body'** but unable to explain its significance. Later, it was found that **'X-body'** of **Henking** was in fact a chromosome and that is why it was given the name **X-chromosome**. Due to the involvement of the X-chromosome in the determination of sex, it was designated as **sex chromosome** and the rest of the chromosomes was named as **autosomes**.

## Basis of Sex Determination

Sex of individuals in different organisms is determined by three methods - **environmental**, genic and **chromosomal**.

1. **Environmental determination of sex.** In crocodiles and some lizards, high temperature induces maleness and low temperature femaleness. In turtles, males are predominant below 28°C females above 33°C and equal number of the two sexes between 28°C - 33°C.
2. **Genetic determination of sex.** In bacteria, fertility factor present in plasmid, determines sex. *Chlamydomonas* possesses sex determining genes.
3. **Chromosomal determination of sex. E.B. Wilson** and **Stevens** (1905) put forward this theory and was further extended by **T.H. Morgan** and **C.McClung**. This theory suggests that all unisexual organisms have one or more sex chromosomes, which are different in male and female individuals. The X and Y bodies were named as sex chromosomes X and Y. (Initial clue about X chromosome as X body was given by **Henking**). In humans as well as *Drosophila*, the female has two X chromosomes, while male has one X and one Y chromosomes. Hence, female produces only one type of gametes with X chromosomes (homogametic) and males produce two type of gametes with X and Y chromosomes (heterogametic). Chromosomal determination of sex is of following types :

**(i) Female is Homogametic and Male is Heterogametic :**

**(a) XX – XY Type :**

**Examples.** Human beings, *Drosophila* (fruit fly) and many angiospermic plants.

In this case female produces one kind of gametes only, containing X chromosomes (XX) but males produce two kinds of gametes, one with X chromosome and the other with Y chromosome (XY).

Female is homogametic with AA+XX chromosome

Male is heterogametic with AA+XY chromosome *(Fig. 22)*.

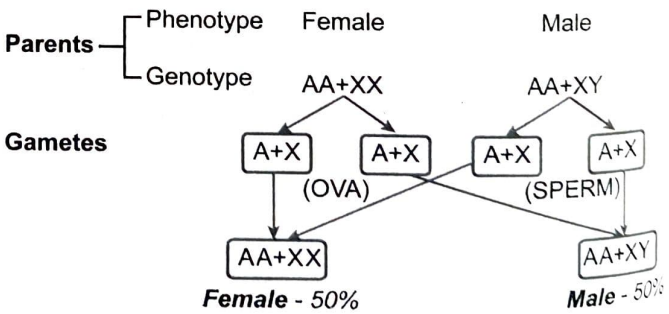


Fig. 22 Sex determination : XX-XY type.

**(b) XX – XO Type :**

**Examples.** Some plants like *Vallisneria*, *Dioscorea* and many insects like true bugs, grasshoppers and cockroaches.

In this case, female produces one kind of gametes only containing X chromosomes (XX) but male produces two kind of gametes, one with X chromosome (only one sex chromosome X) and there is no second sex chromosome.

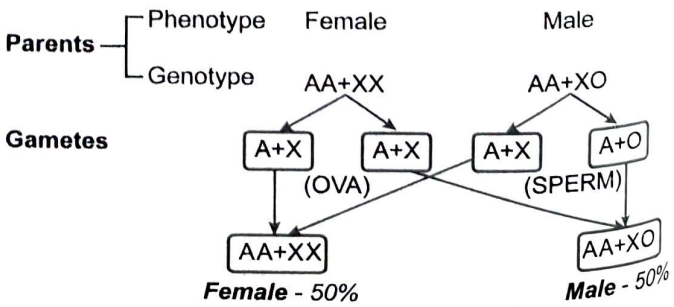


Fig. 23 Sex determination : XX-XO type.

Therefore, males are designated as XO.

Female is homogametic with AA+XX chromosome *(Fig. 23)*.

Male is heterogametic with half male gametes (gynosperm) carrying X chromosome (A+X) while the other half (androsperms) being devoid of it (A+O).

**(ii) Male is Homogametic and Female is Heterogametic**

Here, the female produces two types of gametes (heterogametic), whereas the males produces only one type of gametes (homogametic). Letters Z and W are used instead X and Y, in order to avoid any confusion.

**(a) ZW – ZZ Type :**

**Examples.** Birds, many fishes and reptiles.

In this case, the female produces two types of eggs(ova). One with single Z chromosome and another with single W chromosome. The males on other hand produce only one kind of sperm carrying only Z chromosome.

Female is heterogametic and produces two types of eggs A+Z and A+W.

Male sperms are of one type A+Z, *(Fig. 24)*.

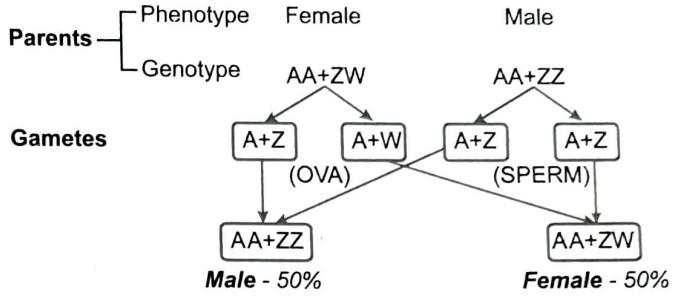


Fig. 24. Sex determination : ZW-ZZ type.

**(b) ZO – ZZ Type :**

**Examples.** Many moths and butterflies.

In this case, the female produces two types of eggs (ova) with single Z chromosome and without any sex chromosome. The males on other hand produce only one kind of sperm carrying Z chromosome only.

Females are heterogametic with female gamete A+Z and A+O.

Males are homogametic with AA+ZZ chromosome *(Fig. 25)*.

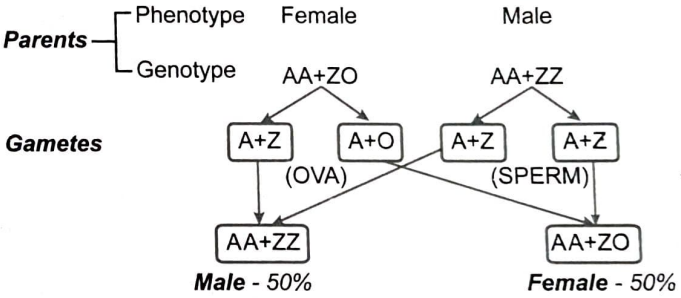


Fig. 25 Sex determination : ZO-ZZ type.

## Sex determination in honey bee (Haplodiploid mechanism)

In honeybees, the females (queens and workers) are diploid. They develop from fertilized eggs having $2n$ number of chromosomes. The males (drones) are haploid and have only $n$ number of chromosomes. They develop from unfertilized haploid eggs by parthenogenesis. This is called **haplo-diploid mechanism of sex determination** by number of sets of chromosomes and not by X and Y chromosomes *(Fig. 26)*.

The genome (haploid number) in honeybee contains 16 chromosomes.

The individual formed by fusion of male and female gametes is diploid as it receives two genomes (one from sperm and other from ovum). This diploid individual develops into a female honeybee (all female honeybees have 16 + 16 = 32 chromosomes). On other hand, some of the ova remain unfertilized and develop into haploid individuals with only 16 chromosomes (**Parthenogenesis**). They develop into male honeybees with only one genome (haploid with 16 chromosomes).

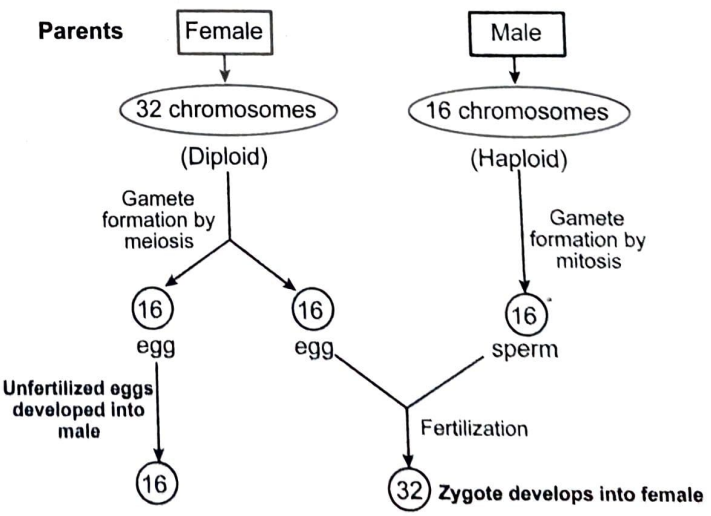


Fig. 26. Sex determination in honeybee.

# SEX-LINKED INHERITANCE

Mendel's laws of inheritance indicate that reciprocal crosses give the same results despite the fact that a particular character is present in male or female parent. For instance, in pea, when tall plants are crossed with dwarf plants, tall plants are invariably obtained in $F_1$ generation, whether tall plants were used as male or as female parent. Such a situation always exists in bisexual plants or in hermaphrodite animals. However, in dioecious individuals, there can be two kinds of characters : (i) characters which do not show any differences in reciprocal crosses (male A X female B ; male B X female A), and (ii) characters which show a difference in reciprocal crosses. The former characters are located on autosomes, whereas the latter are present on sex chromosomes.

# SEX-LINKED TRAITS

In most unisexual organisms, a pair of sex chromosomes is found besides a set of autosomes. All genes situated on the sex chromosome will show linkage. To understand the inheritance of characters present on sex chromosomes, let us take the example of *Drosophila* or man. In the male individuals of these organisms, the sex chromosome pair is heteromorphic (XY) involving one X-chromosome and one Y-chromosome, while in females it is homomorphic (XX) with two X-chromosomes. As shown in figure 27 A, X-chromosome from the male individual invariably goes to the daughter, while X-chromosomes from the female individual may go to the daughter or son. Consequently, if female parent carries recessive character in homozygous condition and male parent carries dominant allele, in $F_1$ generation the female individuals will always show dominant phenotype (heterozygous) and the male individuals only recessive phenotype *(Fig. 27 B)*.

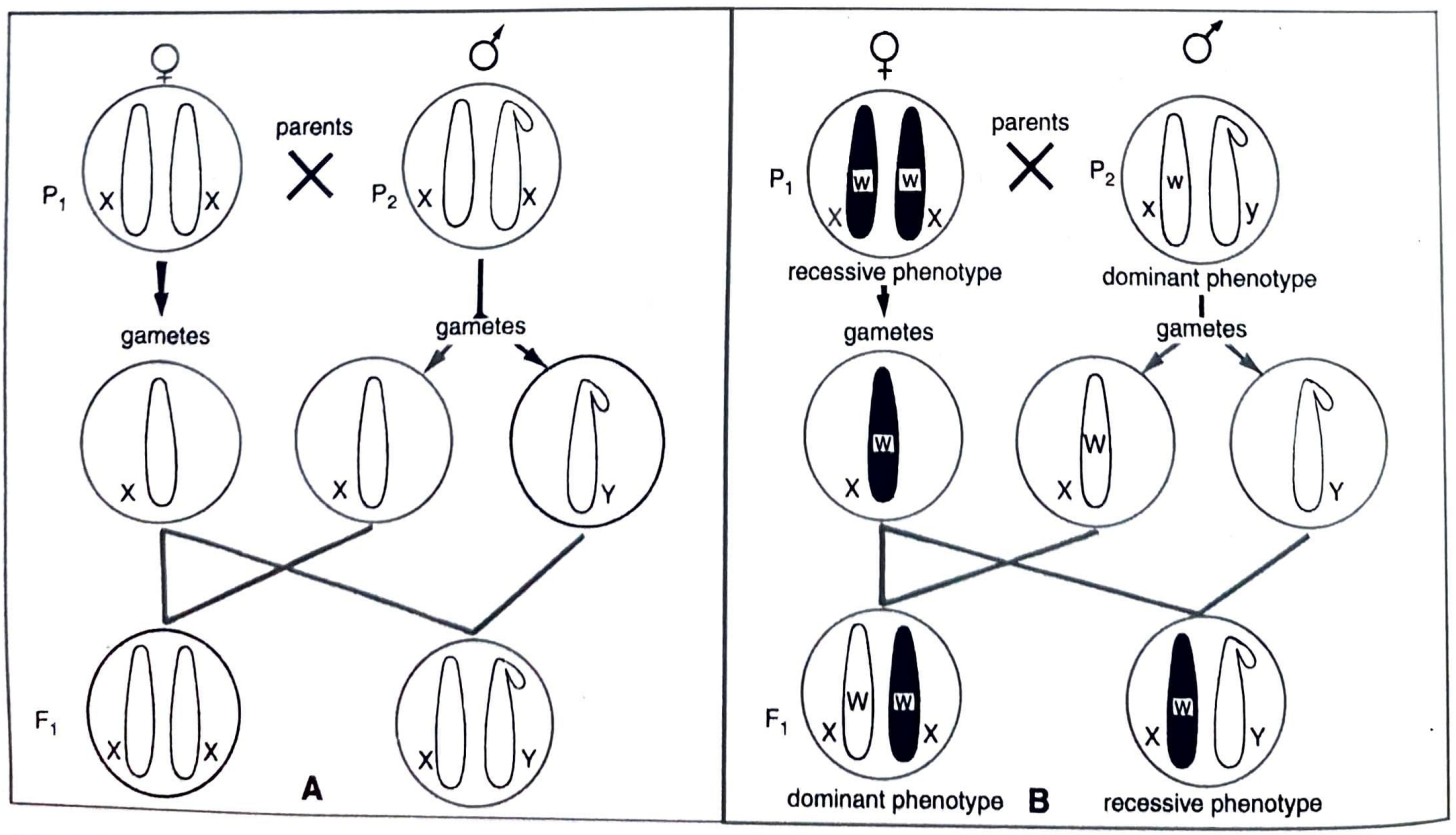


**Fig. 27 A-B.** Sex-linked characters : **A.** transmission of sex chromosome (x) from two parents to their female and male offsprings; **B.** inheritance of sex-linked characters.

Give an example of lethal genes.
Give the meaning of term allele.
What was the experimental material used by Mendel?
What are pleiotropic genes?
Define $F_1$ and $F_2$ generations.
What for the genetic symbols AA and Aa stand?
Mendel studied seven characters in garden pea. Which one or more of the following were recessive : Wrinkled seed, axillary flower, yellow colour of food?
What is allelomorphic pair?
What is pleiotropy?
Name any one plant and its feature that shows the phenomenon of incomplete dominance.
What are the total number of true breeding varieties of garden pea which Mendel had taken to start his experiments?
What do the symbols square and circle in a pedigree chart indicate?
What is meant by polygenic trait?
What for genetic symbols AA and Aa stand?
Name the process which is antagonistic to linkage.
What are the units of heredity?
Name the type of cell division in which number of chromosomes halved in daughter cells.

1. Name two kinds of linkage.
2. Recalling meiosis, indicate at what stage a recombinant DNA is made?
3. What is allelomorphic pair?
4. What is allele?
5. Mention how does DNA polymorphism arise in a population.

In garden pea smooth seeded (S) character is dominant over wrinkled seeded (s) character and red flower (R) over white (r) flowers. Determine the genotype and phenotype of the following crosses :

(a) Ssrr × SsRR (b) SsRr × Ssrr (c) SsRr × ssrr (d) SSRR × SSRR (e) SsRr × SsRr

Explain the basis of sickle cell anaemia with the help of genotype outline.

(a) What offspring would be expected from mating a roan cow with a roan bull? A roan with a white? A red with a white?

(b) Give an example of genotypes such that two left handed parents would produce only right handed children. Only left handed. Both.

A man with AB blood group has married a woman with O group. Show the possible genotypes and phenotypes of the progeny.

Study the following carefully and explain why mutation (A) did not cause any sickle cell anemia inspite of change in the