ST. LAWRENCE HIGH SCHOOL

A JESUIT CHRISTIAN MINORITY INSTITUTION

STUDY MATERIAL -6

Class: XII **Sub: BIOLOGICAL SCIENCE** **Date: 27.06.2020**

Topic - Ch 5 (part 2) Principles of inheritance and variations

NON-MENDELISM

PLEIOTROPY OR PLEIOTROPISM

Definition : When a single pair of genes influence more than one character, this phenomenon of controlling more than one characters at the same time is called pleiotropism. Such genes are called pleiotropic genes. These genes produce more than one phenotypic effects which are totally unrelated.

Explanation : Mendel proposed law of unit character which means each character is controlled by one gene. A pleiotropic gene along with controlling the manifestation of its phenotypic trait, also influences some other related or unrelated character/ characters, i.e., a single gene produces more than one phenotypic effects on a number of body characters.

Mechanism of Pleiotropy : The pleiotropic effect is produced by a gene owing to a cascade of reations during some metabolic pathway which are influenced from the original gene product and contribute to different phenotypic effects.

Example 1 : Phenylketonuria (PKU) : Phenylketonuria is an autosomal recessive character controlled by a mutant gene present on 12^{th} chromosome. The mutant gene fails to code for the enzyme phenylalanine hydroxylase (PAH) needed for the normal metabolism of amino acid phenylalanine to tyrosine. This results in the accumulation of phenylalanine in the body fluids lie blood, sweat and cerebrospinal fluid and appearance of an abnormal breakdown product phenylketone in urine. Accumulated phenylalanine and breakdown products cause severe brain damage leading to mental retardation. Such persons are fair-skinned with blonde hair and blue eyes because of failure of melanin synthesis.

Example 2 : Sickle cell anaemia : A recessive gene causes production of abnormal haemoglobin .As a consequence the shape of the red blood cell containing it becomes distorted and sickle shaped .Sickle shaped red blood cells not only transport oxygen poorly ,resulting in anaemia ,but also tend to clump together and clog small arteries. Thus, homozygous individuals for sickled shaped cells normally die early in life due to severe anaemia ,whereas heterozygous state confers protection agaist malaria as the parasite cannot live on distorted cells.

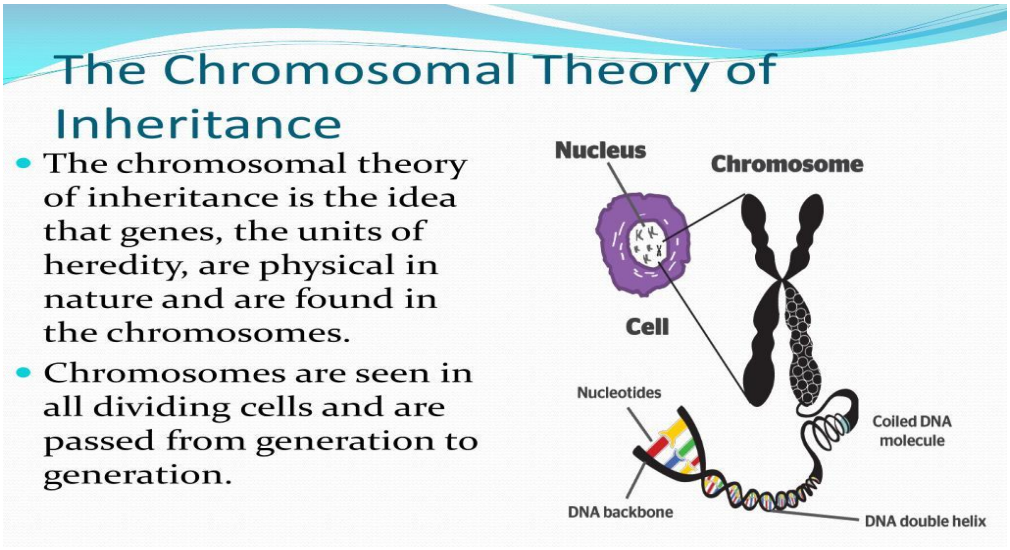
THE CHROMOSOME THEORY OF INHERITANCE

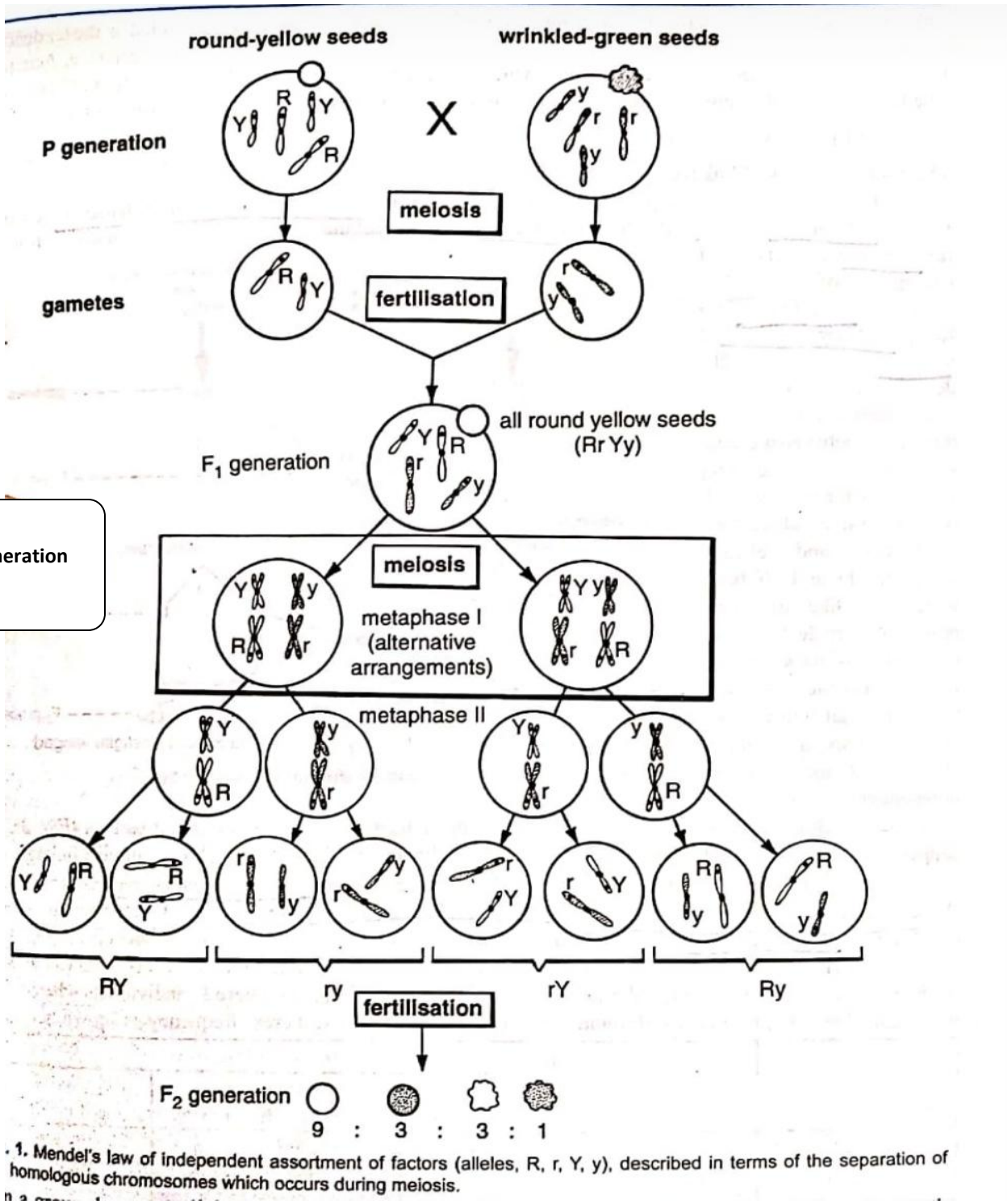
Soon after the rediscovery of Mendel's laws in 1990, Walters S. Sutton (an American graduate student) and Theodor Boveri (a German biologist), in 1903, have independently

observed a parallelism between the behaviour of chromosomes and mendelian characters. They noticed the following similarities between the two.

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

The similarities led Sutton and Boveri to postulate the Chromosome Theory of Inheritance. **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**. A simplified version of the chromosomal theory is presented in figure 1 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 paired homologous chromosomes (a tetrad of four chromatids) assembled at the metaphase stage of meiosis 1 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 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.





F_2 generation

1. 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.

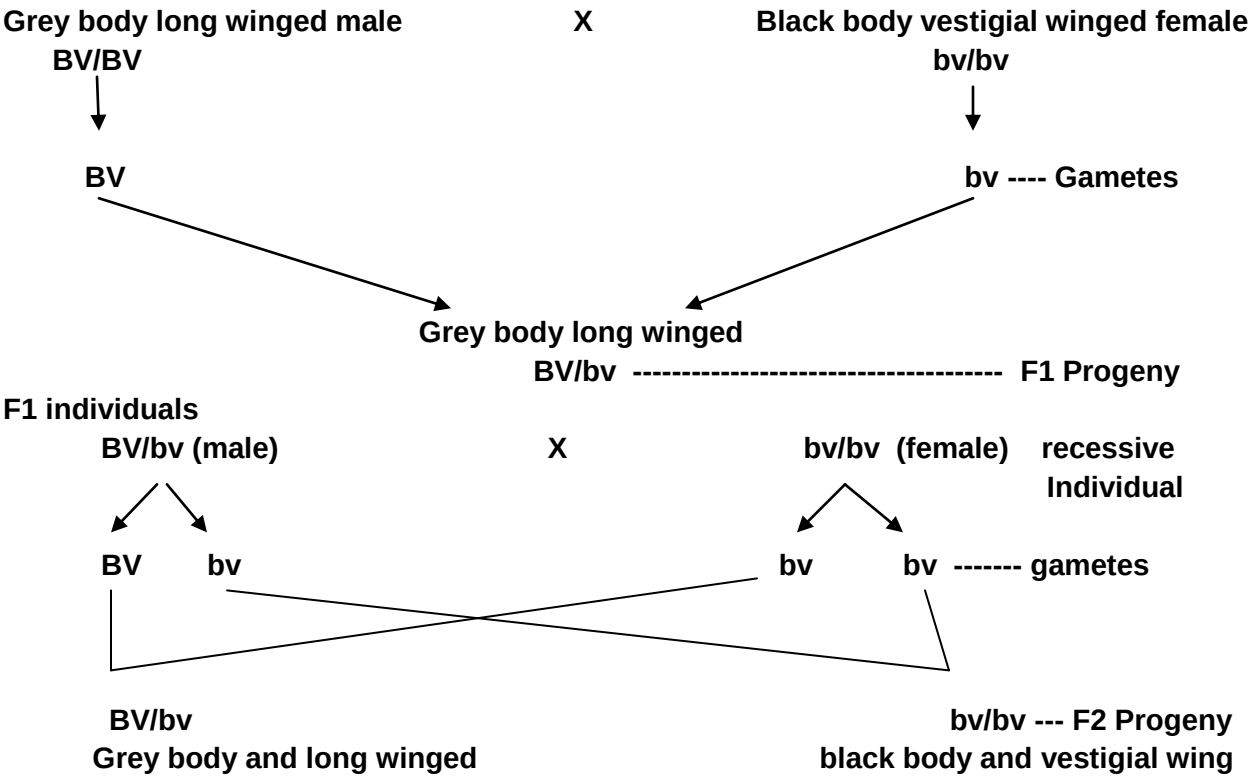
LINKAGE

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 4 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 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.**

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. 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 back crossed with a double recessive black vestigial female, four kinds of offsprings in equal numbers as the result of independent assortment are expected. But, there are only two types off offsprings which resemble the two grand parents. This suggests that the genes for grey body and long wings are linked together and thus show complete linkage.



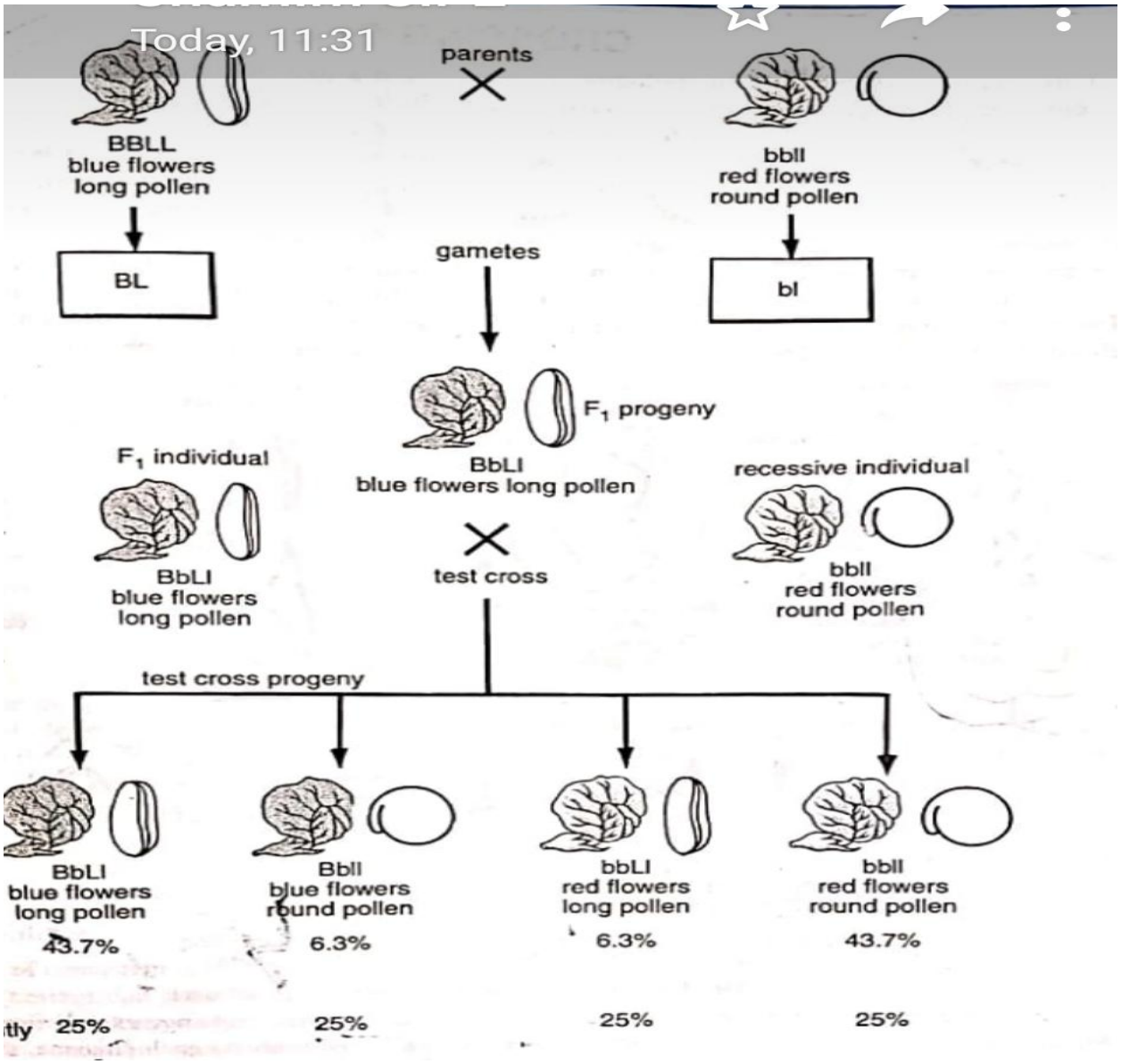
Complete linkage in *Drosophila* fly

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. 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 3). 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%	50%
Red-long	Recombinant	6.3%	25%	50%
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.



Incomplete linkage in *Lathyrus odoratus*

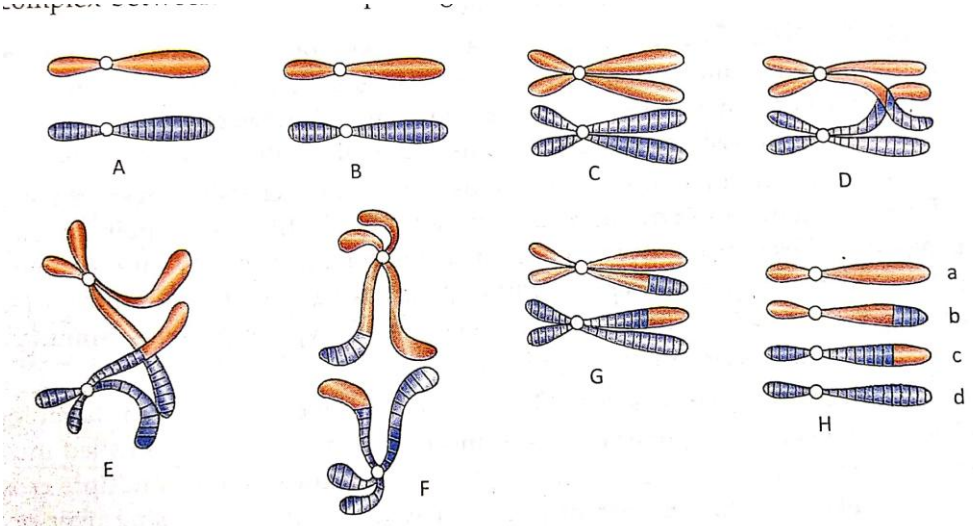
The above results show that two new combinations (blue-round and red-long) also occurs in F_2 generation, **but their frequency is only 12.6 per cent. The fully linked genes, however, should exhibit zero per cent recombination. The answer lies in the crossing over,** i.e., there was physical exchange of parts of non-sister chromatids of homologous chromosomes following synapsis at meiosis.

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.

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. Over involves breaking and rejoining of chromosomes in the synaptonemal complex. During zygotene and pachytene stages of the prophase 1 of meiosis, pairing takes place in such a way that similar parts of chromosomes like 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 of places where homologues are held together and exchange bits of chromatids are known as **chiasmata.** The number of chiasmata in a chromosome depends on its length.



The events at Crossing over : A. Leptotene, B. Zygotene, C. Pachytene, D. Chiasma and Crossing Over, E. Diplotene, F. Segregation of chromosome G. End of segregation of chromosome, H. a and d non crossed, b and c crossed gamete

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 recognized depending on the number of chiasmata.

- **Single crossing over,** In single crossing over, there is only one chiasmata where chromatids of homologous chromosome contact. The chromosomes break only at one

point along their entire length. Bivalents having a single chiasmata appear as open crosses.

- **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.
- **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.

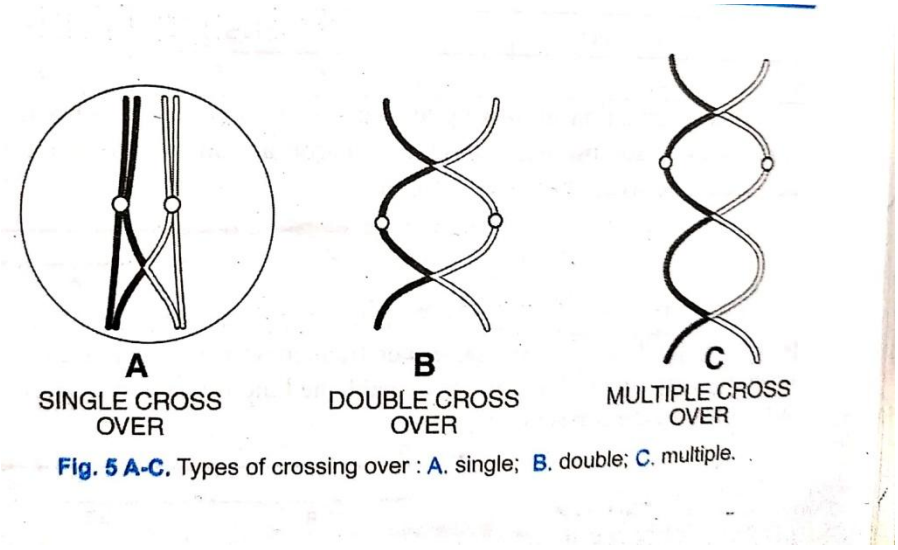


Fig. 5 A-C. Types of crossing over : A. single; B. double; C. multiple.

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 plat 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.

CHROMOSOMAL MAPPING

The representation in figure of relative position of genes on the chromosome is known as **chromosome map** and the process of identifying gene loci is called **mapping.** The chromosome is based on two important assumptions :

- That genes are arranged on a chromosome in a **linear** fashion.
- The percentage of crossing over (recombination frequencies) between the two genes is an index of their distance apart. The relationship between the cross over frequency and the distance between loci was first suggested in 1913 by A. H. Sturtevant. Thus, the chromosome map is a condensed graphic representation of relative distances between the linked genes, expressed in percentage of recombinations among the gene sin one

linkage group. **Distances between genes can be expressed in map units, where one map unit is defined as 1 per cent recombination.**

SEX DETERMINATION

Sex

Sex in animals can be defined as the hereditary difference between male and female individuals of the same species. It behaves as a Mendelian factor. Usually, genes determining sex are located on a special pair of chromosomes. These are called **Sex chromosomes.** Therefore, chromosomes of an individual are basically of two types.

- Autosomes : These carry gene for body characters (somatic characters) and general physiology. These are represented by A.
- Sex Chromosomes or **Allosomes** : These carry genes for determining the sex. These are also called **Idiochromosomes** and are represented by X and Y or Z and W chromosomes.

DISCOVERY OF SEX CHROMOSOMES

Sex chromosomes and Chromosomal basis of sex dertermination was established by German biologist, **Henking** (1891) while studying spermatogenesis in insects. He observed a dark coloured body in the nuclei of 50 per cent sperm and named it **X-body.**

X-body of Henking was later named as X chromosome. It was later discovered by Mc Clung (1902) that in insects, the sex determination mechanism is XX-XO type. The female insects have 2X (XX) chromosomes along with autosomes and male insects have XO (only one X) chromosome.

CHROMOSOMAL MECHANISMS OF SEX DETERMINATION

In majority of animals, one pair of sex chromosomes (X and Y or Z and W) are associated with sex determination. These are called allosomes or idiochromosomes or sex chromosomes. One sex is heterogametic, i.e., produces two types of gametes in equal proportion and other sex is homogametic, i.e., produces only one type of gametes. It may be:

- Male Heterogamy : Male produce two types of sperm in equal proportion. The females are homogametic and produce only one type of eggs as in mammals.
- Female Heterogamy : Females produce two types of eggs. Males produce only one type of sperm and are homogametic as in birds.

(a) **XX-XY type of Sex Determination**

In mammals including Man and *Drosophila*, sex is determined by X and Y chromosomes. X-chromosomes possess female dertermining genes and Y-chromosome carries male determining genes.

- Female is homogametic sex. It has chromosomes AA+XX. All the eggs produced are only of one type with chromosomes A+X.
- Male is heterogametic sex. It has chromosomes AA+XY. It produces two types of sperm: with A+X and A+Y chromosomes. The condition is called male digamety. The sperms with X-chromosome are known as gynosperms (female determining) and with Y-chromosome as androsperms (male determining).

  The sex is determined at the time fo fertilization by the type of sperm fusing with the ovum. The ovum with X-chromosome when fertilised by a sperm with X-chromosome, develops into a male XY-chromosomes.

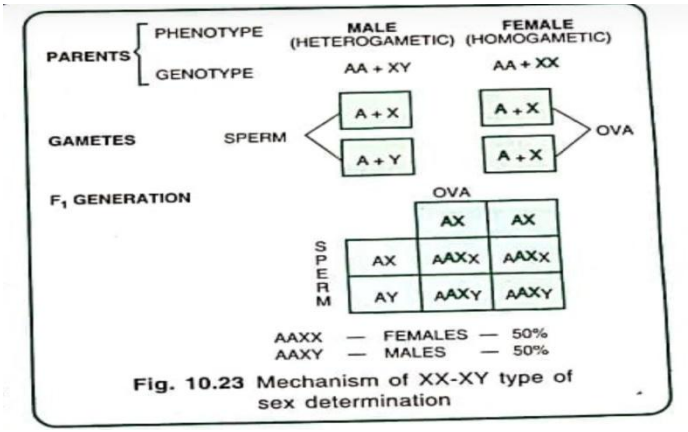


Fig. 10.23 Mechanism of XX-XY type of sex determination

(b) **ZZ-ZW Type of Sex Determination**

It is found in butterflies, moths, fishes, reptiles and some birds where :

- Male is homogametic. It has homomorphic sex chromosomes ZZ. Its chromosomal composition is AA+ZZ. It produces sperm only of one type A+Z.
- Female is heterogametic. Its sex chromosomes are heteromorphic,

Represented by ZW. Therefore, female has chromosomes AA+ZW. It produces eggs of two types with A+Z and A+W chromosome.

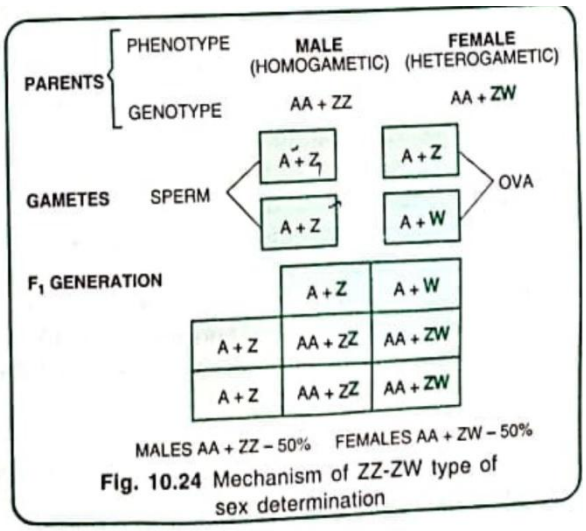


Fig. 10.24 Mechanism of ZZ-ZW type of sex determination

(c) Haplo-diploid Mechanism of Sex Determination in Honeybee:

In honeybees, wasps, ants and sawflies, females (the queens and workers) are diploid. They develop from fertilised eggs having 2n number of chromosomes. The males (drones) are haploid and have only n number of chromosomes. These develop from unfertilized haploid eggs by **parthenogenesis**. This is called haplodiploid mechanism of sex determination, wherein sex is determined by the number of sets of chromosomes and not by the number of X and Y chromosomes. Males produce haploid sperms by mitosis. The eggs that are fertilised restore two sets of chromosomes and develop into females. The unfertilized eggs develop into males. In honeybee, a male has 16 chromosomes while a female has 32 chromosomes.

Haplo-diploid mechanism of sex determination exhibits the following peculiarities:

- Sperms are produced by mitosis.
- The male has no father but has grandfather.
- A male cannot have sons but has grandsons.
- If a queen bee mates with only one drone, any two of her daughters will have, on an average, ¾th of their genes similar.

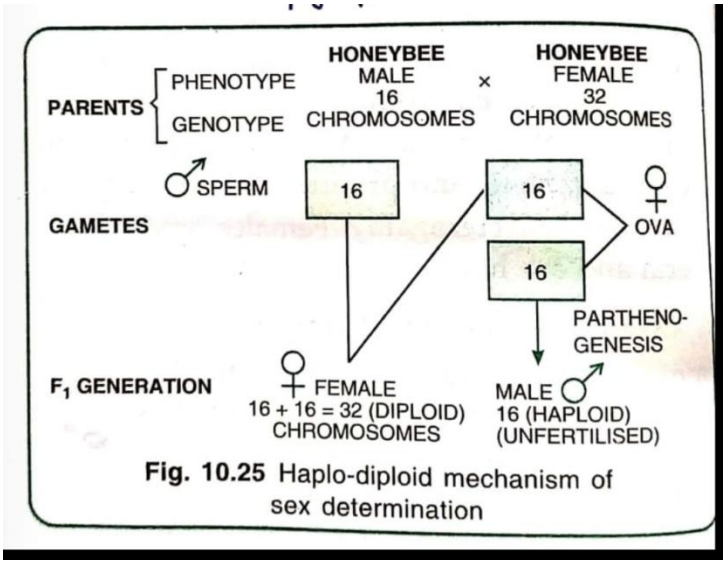


Fig. 10.25 Haplo-diploid mechanism of sex determination

SEX DETERMINATION IN MAN

The total number of chromosomes in man is 46 chromosomes or 23 pairs. Out of these 23 pairs, 22 pairs or **44 chromosomes are autosomes** and **one pair is sex chromosomes** or **allosomes**. Autosomes are similar in both males and females or men and women. The two sex chromosomes are similar in women and are represented by XX. In men, they are different; one of them is similar to X-chromosome while the other one is called the Y-chromosome. Therefore, chromosomes in man are 44+XY and in women are 44+XX.

Thus, men are heterogametic and produce two types of sperm, 50 per cent with 22+X and 50 per cent with 22+Y chromosomes. Women (females) are homogametic. They produce only one type of ova having 22+X chromosomes.

- In case the ovum is fertilised with a sperm carrying X-chromosome, the zygote with 44+XX develops into a female child.
- When ovum is fertilised with a sperm carrying Y-chromosome, the zygote with 44+XYchromosomes develops into a male child. Thus, this is clear that :
  - The sex of child is determined at the time of fertilization.
  - The sex is determined by the genetic make up of the sperm.
  - There is always 50 per cent probability of a child being born to be male or female.

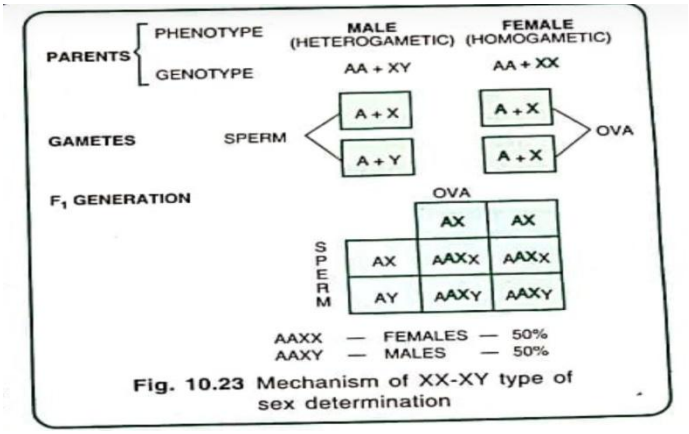


Fig. 10.23 Mechanism of XX-XY type of sex determination

Shaista Ahmed