



CSIR-NET

Council of Scientific & Industrial Research

LIFE SCIENCE

VOLUME – 6

**DEVELOPMENT BIOLOGY, PLANT
& ANIMAL PHYSIOLOGY**



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GENETICS (INHERITANCE BIOLOGY)

Gene interaction :-
 → when two or more different genes influence the outcome of single trait, this is known as a gene interaction.
Discovery - William Bateson & Reginald Punnett in 1906.
 They discovered an unexpected gene interaction when they studied crosses involving the sweet pea, *Lathyrus odoratus*.
 ex. P generation white flowered plant × white flowered plant
 CCPP ccPP

All Purple
 $CcPp$ → F₁ gen
 The F₁ hybrid plants are allowed to self fertilize
 $CcPp \times CcPp$
 F₂ gen → Purple : White
 9 : 7

9 : 3 : 3 : 1
 ↓ convert
 9 : 7

★ Epistasis :-

- Epistasis → Greek for standing upon
 A Type of gene interaction when one gene masks or modifies the expression of another gene at distinct locus.
 → Any gene that masks the expression of another non-allelic gene is epistatic to that gene.
 → The gene suppressed is hypostatic.
 → Epistasis is diff from dominance. Epistasis is the interaction b/w diff genes (non-alleles). Dominance is the interaction b/w diff alleles of the same gene that is intrallelic.
 ① Dominant Epistasis :-
 Dominant allele of one gene masks the effects of either allele of the second gene. It is termed as dominant epistasis

⊛ ex. P₁ AA bb (Purple) P₂ aa BB (Red)
 ↓
 F₁ Aa Bb (Purple)
 ↓
 F₂ 12 Purple : 3 Red : 1 white

★ Recessive epistasis:-

In the case of recessive epistasis, in a pair of non-allelic gene one produces its phenotypic effect independently in a dominant state, but another cannot produce a phenotypic effect independent.

Ratio $9:3:3:1$ become $9:3:4$

Purple : 3 Red : 4 White

★ Duplicate recessive epistasis:- (complementary gene interaction)

If two non-allelic gene are involve in a specific pathway & final product from both are required for expression, then one Homozygous recessive allele in either allelic pair would result in the mutant phenotype

$9:3:3:1 \rightarrow 9:7$

★ Duplicate dominant interaction:-

$9:3:3:1 \rightarrow 15:1$

★ Pleiotropy:-

→ Most of the biochemical pathways in the living organism are interconnected.

→ The term pleiotropy refers to the effect of a single gene on more than one character/trait
 Some time one trait will be very evident & others will be less evident.

- Several characteristics may differ b/w individuals belonging to the same spp. These differences are termed variations.
- The mechanism of transmission of characters, resemblances as well as differences, from the parental generation to the offspring is heredity. (आनुवंशिकता)
- ⇒ The study of heredity, variations & envt. factor known as genetics.

Greek word genno = give birth

Genetics → (3 types)

- 1) Classical genetics → Mendel's principle, sex determination
- 2) Molecular genetics → Genetic material
- 3) Evolutionary genetics → Pop'n genetics

Gregor Johann Mendel (1822-1884)

Rediscovered the Mendel's law -

- ① Hugo de Vries (Holland)
- ② Carl Correns (Germany)
- ③ Erich Tschermak (Austria)

⇒ He discovered that individual traits are inherited as discrete factors which retain their physical identity in a hybrid.

⇒ Later, these factors came to be known as gene.

⇒ A gene is defined as a unit of heredity.

* Allele :-

Each gene may exist in alternative forms known as allele.

* Homologous - chromosome that carry the same set of genes in the same seq., although they may not necessarily carry identical alleles of each gene.

★ Rh Blood group system :-

- Rh antigen & Factor fut on the plasma membrane.
- Macacus rhesus → Monkey
- The Rh antigens are located on two Rhesus protein RHD and RHCE
- RHD and RHCE are expressed only in RBCs.
- More than 170 alleles of RHD gene have been found.
RHD protein carries for D antigen
RHCE protein carries for CE antigens in various combination (Ce₁, Ce₂, cE or CE)
- The classification of Rh-positive and Rh-negative individuals is determined by the presence or absence of the D antigen on the surface of RBCs.
- D antigen fut on a person's RBC, the person is Rh⁺
if absent Rh⁻
Erythroblastosis Fetalis (or hemolytic disease)

★ Lethal allele :-

The alleles created by mutation in these gene are called lethal alleles. Lethal alleles may be recessive or dominant.

- 4 categories
- ① Early onset → early death of an organism
 - ② Late onset → delayed effect
 - ③ Conditional → kill organism under certain env. condition
 - ④ Semilethal → kill only some individuals but not all.

★ Penetrance :-

The percentage of individuals that shows a particular phenotype among those capable of showing it, is known as penetrance. ex. - Polydactyly in human (More than 5 finger)

- A particular gene may produce diff. degree of expression in diff. individuals. This is known as expressivity.

★ Phenocopy :-

A phenotype that is not genetically controlled but looks like a genetically controlled one is c/d phenocopy. It is an environmentally induced phenotype that resembles the phenotype determined by the genotype.
ex. → phenocopy of vit-D resistant rickets.

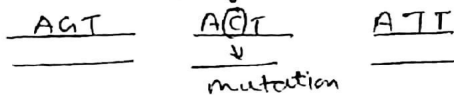
LAW OF GENETICS

- **Allele**: - Alternate form of the gene
- Different nose shape - Polymorphism
- Eye colour is a gene

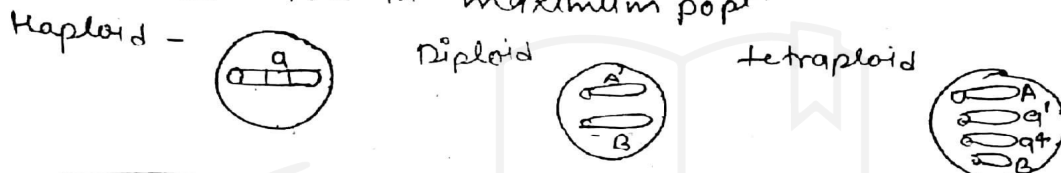
Diff nose shape

- but diff. Eye colour like blue, black, white eye is Allele
- Mutation create Allele.

Mutation $\leq 1\%$ of population



- Local population in a particular area known as wild type
- gt is +nt in maximum popⁿ



→ Alleles present on chromosome
different allele representation -
eye colour - (Black eye - b^+ , B , B^1 , B^A)

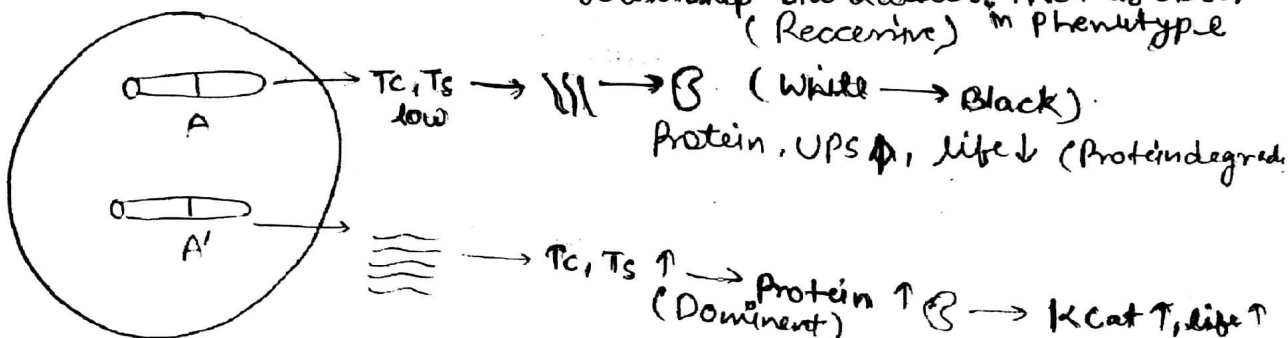
- * Diploid - two alleles of a gene
- * Haploid - Single alleles of a gene

- Crease is not seen → The wrinkled phenotype of seed is due to the absence of Amylopectin

* Law of Genetics

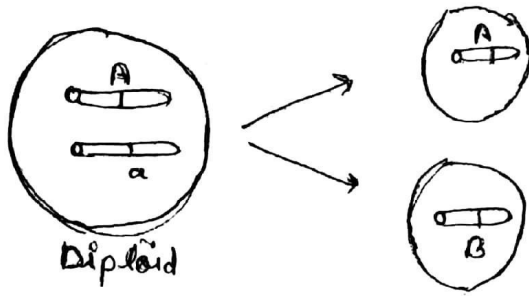
(1) Law of Dominance:

→ Dominant & Recessive are not the properties of alleles or gene. It is a relationship b/w 2 alleles that is observed (Recessive) in phenotype



- Blue colour is show in ~~off~~ ^{off} generation.

② Law of Segregation :- (Law of purity of gamete)



① Law of Dominance :-

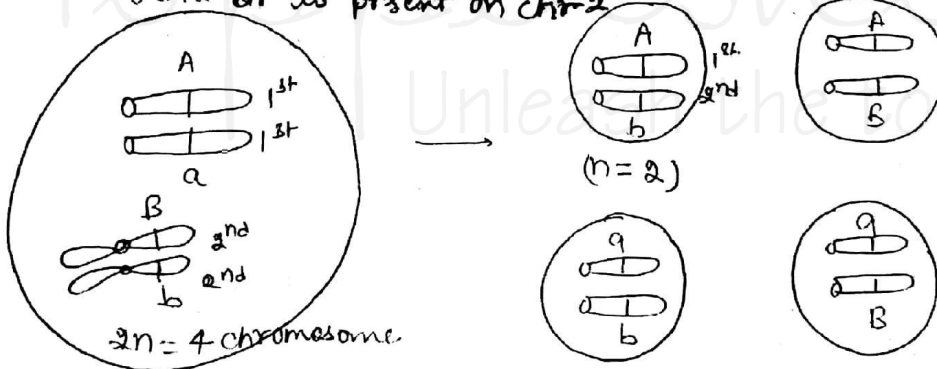
→ One allele of a gene cancel the effect of other allele of same gene

② Law of Segregation :-

Gametes are Haploid

③ Law of Independent Assortment :-

Gene B is present on chr-1
Gene A is present on chr-2



Phenotype = 1:1:1:1

→ Allele can combined with any other allele of other gene
two gene are not on diff chromosome

→ One allele of a gene can combined with any other allele of other gene. This is known as law of Independent Assortment.

→ When 2 genes are not on diff chromosome their gametic ratio is 1:1:1:1

LINKAGE

Linkage :-

- two or more genes reside on the same chromosome, they are said to be linked and their transmission pattern is called linkage.
- They may be linked together on the autosomes or on the sex chromosome. Gene not on diff. non-homologous chroma are called unlinked gene.
- linked gene (genes on the same chromosome), however tends to stay together during the form of gametes.
 linked gene - on same chromosome
 unlinked gene - (on different chromosome)

* Limits of recombination :-

If two gene loci are, so far, apart in the chromosome that the probability of a chiasma forming b/w them is 100%. then 50% of the gametes will be the parental type (non-crossover) & 50% recombinant (crossover) type. when such dihybrid individuals are testcrossed, they are expected to produce progeny in a 1:1:1:1 ratio as would be expected for genes on diff chromosome. Recombination b/w two linked genes cannot exceed 50% even when multiple crossovers occurs b/w them.

* Morgan's work :-

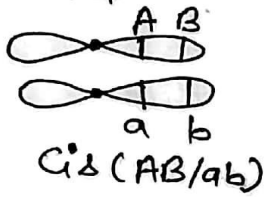
The effects of linkage were first evident in the result of a dihybrid cross in sweet pea by Bateson & Punnett in 1906.

- Morgan who first discovered the phenomenon of linkage. Morgan postulated that separation of linked gene occurs due to form of chiasmata which represent points of genetic exchange. He used the term crossing over to describe the physical exchange leading to recombination.
- Morgan also proposed that two genes located relatively close to each other along a chromosome are less likely to have a chiasma b/w them as compared to the two genes which are farther apart on the chromosome.
- Drosophila is unusual in that crossing over (hence recombination) does not occur in ♂ male.

The results is that alleles located in a particular chromosomes show complete linkage in drosophila males. Crossing over is also completely suppressed in female silkworms.

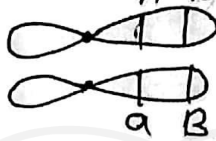
★ Cis and trans configuration :-

Doubly heterozygous genotypes can be in two diff. configurations. When two linked genes on each chromosome are of the same type (i.e. both dominant, AB, or both recessive ab) the arrangement is called the coupling or cis. Alternatively, when two genes on each chromosome are of diff. type (i.e. one dominant and one recessive allele, aB or Ab) the arrangement is the repulsion or trans.



↓

AB = Parental combination
Ab = Recombinant



Trans (Ab/aB)

Ab = Parental combination
AB = Recombinant.

★ Genetic mapping :-

The linkage of the gene in a chromosome can be represented in the form of a genetic map or linkage map or chromosomal map.

Discovery - Alfred Sturtevant

→ He used the frequency of crossing over (i.e. recombination frequency) b/w two genes to prepare the first genetic map of X-chromosome of *Drosophila*.

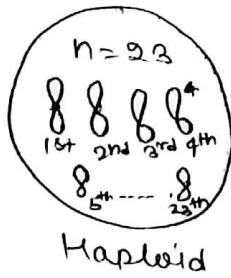
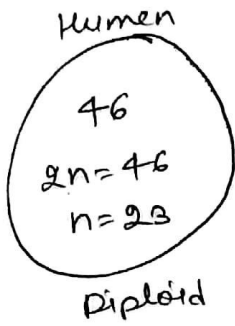
1% crossing over or recombination is equivalent to 1 map unit or 1 cM.

By analyzing percent recombination among the progeny of parents that are heterozygous for a no. of linked genes, a genetic map that places the gene in a linear array can be constructed & o.

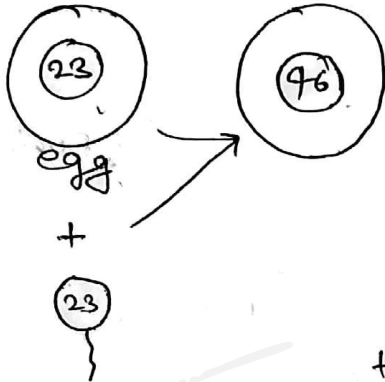
$$\text{Map distance} = \frac{\text{No. of Recombinant offspring}}{\text{Total no. of offspring}} \times 100$$

Unit = Map Unit or Centimorgan (cM)

$$1\text{cM} = 1 \times 10^8 \text{ bp}$$

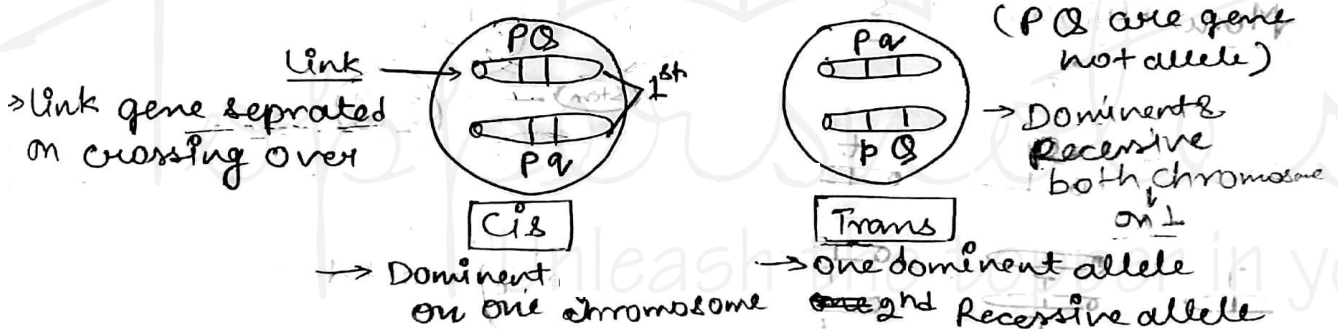


25000 gene
↓
23 Chromosome
↓
1 chr. - 1000 gene

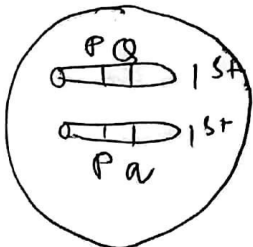


In one sperm & one egg 23 chromosome is present

Linked gene :- (Pair of the gene) when two or more gene but on same chromosome they are known as linked gene



The linked gene are but on same chromosome
→ Chromosomal map = T. H Morgan

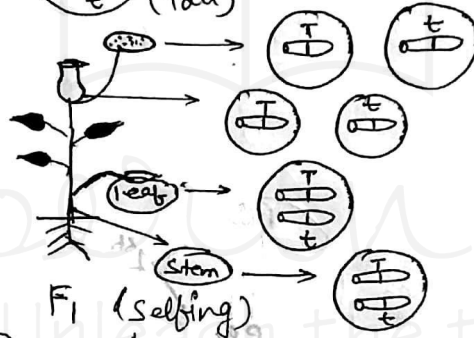
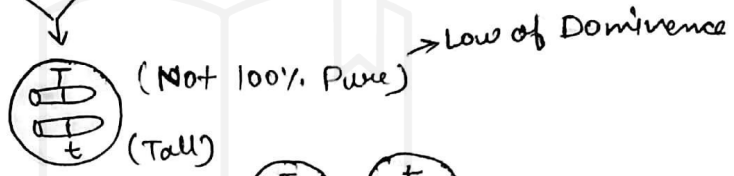
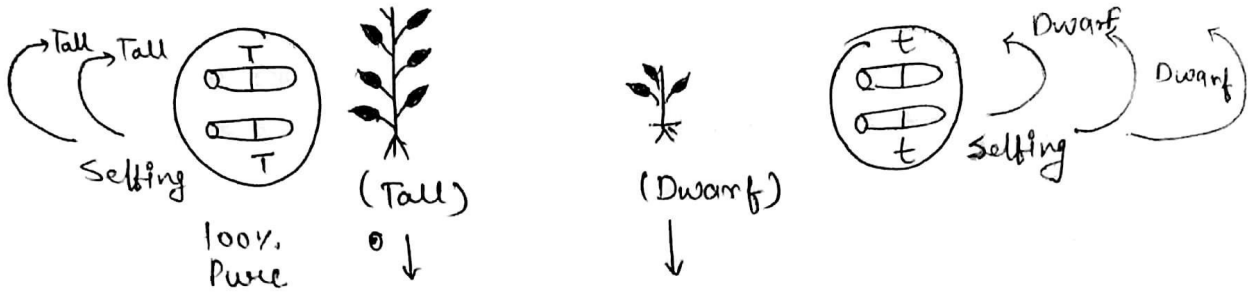


Phenotype = 1 : 0 : 0 : 1



Genetic ratio deviates from 1:1:1:1 and it is 1:0:0:1
linkage group = Haploid no. of chromosome are c/d as linked group
Haploid no. of chromosome & sex chromosome
Female = 22+X Male = 22+X+Y

MONOHYBRID AND DIHYBRID CROSS



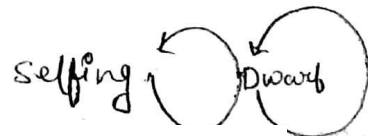
Mono Hybrid cross
Punnett square

| | | | | | | | | | | |
|---|---|---|------------|---|------------|--|---|------------|---|-------------|
| ♂ | T | t | | | | | | | | |
| ♀ | <table border="1"> <tr> <td>T</td> <td>TT Tall</td> </tr> <tr> <td>t</td> <td>Tt Tall</td> </tr> </table> | T | TT Tall | t | Tt Tall | <table border="1"> <tr> <td>T</td> <td>Tt Tall</td> </tr> <tr> <td>t</td> <td>tt Small</td> </tr> </table> | T | Tt Tall | t | tt Small |
| T | TT Tall | | | | | | | | | |
| t | Tt Tall | | | | | | | | | |
| T | Tt Tall | | | | | | | | | |
| t | tt Small | | | | | | | | | |

F₂ gen all dwarf are pure
Phenotype
3:1
Tall: Small Dwarf
Genotype = 1:2:1
1/4, 2/4, 1/4

tall, tall, tall, Dwarf
Tt = 3:1
50% pure

| | |
|------------|-------------|
| TT Tall | Tt Tall |
| Tt Tall | tt Small |

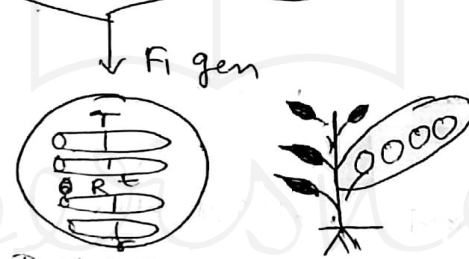
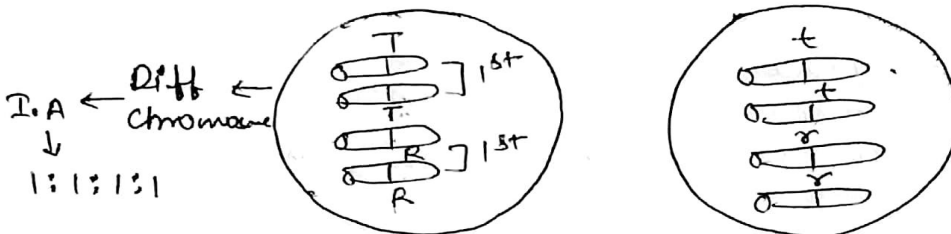




Dihybrid cross



Dwarf & wrinkled



Dihybrid cross →

Tall & Round

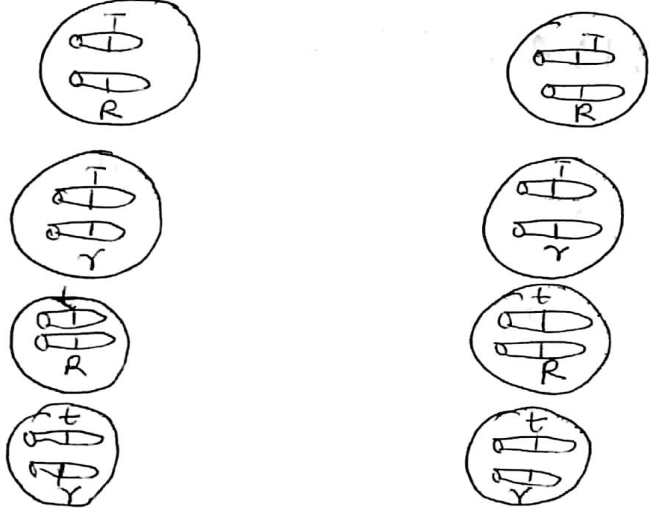
Tall & Round

Phenotype :-

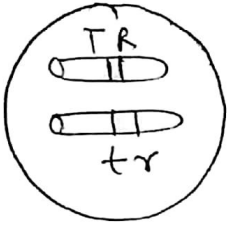
Tall & Round : Tall & wrinkled : Dwarf & Round : Dwarf & wrinkled
9 : 3 : 3 : 1

Gamete Male

Female



Linked gene



→ linked gene में both gene साथ में रहते हैं जिससे Tall & Round, Small & Wrinkled के साथ नहीं आ सकता।

Blue eyes
&
Normal Nose

Black eye
&
Pointed Nose

Gametes -

① Blue eyes
&
Normal Nose

② Blue eyes
&
Pointed Nose

③ Black eyes
&
Normal Nose

Black eyes
&
Pointed Nose

Punnett square

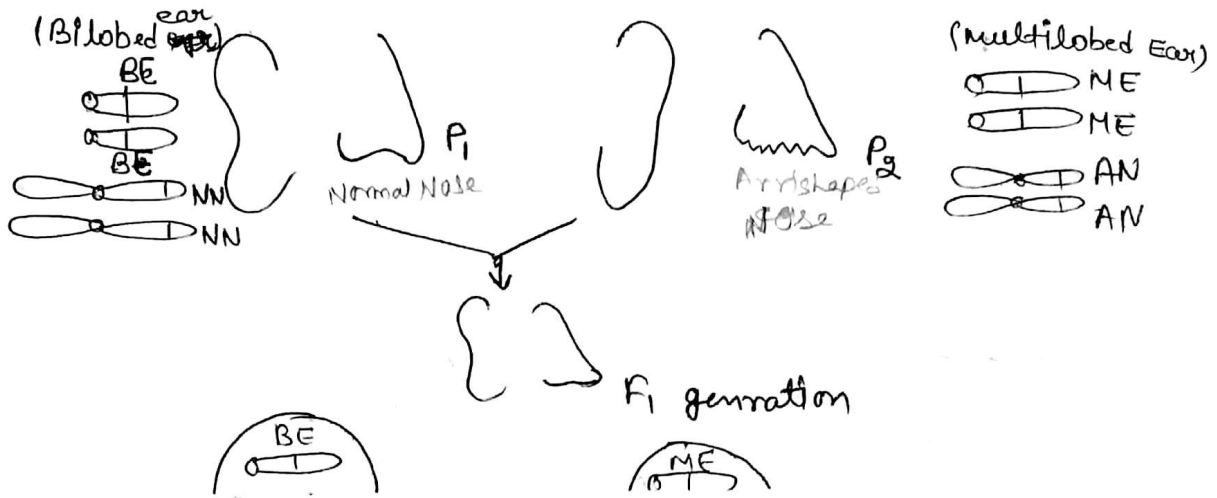
| ♀ ♂ | TR | T γ | tR | t γ |
|------------|------------------------------|---|-------------------------------|--|
| TR | TTRR Tall & Round | TTR γ Tall & Round | TtRR Tall & Round | TtR γ Tall & Round |
| T γ | TTR γ Tall & Round | T γ γ Tall & wrinkled | TtR γ Tall & Round | Tt γ γ Tall & wrinkled |
| tR | TtRR Tall & Round | TtR γ Tall & Round | ttRR Small & Round | ttR γ Small & Round |
| t γ | TtR γ Tall & Round | Tt γ γ Tall & wrinkled | ttR γ Small & Round | tt γ γ Small & wrinkled |

Phenotype :- 9:3:3:1

Genotype :- 1:2:2:4:1:2:1:2:1

→ All alleles for any particular gene are found at a specific place on a chromosome called the locus for that gene.

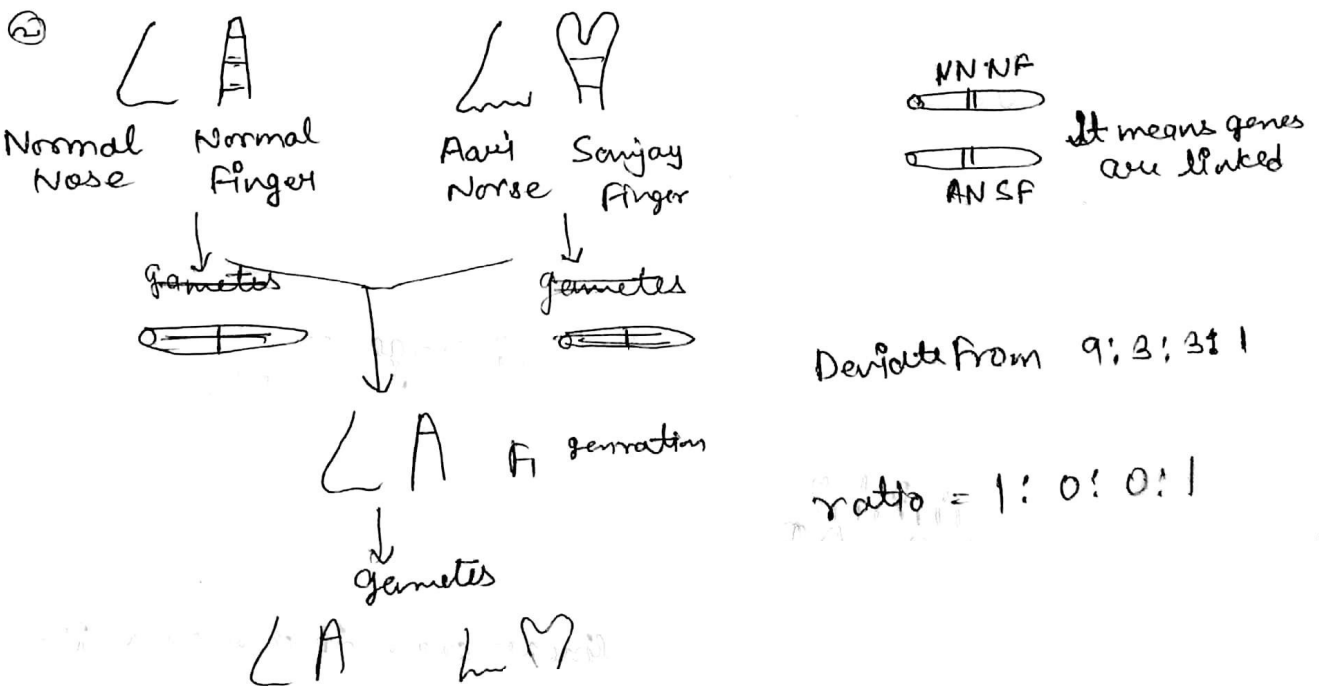
* Test cross - The cross b/w F₁ & its recessive



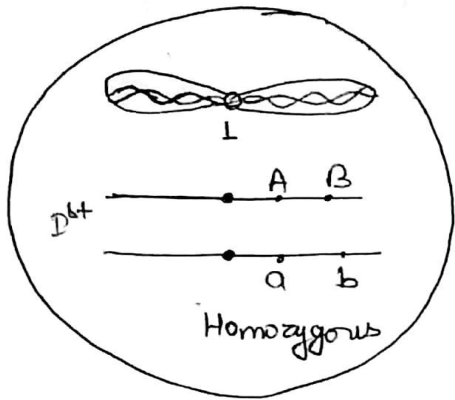
Linkage group :-

→ All the gene that on a particular chromosome form a linkage group. ex-

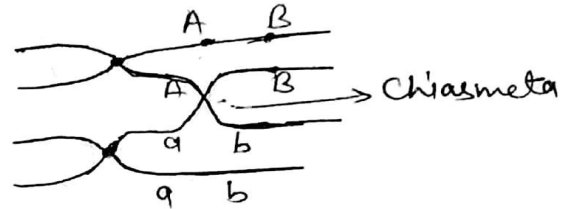
In human - Male = 22 pairs autosome + 1X chro. + 1Y chro.
= 24 linkage group
Female = 22 pair autosome + 2X chromosome
= 23 linkage group



Crossing over means recombination.
Chiasmata = Evidence of crossing over



G₁ phase



→ Normally 1 chiasmata is formed.

4 & 5 vary rate

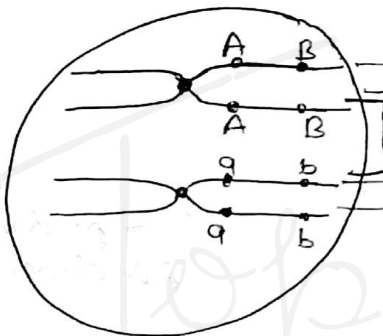
→ Chiasmata no. is variable

like -

A = 1 C = 4

B = 3 D = 2

E = 7 → possible



S phase (meiosis)

Sister chrm.

Non-sister Chromatid

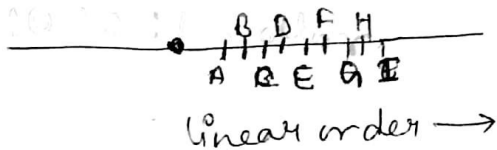
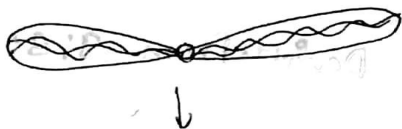
Sister Chromatid

(Replication)

→ crossing over takes place in non-sister chromatid.

→ In one chromosome on one crossing over second crossing is not found. because by crossing over chromosome is broken

→ 1 chromosome = 1 DNA



→ genes are arranged in linear order that means distance b/w diff. genes pair is diff.

$$\text{Linkage} \propto \frac{1}{\text{Distance}}$$

→ जितना Distance उतना होता है।
इसलिए linkage कम होता है।

Linkage frequency -

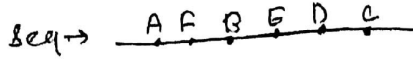
A-B = 90%

A-C = 10%

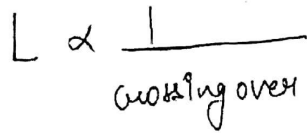
A-D = 20%

A-E = 60%

A-F = 89%



→ F, A के पास

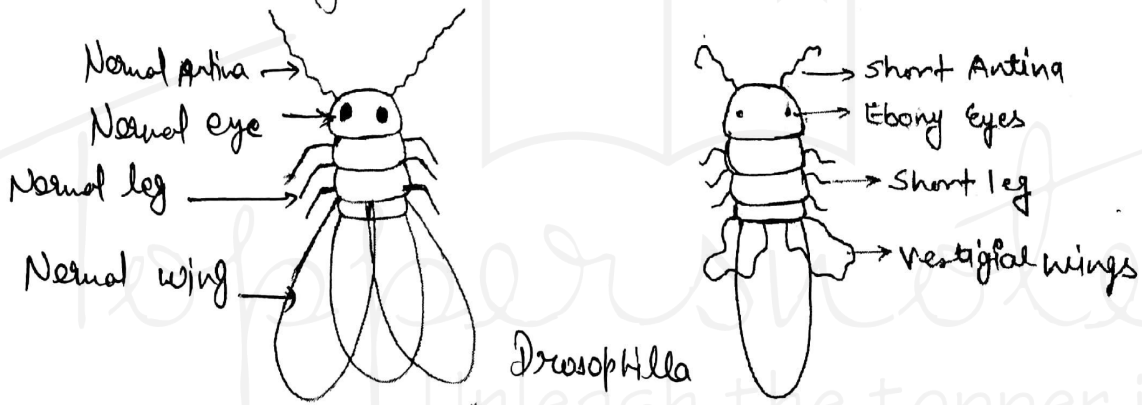


crossing over ↑ → Linkage कम
→ crossing over is a feature of germ cell.

→ की following type 2 identified कर (कैसे) -

① according to F₁ gen. gametes

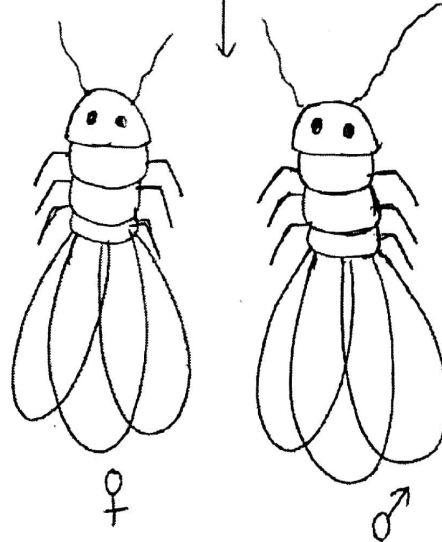
② By selfing



Selfing

9 : 3 : 3 : 1

| | | | |
|-------|-------|-------|-------|
| N.w | N.w | U.w | U.w |
| N.leg | S.leg | N.leg | S.leg |



F₁ generation
(Heterozygous)

(Progeny Male & Female की भी है समान ही)