

MUTATION

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INTRODUCTION

- The term mutation was coined by Hugo De Vries in 1900.
- Sudden heritable changes in the genetic material are called mutation.
- Mutation is not always bad most DNA changes fall in the large area genome that sit between and usually they have no effect.

TYPES OF MUTATION

(A) On the basis of origin-

1. Spontaneous mutation
2. Induced mutation

(B) chromosomal mutation-

1. Change in the numbers of chromosome- Ploidy

Two types-

1. **Euploidy**- it include addition or loss of one or more than one set chromosome in diploid organisms.

1. monoploidy

2. polyploidy

2. **Aneuploidy**- it is the changes in one or more pair of chromosomal set.

1. Hypoploidy- reduction in the normal no. of chromosome.

1. monosomy

2. nullisomy

2. Hyperploidy- addition of one or more set of chromosome

1. Trisomy

2. Tetrasomy

2. Changes in the structures of chromosome-
it include changes are-

1. The no. of gene in chromosome

1. Deletion

2. Addition

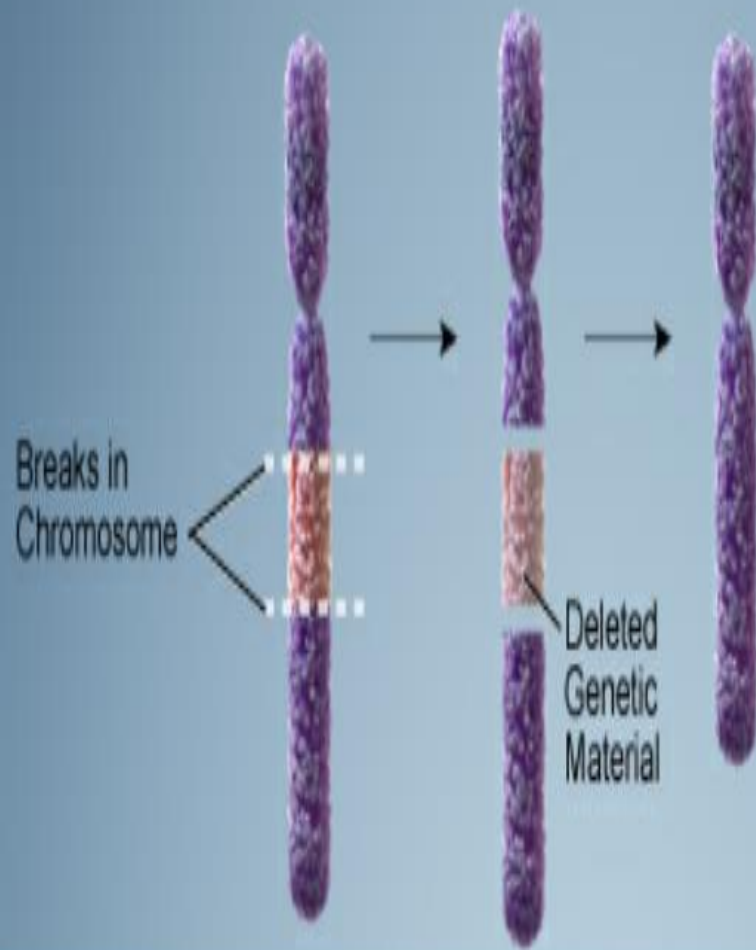
3. Duplication

2. Arrangement of gene in chromosome-

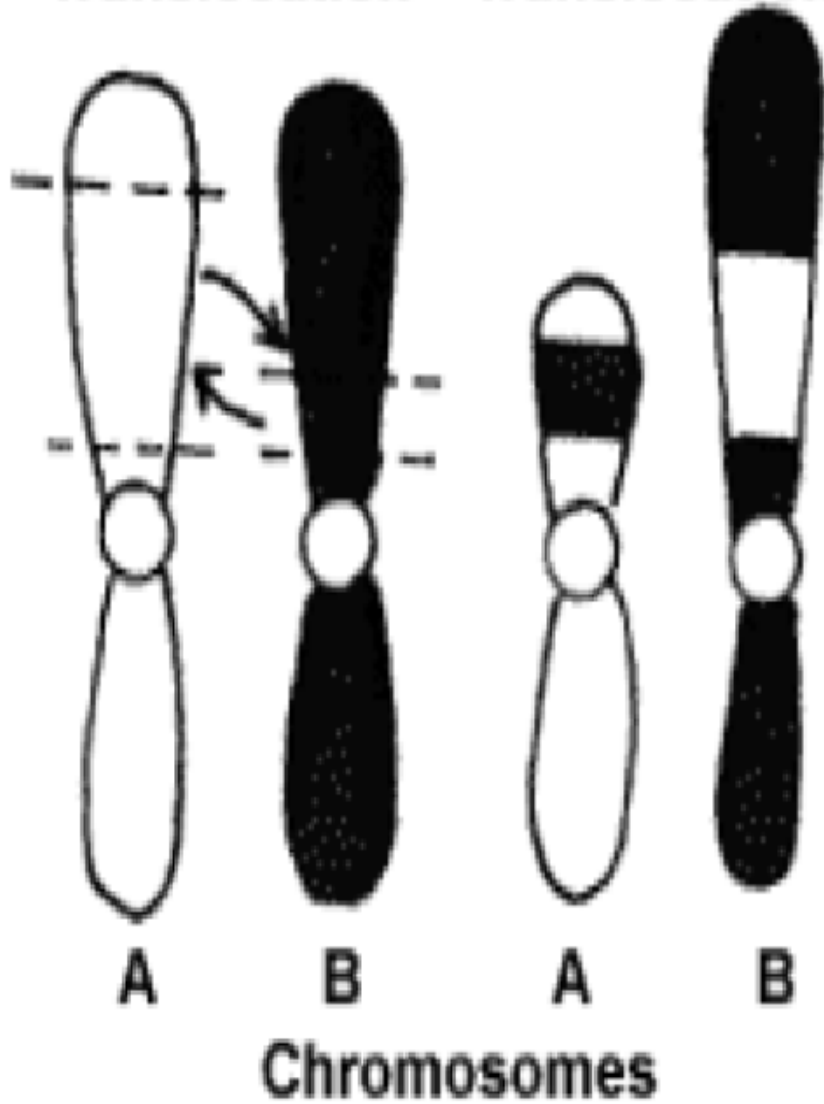
1. Inversion

2. Translocation

Deletion

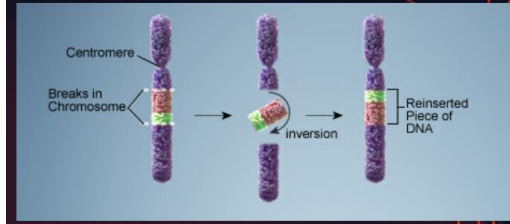


Before Translocation After Translocation



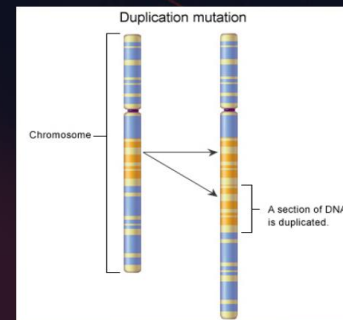
backwards

- Segment **reattaches**



Duplication

- Occurs when a gene **sequence is repeated**



3. **Point mutation-** Deletion ,duplication, insertion of nucleotide cause heritable alternation in the DNA sequence.

- Silence **mutation** : no change in the overall protein
- **Missense mutation** : change in one amino acid
- **Non-sense mutation**: a stop codon

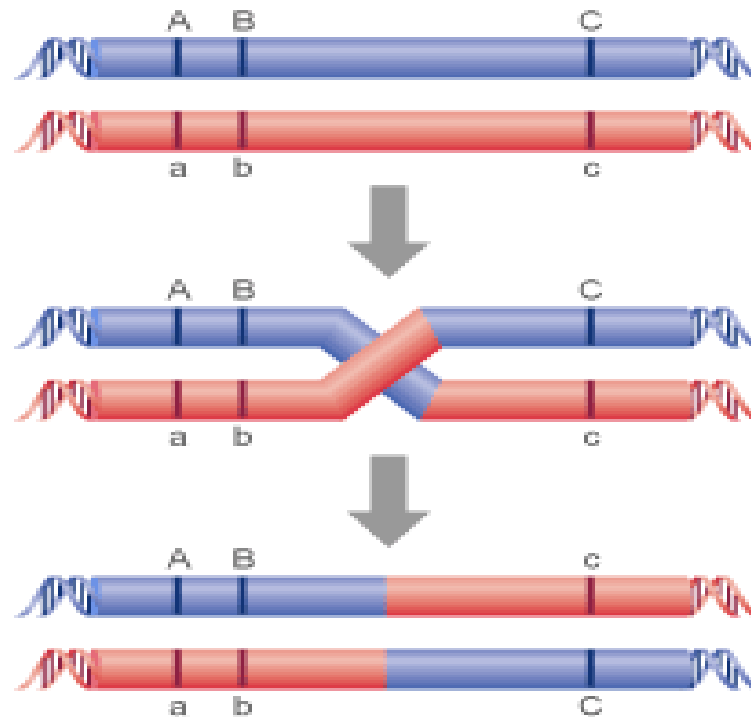
Effect of point mutation-

Mutation may have no effect if base substitution in gene does not alter the amino acid sequence.

- Mutation may result in the change of nucleotide sequence leading to the alteration of amino acid sequence in the poly peptide chain. And is called as missense mutation.
- A change in nucleotide sequence may sometime produce a stop codon within a gene sequence. Due to that termination occur this type of mutation is called non-sense mutation.

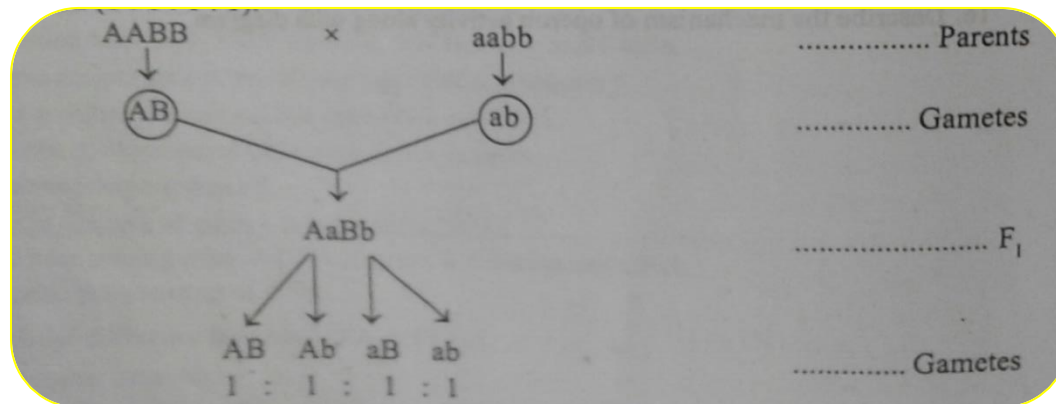
- If mutation involves the insertion or deletion of nucleotides which are not a multiple of three it will alter the reading of the amino acid sequence and can be changed drastically, this is called frameshift mutation.

Linkage in genetics



Introduction

- Mendel's law of segregation states that when two or more than two factor are considered together, these factors show independent and random assortment during distribution of gametes.
- During meiosis in F₁ generation progeny (AaBb) maternal and paternal factors are independently assorted into gametes.
- Four types of gametes (AB , Ab , aB & ab) will be produced in equal proportion (1:1:1:1).



- Dihybrid F₂ ratio of 9:3:3:1 and test cross ratio (1:1:1:1) are actually based on the law of independent assortment.
- This assortment of characters is based on independent assortment of non-homologous chromosomes; these will be located on separated non-homologous chromosomes.

Conti

- “Genes present on same chromosomes and inherited together are called as **linked gene** and the process is called as **linkage**. “
- The principle of linkage was 1st discovered by **Bateson** and **Punnet** in 1906 in **sweet pea** (*Lathyrus odoratus*) .
- Total number of linkage group in an organism is equal to the number of homologous chromosomes (pair of chromosomes) .
- Linkage group – Human beings – 23
Drosophila - 04
Sweet pea – 07

Definition

- According to Morgan (1910) he define linkage as -

“The tendency of gene present on the same chromosome , to remain in the original combination and enter together same gamete”.

Theory of linkage

1. Coupling and repulsion theory of linkage
2. Chromosomal theory of linkage

1. Coupling and repulsion theory of linkage – this theory was proposed by **W. Bateson & R. C. Punnet** (1906), after the study of pea plant

☐ He take two varieties of pea plant -

I. Plant with purple flower 'P' and long pollen (L).

II. Plant with red flower 'p' and round pollen(l).

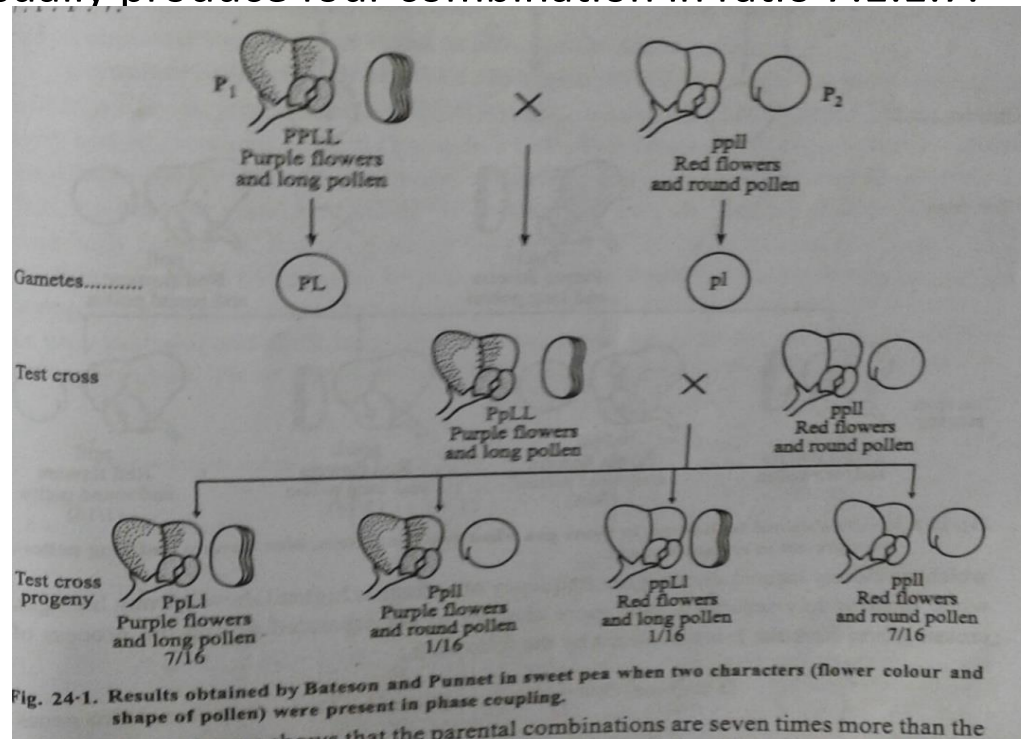
Here purple flower 'P' & long pollen 'L' are dominant and red flower 'p' & round pollen 'l' are recessive.

When a cross is made b/w purple flower long pollen "PPLL" and red flower round pollen "ppll".

F1 gen.... Only "PpLl" produce..

Conti....

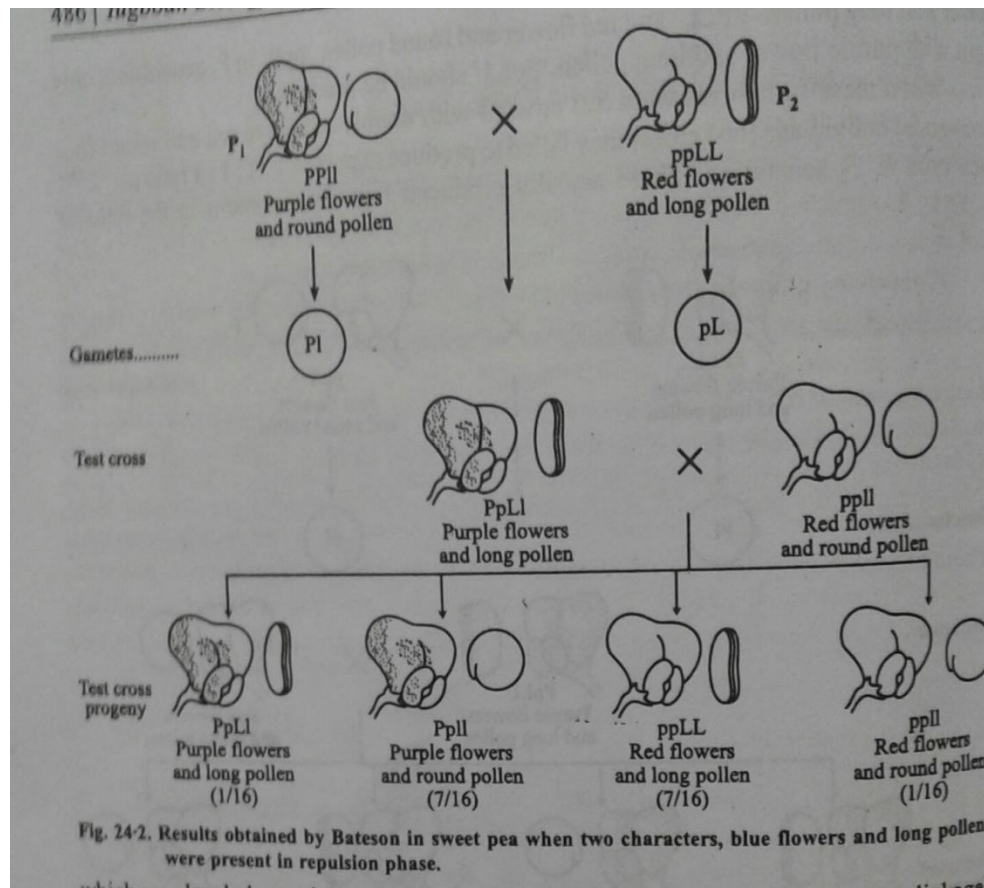
- When f1 hybrid were test cross with double recessive red & round (homozygous) they failed to produce expected ratio 1:1:1:1ratio
- These usually produce four combination in ratio 7:1:1:7.



- Above test cross show the parental combination are seven time more than the non parental combination.
- Bateson & punnet suggest “ the alleles coming from the same parent tend to enter same gamet and to be inherited together . This is called **genetic coupling**.

Cont...

- In same way “ the genes or alleles coming from two different parents tend to enter different gametes and to be inherited independently”. This is called genetic repulsion.



Conti...

2. Chromosomal theory propose by Morgan and Castle in 1910

According to this theory –

1. Gene which is locate on the same chromosome show linkage.
2. The linked genes are locate same chromosome in a linear sequence with a definite and constant order in its arrangement.
3. The strength of linkage depends upon the distance b/w gene .

$$D \propto 1/S \text{ or } S \propto 1/D$$

Here D = distance b/t gene , S = Strength of linkage

$$D = K. 1/S$$

K = constant known as linkage

4. Cis – arrangement & trans arrangement of genes

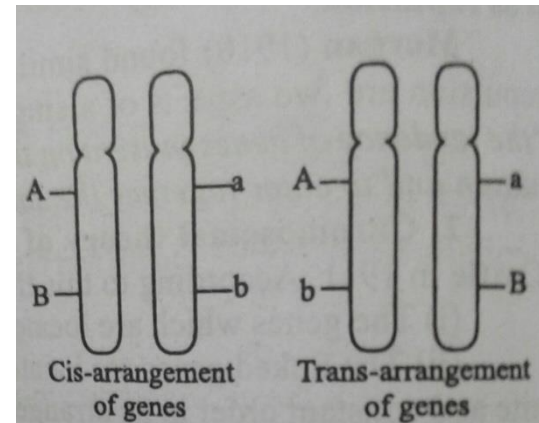


Fig. 24-3. Cis-trans-arrangement of linked genes.

Types of linkage

1. Complete linkage
 2. Incomplete linkage
1. **Complete linkage** – In a complete linkage two or more character are inherited together and these character are appear in two or more generations. Ex. *Drosophila* (male)

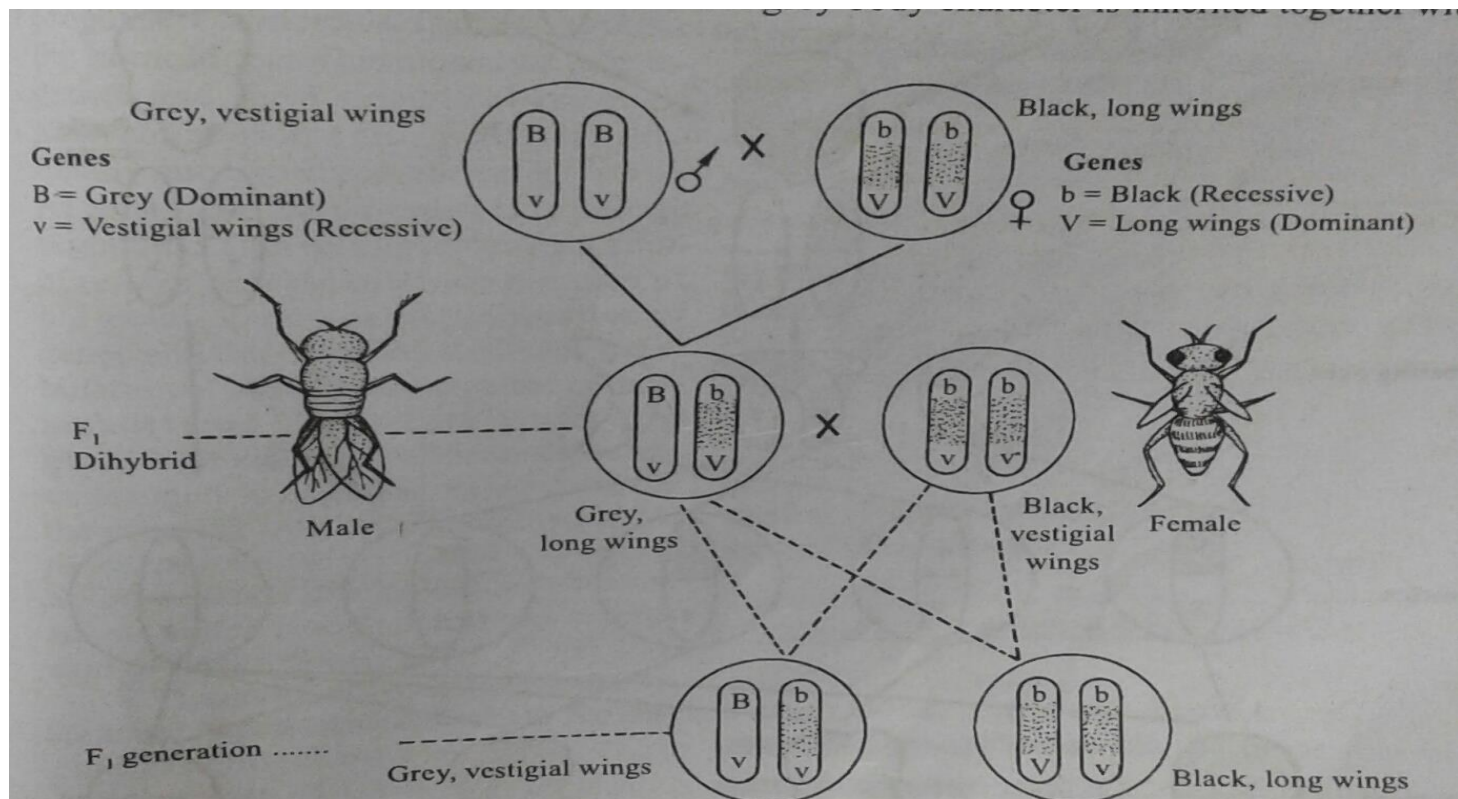


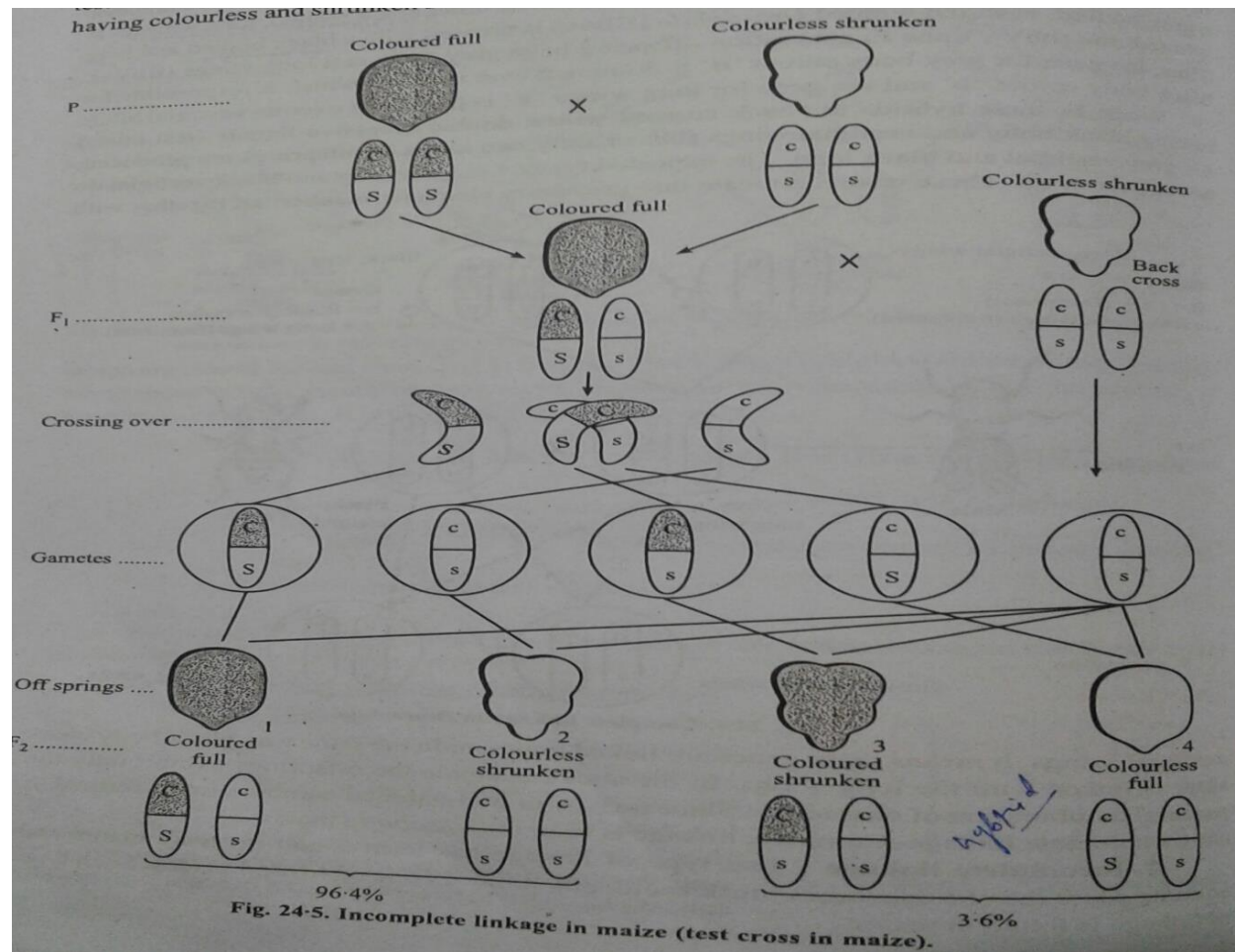
Fig. 24-4. Complete linkage in *Drosophila*.

Cont....

2. Incomplete linkage – this type of linkage found many plant and animals.

Here chromosomes break accidentally either in one or several places. Ex.

maize.



Important of linkage

1. Linkage help to know the characters of parents.
2. It helps to know the probability of origin of new characters in off springs.
3. It helps in the control of many characters of organisms by controlling inheritance of linkage groups.

Thank you for your attention

