PRE-MEDICAL

ZOOLOGY

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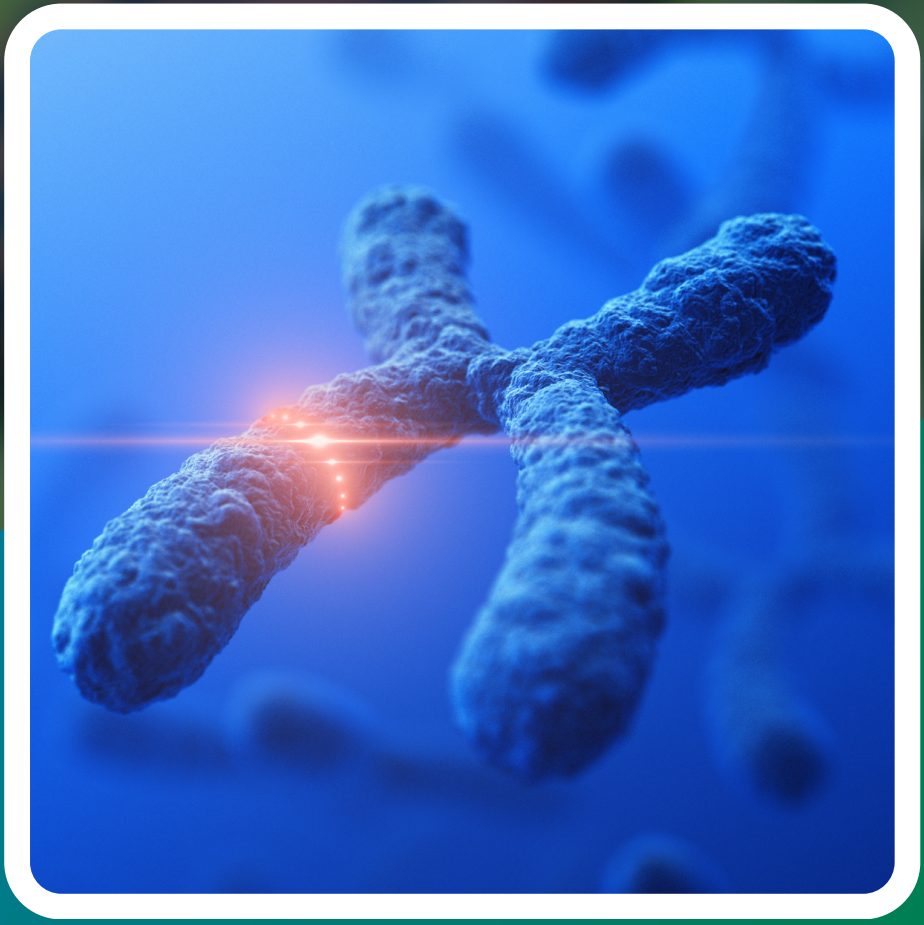

STUDY MATERIAL

Genetics : Principles of Inheritance and variations

ENGLISH MEDIUM

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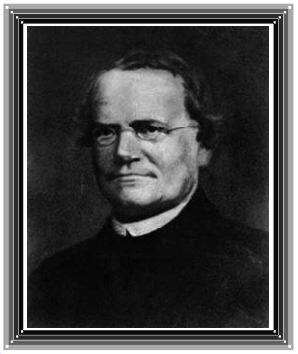
GREGOR JOHANN MENDEL (1822-1884)



In 1822, in a small village of Heinzendorf (now a part of Czech Republic), in a peasant family was born a boy named Johann Mendel, who was to make great contributions in the field of Genetics. Mendel was a brilliant student in school and studied philosophy for several years before joining the Augustinian Monastery of St. Thomas in Brno in 1843. There he took the name Gregor and received support for his studies and research throughout his life. In 1849, he took up a teaching appointment. Later, he joined the University of Vienna from 1851-1853 to study Physics and Botany and returned to Brno to teach physics and natural sciences. In 1856, Mendel performed his first set of hybridisation experiments with the garden pea and followed it up very meticulously for the next ten years or so. His interest in genetics remained alive even when he did not get any recognition for his work. He died in 1884 of a kidney disorder.

THOMAS HUNT MORGAN (1866-1945)

American geneticis, Thomas Hunt Morgan studied at Johns Hopkins University. Morgan's interest turned from embryology to the mechanisms involved in heredity. Morgan found that the rapidly multiplying Drosophila, the fruit fly ideal for studying how specific traits are transmitted through many generations. Charting the family trees of fruit flies with such mutations as stunted wings, asymmetric bodies and mismatched eye colouring, following the rediscovery of Austrian Scientist Gregor Mendel's work, Morgan elaborated the details of inheritance. He realised that there were more genes than chromosomes in Drosophila. He invented the techniques of genetic mapping. Thanks largely to Morgan's book, **The theory of the Gene (1926),** genetics was accepted as a legitimate branch of biology. He was awarded the Nobel Prize for Physiology or Medicine in 1933.

GENETICS

PRINCIPLES OF INHERITANCE AND VARIATIONS

01. INTRODUCTION

- **Introduction**
- **Mendel's Laws of Inheritance**
- **Inheritance of One Gene**
- **Inheritance of Two Genes**
- **Linkage**
- **Sex Determination**
- **Genetic Disorders**

- Genetics term was given by **W. Bateson** (Gr : Genesis, to become or to grow into)
  Genetics = Collective study of heredity & Variations.
- **Heredity :-** Transmission of genetic characters from parents to offsprings.
- **Variation :-** Differences that are seen among the members of same species.
- **Inheritance :-** The process by which characters are passed on from parents to progeny; it is the basis of heredity.

History of researches in genetics

- **G.J. Mendel** – Father of Genetics.
- **W. Bateson** – Father of Modern Genetics.
- **Morgan** – Father of Experimental genetics
  He performed experiment on *Drosophila* & proposed various concepts, like Linkage, Sex linkage, Crossing over, Criss - cross inheritance.
- **A. Garrod** = Father of human genetics & Biochemical genetics. Garrod discovered first human Metabolic genetic disorder which is called **alkaptonuria**(black urine disease).

02. SOME GENETICAL TERMS

(1) CHARACTER

- Characteristic feature of an organism. e.g. Stem height , Flower colour

(2) TRAIT

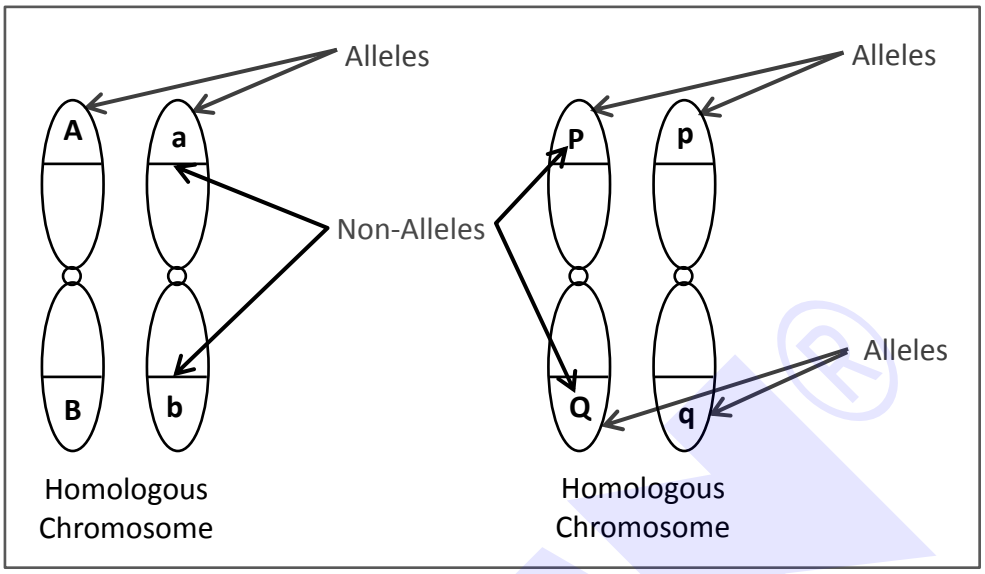
- Variable forms of a character.
  e.g. Tall/Dwarf.

(3) FACTORS/GENES

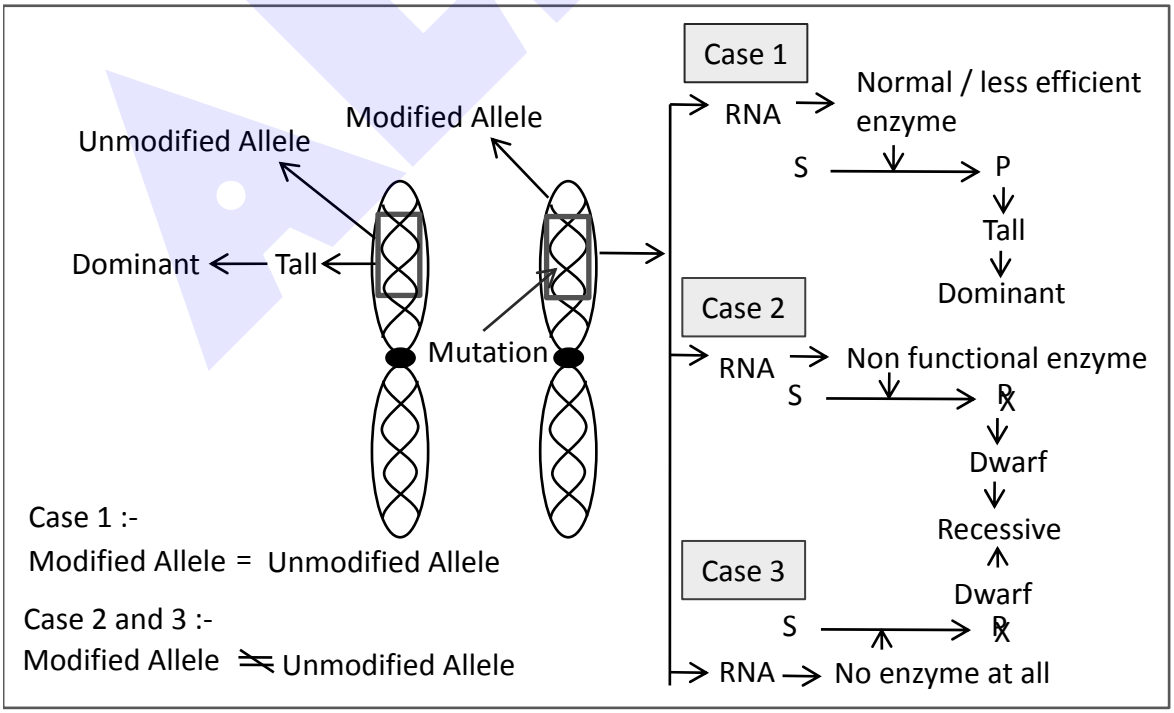
- Unit of heredity.
- Segment of DNA which is responsible for inheritance and appearance of characters. These factors were referred as **genes** by **Johannsen**.
- Mendel used term **"element"** or **"factor".**
- Dominant factors are represented by capital letters while recessive factors by small letters.

(4) ALLELE

- Alternative forms of a gene which are located on same position [loci] on the homologous chromosome are called Alleles.



- Let's take an example of a gene that contains the information for producing an enzyme. Now there are two copies of this gene, the two allelic forms.
- Let us assume (as is more common) that the normal allele produces the normal enzyme that is needed for the transformation of a substrate S.
- Theoretically, the modified 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 the first case, the modified allele is equivalent to the unmodified allele, i.e., it will produce the same phenotype/trait, i.e., result in the transformation of substrate S. Such equivalent allele pairs are very common.
- If the allele produces a non-functional enzyme or no enzyme, the phenotype may be affected. The phenotype/trait will only be dependent on the functioning of the unmodified allele.
- The unmodified (functioning) allele, which represents the original phenotype is the dominant allele and the modified allele is generally the recessive allele. Hence, in the example above the recessive trait is seen due to non-functional enzyme or because no enzyme is produced.

(5) HOMOZYGOUS/PURE

- Presence of two similar alleles of a gene in diploid organisms. Ex. TT, RR, tt

(6) HETEROZYGOUS/IMPURE

- Presence of two dissimilar alleles of a gene in diploid organisms. Ex. Tt, Rr

(7) HEMIZYGOUS

- In Diploid organism, presence of single allele of a gene. Human male are Hemizygous for sex linked genes. (genes located on sex chromosome)

(8) PHENOTYPE

- It is the external and morphological appearance of an organism for a particular character. Ex. Tall/dwarf

(9) GENOTYPE

- The genetic constitution or genetic make-up of an organism for a particular character. Ex. TT/Tt/tt

  Genotype & phenotype terms were coined by **Johannsen**.

(10) PHENOCOPY

- If two different genotypes develop similar phenotype under diff. environmental condition then they are called phenocopy of each other.

Genotype	Environment	Phenotype
TT	Normal	Tall
tt	Gibberellin rich	Tall

(11) DOMINANT ALLELE

- Allele that can express itself both in homozygous and heterozygous condition (T- allele).

  Ex- Homozygous $\rightarrow$ TT $\rightarrow$ Tall

  Heterozygous $\rightarrow$ Tt $\rightarrow$ Tall

(12) RECESSIVE ALLELE

- Allele that can express itself in homozygous condition but not in heterozygous condition (t- allele).

  Ex- Homozygous → tt → Dwarf

  Heterozygous → Tt → Tall (not dwarf)

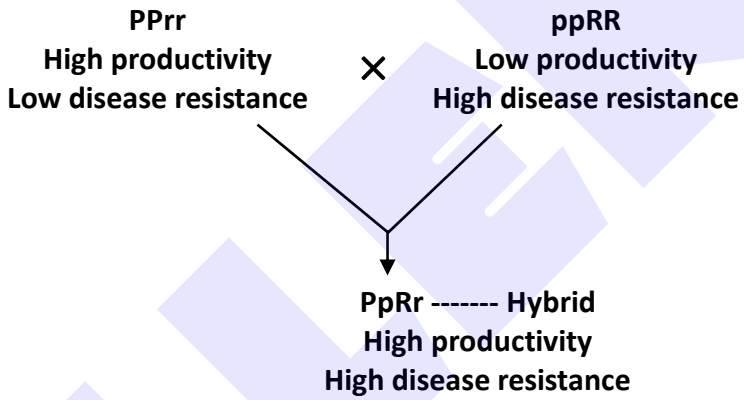
(13) GENOME

- Sum total of all the genes present in haploid number of chromosomes.

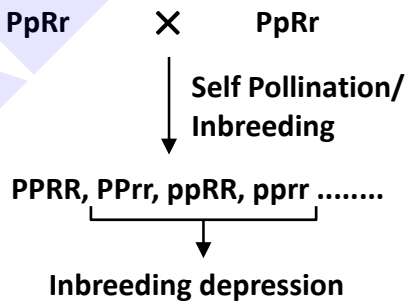
N- Base < **Nucleotide** < **Gene** < **Chromosome** < **Genome**

(14) HYBRID VIGOUR/HETEROSIS

- Superiority of hybrid offsprings over it's parents is called as **Hybrid vigour** & it develops due to Heterozygosity.



- **Inbreeding depression :-** Hybrid vigour can be lost by inbreeding or self-pollination, it is called inbreeding depression.



03. MENDELISM

- Experiments performed by Mendel on genetics and description of mechanisms of hereditary processes and formulation of principles are known as Mendelism.
- Mendel postulated various experimental laws in relation of genetics.
- **Gregor Johann Mendel (1822 - 1884) :-** Mendel was born on July 22,1822 at Heinzendorf in Austria at Silesia village. Mendel worked in Augustinian Monastery as monk at Brunn city, Austria.
- In 1856, he started his historical experiments of heredity on pea (*Pisum sativum*) plant. His experimental work continued on pea plant till 1863 (19th century).
- The results of his experiments were published in 1865.
- A paper of Mendel by the name of **"Experiments in plant hybridization"** was published in the journal.
- Mendel was unable to get any popularity. He died in 1884 without getting any credit of his work.
- After 16 years of Mendel's death, in 1900, Mendel's postulates were rediscovered **by three scientists independently.**
  - **Carl Correns** - (Germany) - (Experiment on Maize)
  - **Hugo de Vries** (Holland) (Experiment on Evening Primrose)
    He republished Mendel's results in 1901 in journal "Flora"
  - **Erich von Tschermak** - (Austria) (Experiment on different flowering plants)
- The credit of rediscovery of Mendelism goes to three scientists.
- Correns converted two postulates of Mendel into two laws of heredity/Mendelism.
  1. Law of segregation.
  2. Law of independent assortment.
- Mendel experiments remain hidden for 34 years.
- **Mendel published his work on inheritance of characters in 1865 but for several reasons, it remained unrecognised till 1900.**
  - **Firstly,** communication was not easy (as it is now) in those days and his work could not be widely publicised.
  - **Secondly,** his concept of **genes** (or **factors**, in Mendel's words) as stable and discrete units that controlled the expression of traits and, of the pair of alleles which did not 'blend' with each other, was not accepted by his contemporaries as an explanation for the apparently continuous variation seen in nature.

- **Thirdly,** Mendel's approach of using mathematics to explain biological phenomenon was totally new and unacceptable to many of the biologists of his time.
- **Finally,** though Mendel's work suggested that factors (genes) were discrete units, he could not provide any physical proof for the existence of factors or say what they were made of.

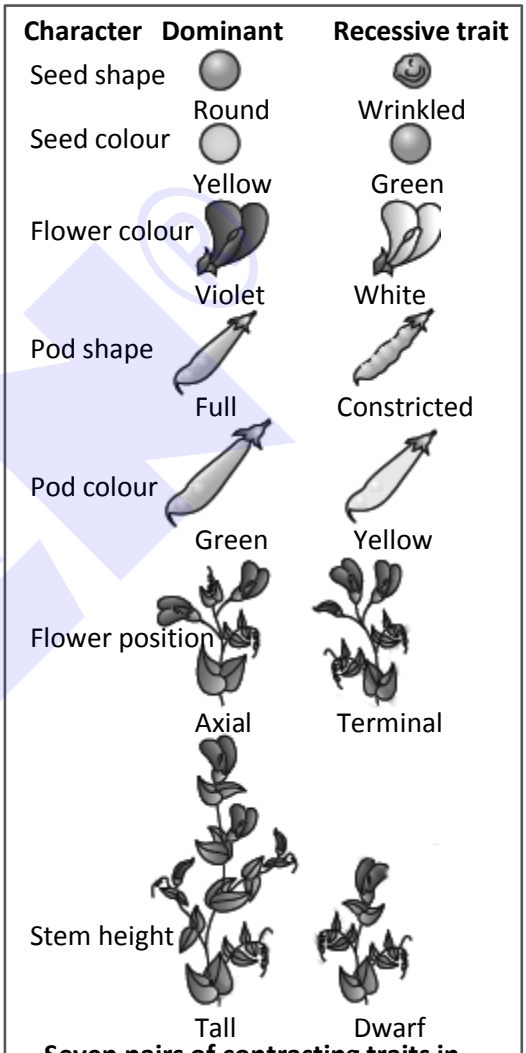
Reasons for Mendel's success :

(a) Mendel studied the inheritance of one or two characters at a time unlike his predecessors who had considered many characters at a time. (Kolreuter-Tobacco plant, John Goss & Knight-Pea plant).

(b) Selection of Material –

Garden pea plant is suitable for studies, which have the following advantages :

- Pea plant is annual plant with short life cycle of 2-3 months so large no. of offsprings can be analysed within a short period of time.
- It has many contrasting traits.
- Naturally pea plant carries out self-pollination.
- Cross pollination can be performed in it artificially so hybridization can be made possible.
- Pea plant is easy to cultivate.
- Pea seeds are large.



Seven pairs of contrasting traits in pea plant studied by Mendel

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Figure No. 5.1

(c) Mendel quantitatively analysed the inheritance of qualitative characters.

(d) He maintained the statistical records of all the experiments.

(e) His experiments had a large sampling size which gave greater credibility to the data that he collected.

(f) Also the confirmation of his inferences from experiment on successive generations of his test plants, proved that his results pointed to general rules of inheritance rather than being unsubstantiated (unverified) idea.

Mendel's work

Mendel studied 7 characters or 7 pairs of contrasting traits.

Contrasting Traits Studied by Mendel in Pea

S.No.	Characters	Ch. No.	Contrasting Traits (Dominant/Recessive)
1.	Length of plant (Stem height)	4th	Tall/Dwarf
2.	Flower position	4th	Axial/Terminal
3.	Shape of pod	4th	Inflated/Constricted
4.	Pod colour	5th	Green/Yellow
5.	Seed shape	7th	Round/Wrinkled
6.	Seed colour	1st	Yellow/Green
7.	Flower colour	1st	Violet/White

MENDEL'S EXPERIMENTAL TECHNIQUE

Steps :-

1. Selection of pure parents :-

- Mendel selected 14 true breeding varieties of garden pea plant.
- True breeding variety is that plant variety that has undergone continuous self pollination and shows stable trait inheritance and expression for several generations.
- Mendel developed true breeding varieties of garden pea plant by continuous self pollination and selection .

2. Hybridisation between pure parents :-

(a) Emasculation :- Removal of anther from bisexual flower before maturity. It is done to prevent self pollination.

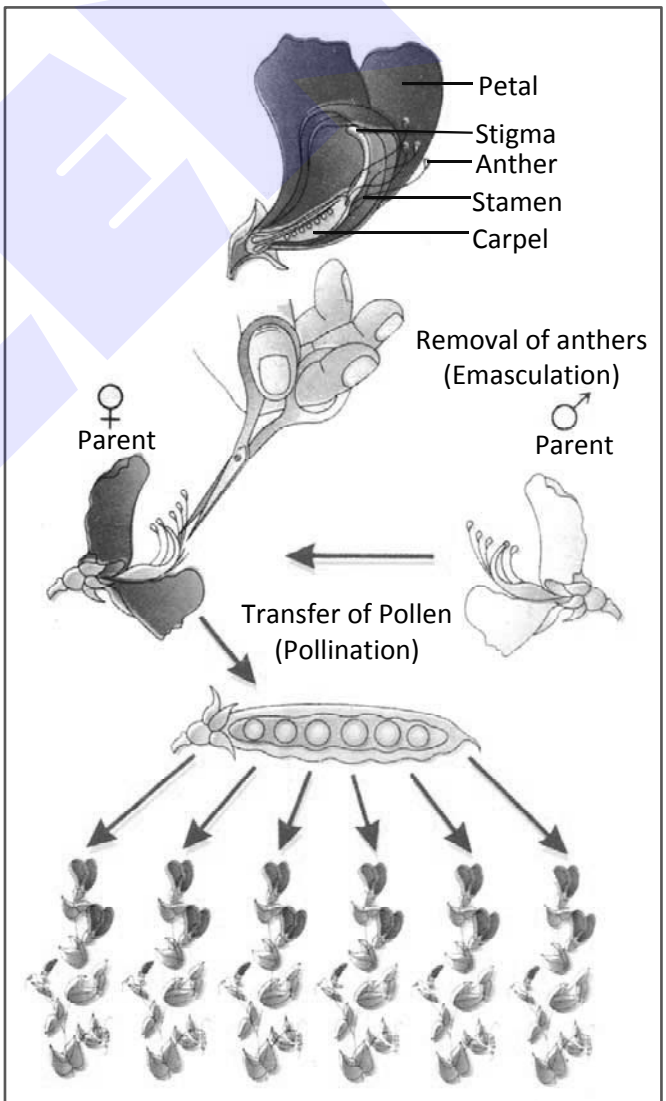
(b) Bagging :- with paper bag so as to prevent undesirable cross pollination.

(c) Tagging :- Emasculated and bagged flower are tagged by writing date and time of every steps.

(d) Dusting :- Pollen grains from selected male plant are dusted over stigma of emasculated flower with the help of a fine brush.

(e) Collection of seeds (F_1 Hybrid)

3. Selfing of F1 Hybrids to obtain F2 Generation

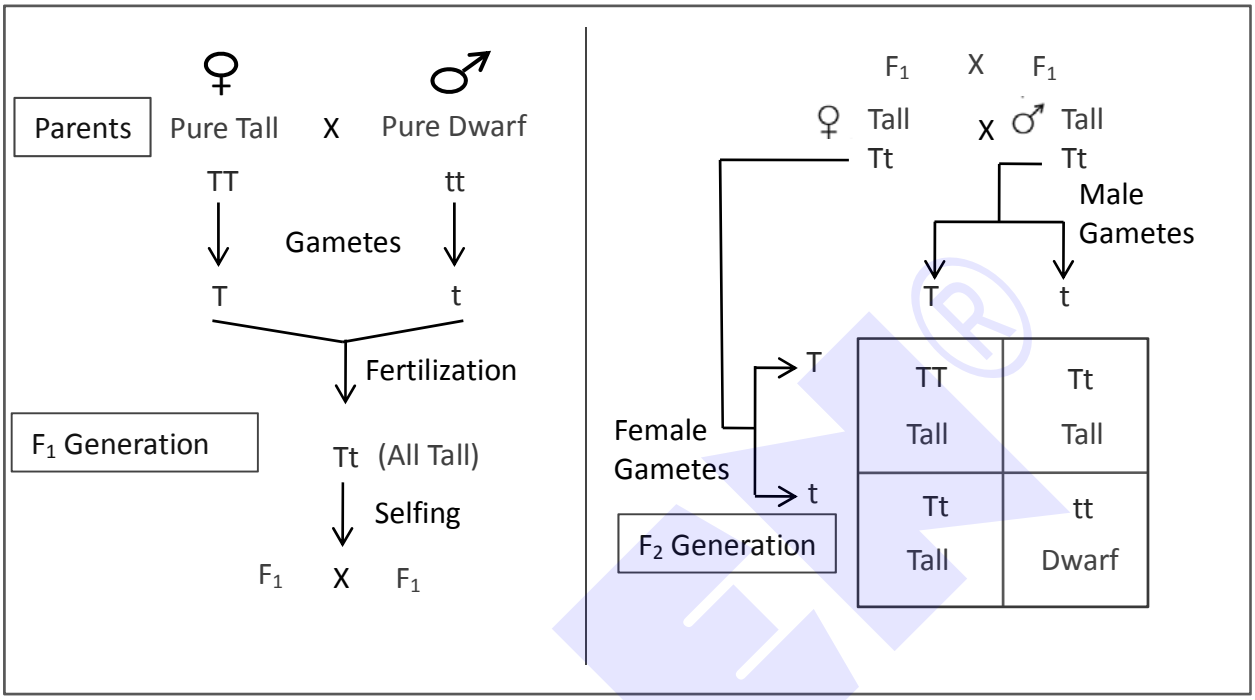


Steps in making a cross in Pea

NCERT XII Page No. 71, Figure No. 5.2

04. MONOHYBRID CROSS (Inheritance of one gene)

- A cross done to study inheritance of one character/one pair of contrasting trait at a time.
- First of all, Mendel selected tall and dwarf plants.



Conclusions of F_2 Generation

(1) Phenotype ratio = Tall : Dwarf

3 : 1

(2) Genotype ratio = TT : Tt : tt

1 : 2 : 1

(3) Types of Phenotype = $2^n = 2^1 = 2$

(4) Types of Genotype = $3^n = 3^1 = 3$

(5) No. of Zygotes/ offsprings = $4^n = 4^1 = 4$

(6) Pure Tall = TT = 1/4

(7) Impure Tall = Tt = 2/4

(8) Pure plants = TT, tt = 2/4

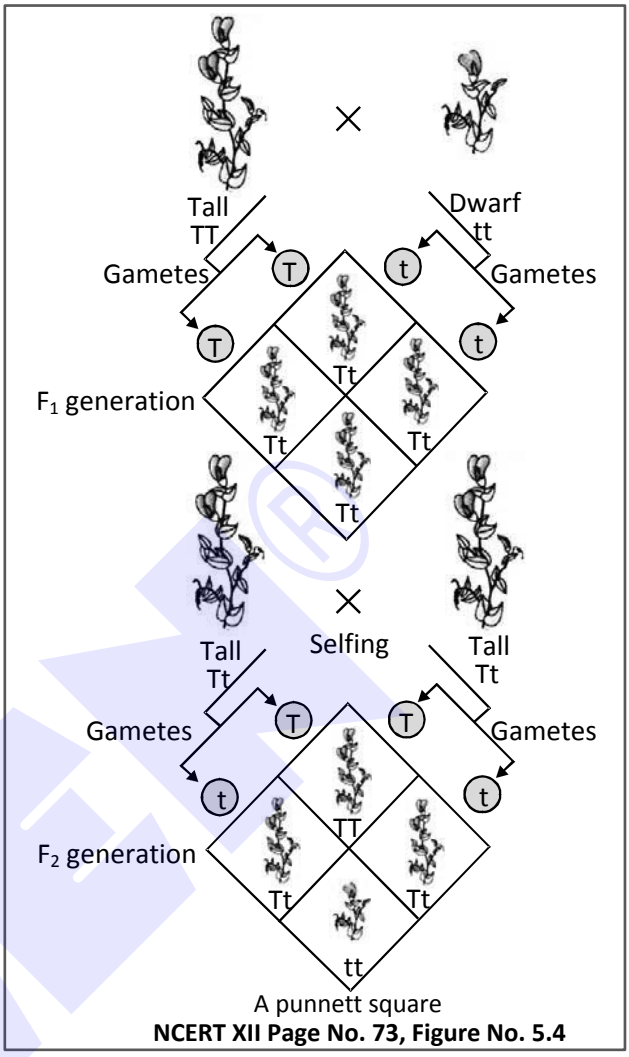
(9) Pure plants : Impure plants = 2 : 2

Checker Board Method/Punnett square

- It is a graphical representation to calculate the probability of all possible genotypes of offspring in a genetic cross.
- The production of gametes by the F_1 plants, the formation of the zygotes in F_2 generation can be understood from a diagram called Punnett Square.
- Male gametes lie horizontally on top row and female gametes lie vertically in left column.

Phenotypic ratio : Tall : Dwarf
3 : 1

Genotypic ratio : TT : Tt : tt
1 : 2 : 1



A punnett square

NCERT XII Page No. 73, Figure No. 5.4

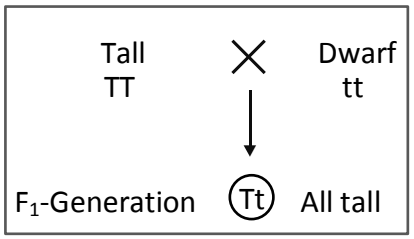
Laws based on monohybrid cross :-

(1) Law of dominance

(2) Law of segregation

(1) Law of dominance :-

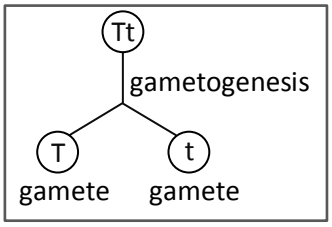
- Characters are controlled by discrete units called factors.
- Factors occur in pairs.
- In a dissimilar pair of factors one member of the pair dominates (dominant) the other (recessive).
- The law of dominance is used to explain the expression of only one of the parental characters in a monohybrid cross in the F_1 and the expression of both in the F_2. It also explains the proportion of 3:1 obtained at the F_2.



- There are two exceptions of law of dominance. [A] Incomplete dominance, [B] Co-dominance,

(2) Law of segregation:-

- This law is based on the fact that the alleles do not show any blending and that both the characters are recovered as such in the F_2 generation though one of these is not seen at the F_1 stage.
- During gamete formation; the unit factors or alleles of a pair segregate randomly and transfer inside different gamete. Each gamete receives only one factor of a pair; so gametes are pure for a particular trait. It is known as law of purity of gametes or segregation.



- This law based on the basis of F_2 generation.
- There is no exception of Law of segregation. The segregation is essential during the meiotic division in all sexually reproducing diploid organisms. (Non disjunction may be exception of this law).

Fork line method for gametes formation

To find out the composition of factors inside the gamete, we use fork line method.

AaBb = 4 types of gamete

A — B – AB = 1/4 = 25%
A — b – Ab = 1/4 = 25%

a — B – aB = 1/4 = 25%
a — b – ab = 1/4 = 25%

Types of gametes /phenotypic categories in $F_2 = 2^n$

Types of genotype in $F_2 = 3^n$

Number of zygotes in $F_2 = 4^n$

n = Number of hybrid character or heterozygous pair.

e.g. AABb
n = 1
$2^1 = 2$

A → B = AB = 1/2
A → b = Ab = 1/2

e.g. aaBBDd
n = 1
$2^1 = 2$

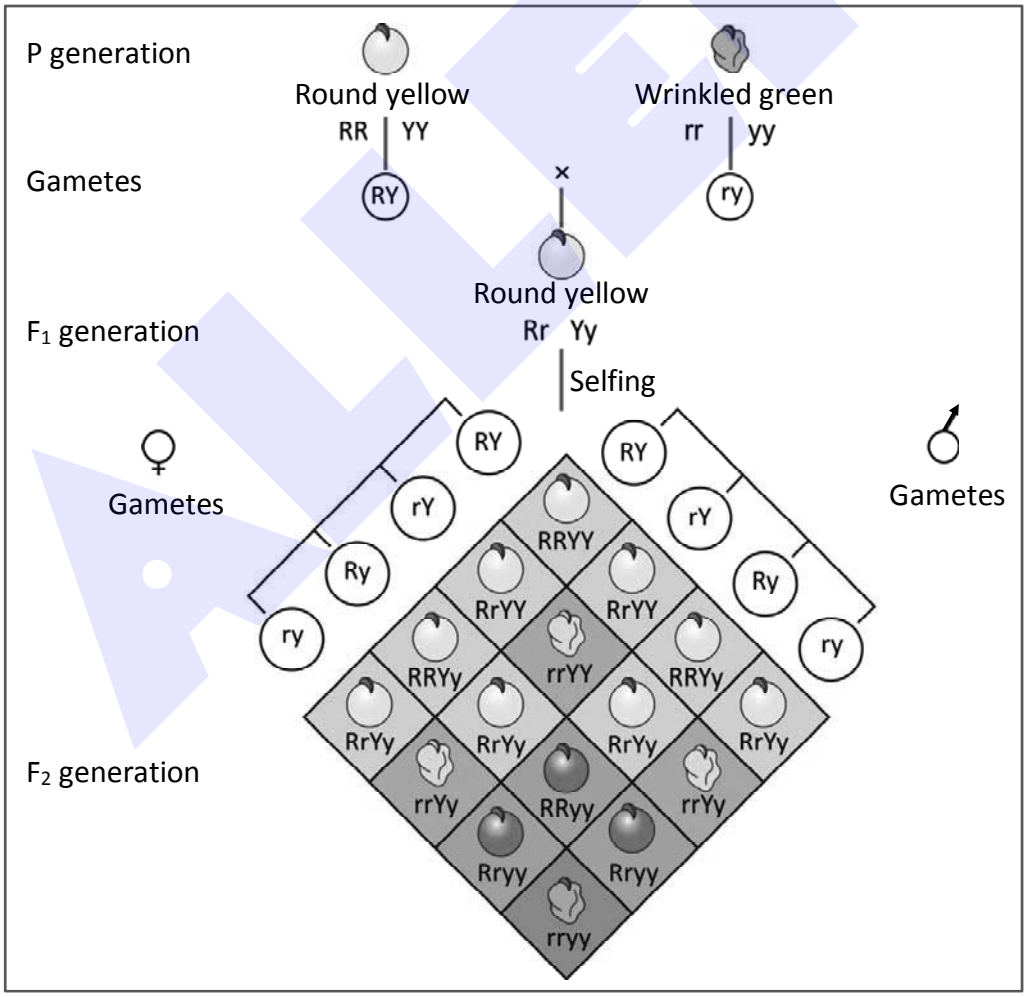
a — B → D = aBD = 1/2
a — B → d = aBd = 1/2

05. DIHYBRID CROSS (Inheritance of two genes)

- A cross done to study of inheritance of two characters or two pairs of contrasting traits.
- Mendel wanted to observe the effect of inheritance of one character on other character.
- Mendel selected :-

In garden Pea
- Seed shape
  - Round [R] - Dominant
  - Wrinkled [r] - Recessive
- Seed colour
  - Yellow [Y] - Dominant
  - Green [y] - Recessive

- Mendel crossed, yellow and round seeded plants with green and wrinkled seeded plants.
- All the plants in F_1–generation had yellow and round seeds.
- When F_1 plants were self pollinated to produce four kinds of plants in F_2 generation such as yellow round, yellow wrinkled, green round and green wrinkled.



- **Phenotypic ratio :** round yellow : round green : wrinkled yellow : wrinkled green
  9 3 3 1

  Parental combination : New combination

  9[Round yellow] + 1 [Green wrinkled] : 3[Round green] + 3 [Wrinkled yellow]

  10 : 6

  Types of phenotype = 4

- **Genotypic ratio** : RRYY RrYY RRYy RrYy rrYY rrYy RRyy Rryy rryy
  1 2 2 4 1 2 1 2 1

  Types of genotype = 9

  (1) Phenotype ratio = 9 : 3 : 3 : 1

  (2) Genotype ratio = 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

  (3) Types of Phenotype $= 2^n = 2^2 = 4$

  (4) Types of Genotype $= 3^n = 3^2 = 9$

  (5) No. of Zygotes $= 4^n = 4^2 = 16$

  Note : Mendel obtained new combinations in F_2 generation due to independent assortment.

Conclusion (Law of Independent Assortment):

- The law states that "When two pairs of traits [2 different characters] are combined in a hybrid, segregation of one pair of character is independent of the other pair of character."
- It is based on F_2 - generation of dihybrid cross.

  Exception : Linkage

06. TRIHYBRID CROSS

- A cross done to study inheritance of three characters or three pairs of contrasting traits.

RRYYTT × rryytt

↓

F_1 = RrYyTt

↓ Selfing

Phenotypic ratio : F_2 = 27 : 9 : 9 : 3 : 9 : 3 : 3 : 1

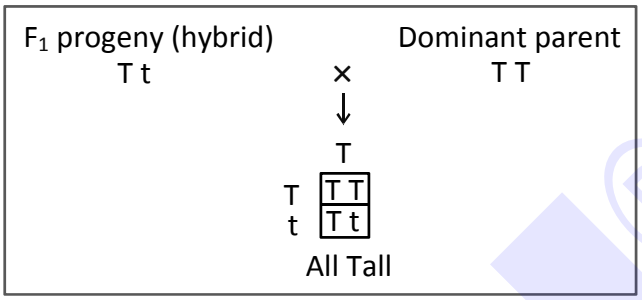
Genotype $3^3 \rightarrow$ 27 types

07. BACK CROSS

F_1 hybrid any one of the homozygous parent.

(1) OUT CROSS

- When F_1 (heterozygous) individual is crossed with dominant parent then it is termed **out cross**.
- The generations obtained from this cross, all possess dominant character. So, any analysis is not possible in F_1 generation.

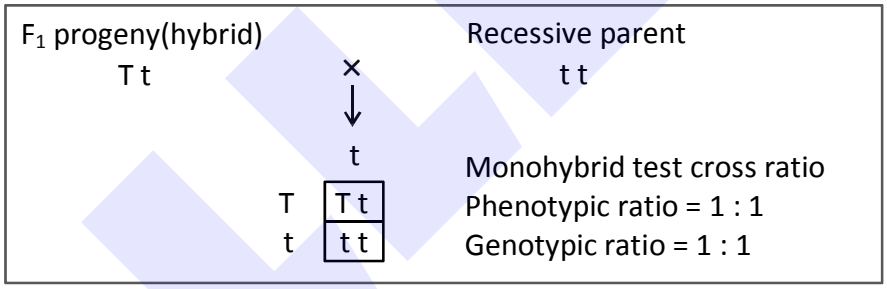


(2) TEST CROSS

When F_1 progeny (heterozygous) is crossed with recessive parent then it is called test cross.

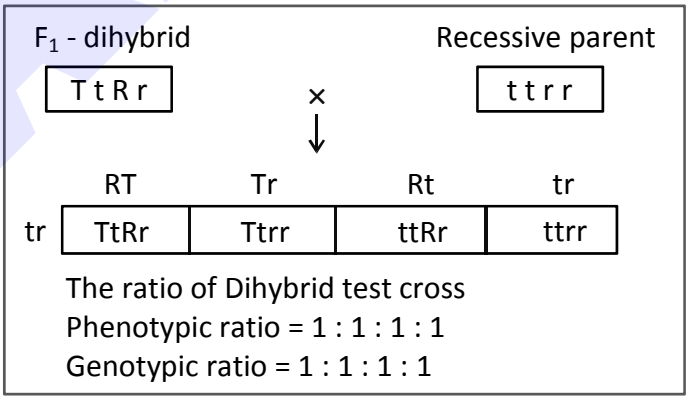
(A) Monohybrid Test Cross :-

- The progeny obtained from the monohybrid test cross are in equal proportion, means 50% are dominant phenotypes and 50% are recessive phenotypes.



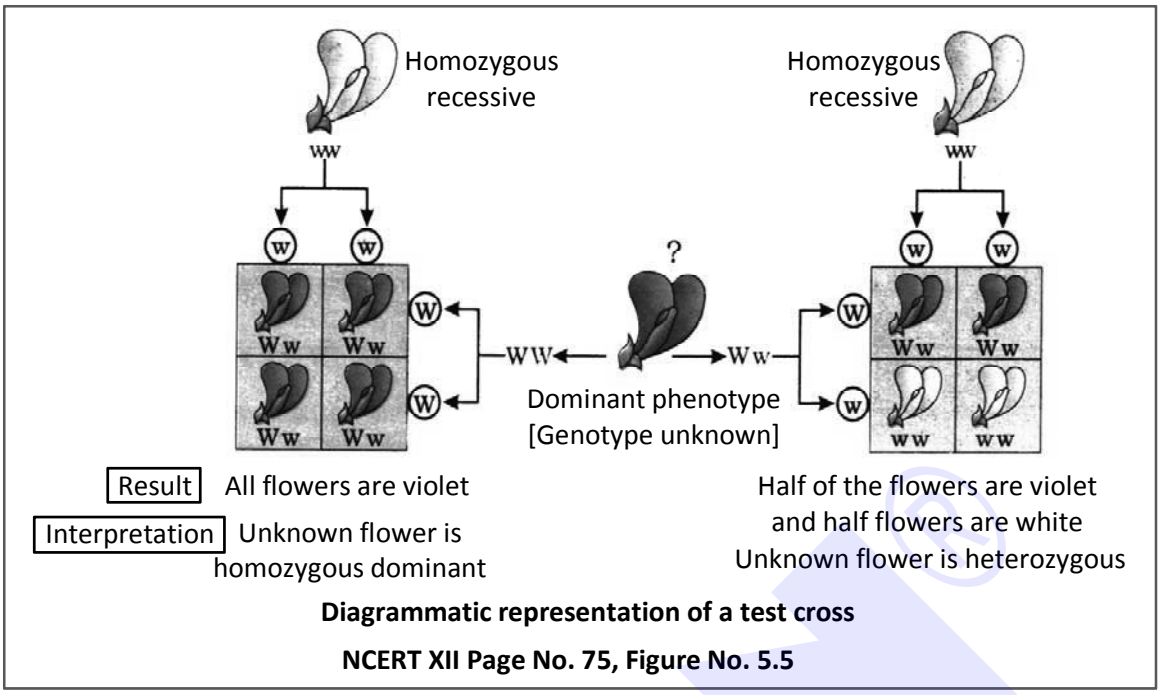
(B) Dihybrid Test Cross :-

- The progeny obtained from dihybrid test cross are of four types and each of them is 25%.



Conclusion :-

- In test cross phenotypic and genotypic ratio are same.
- **Test cross helps to find out the genotype of unknown dominant individual.**

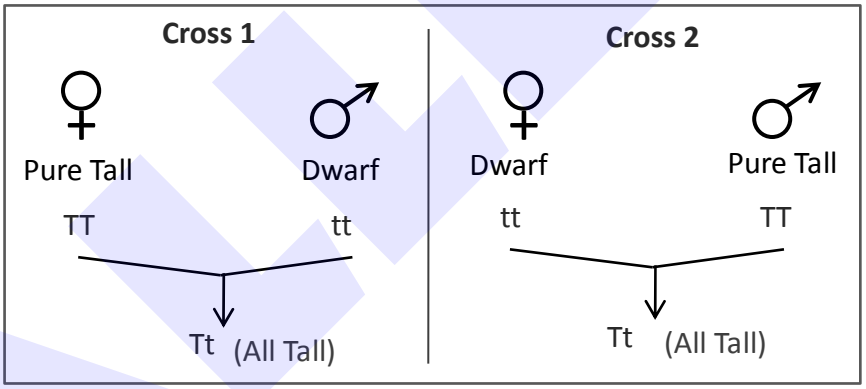


Diagrammatic representation of a test cross

NCERT XII Page No. 75, Figure No. 5.5

08. RECIPROCAL CROSS

- It is a set of two crosses in which sex of the parents are reversed.
- Mendel conducted reciprocal cross to check whether the inheritance of characters is dependent on parental sex or not.



Result :- Mendel obtained similar result in both cross

Conclusion :- Inheritance of characters is not dependent on parental sex

- Characters studied by Mendel were controlled by karyogene and these genes are located on autosome.
- Results of reciprocal cross will change In two cases
  (1) Genes present in cytoplasm (cytoplasmic inheritance)
  (2) Genes present on sex chromosomes (sex linkage)

★ Golden Key Points ★

Monohybrid cross

- Phenotypic ratio = 3 : 1
- Genotypic ratio = 1 : 2 : 1
- Test cross ratio = 1 : 1

Dihybrid cross

- Phenotypic ratio = 9 : 3 : 3 : 1
- Genotypic ratio = 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1
- Test cross ratio = 1 : 1 : 1 : 1

BEGINNER'S BOX
INTRODUCTION TO RECIPROCAL CROSS

1. If the cell of an organism heterozygous for three pairs of genes represented by AaBbCc, undergoes meiosis then possible type of gametes will be :-

(1) 4 (2) 2 (3) 8 (4) 12

2. Which law of Mendel is universal in nature?

(1) Law of dominance (2) Law of indepenent assortment

(3) Law of segregation (4) Linkage

3. When aaBBcc is crossed with AaBbCc then the ratio of hybrid for all the three genes is :-

(1) $\frac{1}{8}$ (2) $\frac{1}{4}$ (3) $\frac{1}{16}$ (4) $\frac{1}{32}$

4. According to Mendelism, which pair of character is showing dominance ?

(1) Terminal position of flower and green colour of seed coat.

(2) Wrinkled seeds and green colour of seed coat.

(3) Yellow pod and round seeds.

(4) Green pod and axial position of flower.

5. A plant of F_1 generation has genotype AABbCc. On selfing of this plant what is phenotypic ratio in F_2 generation?

(1) 3 : 1 (2) 9 : 3 : 3 : 1 (3) 1 : 1 (4) 27 : 9 : 9 : 9 : 3 : 3 : 3 : 1

6. A character which is expressed in a hybrid is called

(1) Dominant (2) Recessive (3) Co-dominant (4) Epistatic

7. Alleles are :-

(1) Alternate forms of a gene

(2) Homologous chromosome

(3) Pair of sex chromosome

(4) None of these

8. When F_1 generation hybrid tall Tt is crossed with dwarf tt parent, it is a case of:-

(1) Dihybrid cross (2) Test cross

(3) Crossing over (4) Reciprocal cross

9. The ultimate biological unit which controls heredity, is called :

(1) Genome (2) Chromosome

(3) Genotype (4) Gene

10. In case of inheritance of one gene, 3 : 1 phenotypic ratio can be explained on the basis of–

(1) Incomplete dominance (2) Codominance

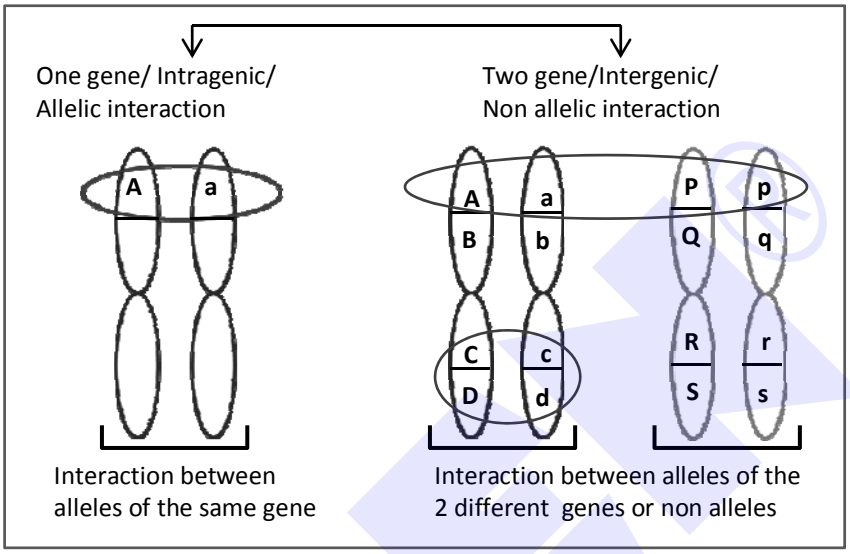
(3) Dominance (4) Linkage

POST - MENDELIAN GENETICS

- After rediscovery of Mendel's work

09. GENE INTERACTION

- Genes interact to modify the phenotype.



Gene Interaction

One gene/ Intragenic/ Allelic interaction	Two gene/Intergenic/ Non allelic interaction
Examples :-	Examples :-
1. Incomplete dominance	1. Complementary gene
2. Co- dominance	2. Epistatic gene
3. Multiple alleles	
4. Pleiotropic gene	

- **Qualitative character** → This character is not dependent on number of dominant alleles.
- **Quantitative character** → This character is dependent on number of dominant alleles.

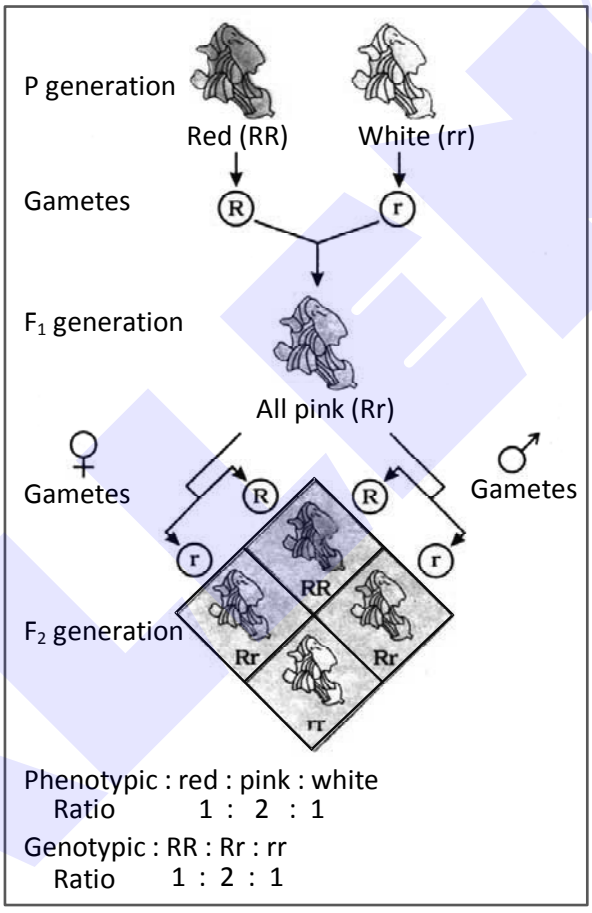
(A) Incomplete Dominance :-

- Exception to Mendel's law of dominance .
- In this interaction dominant allele is not fully dominant over recessive allele so in heterozygous condition an intermediate phenotype appears.

- Thus, F_1 hybrid does not resemble to any of the parent.
- Incomplete dominance was first discovered by **Carl Correns** in *Mirabilis jalapa*. This plant is called as **'4 O' clock plant** 'or' **Gul-e-Bans'**.

(i) Flower colour in *Mirabilis jalapa* :

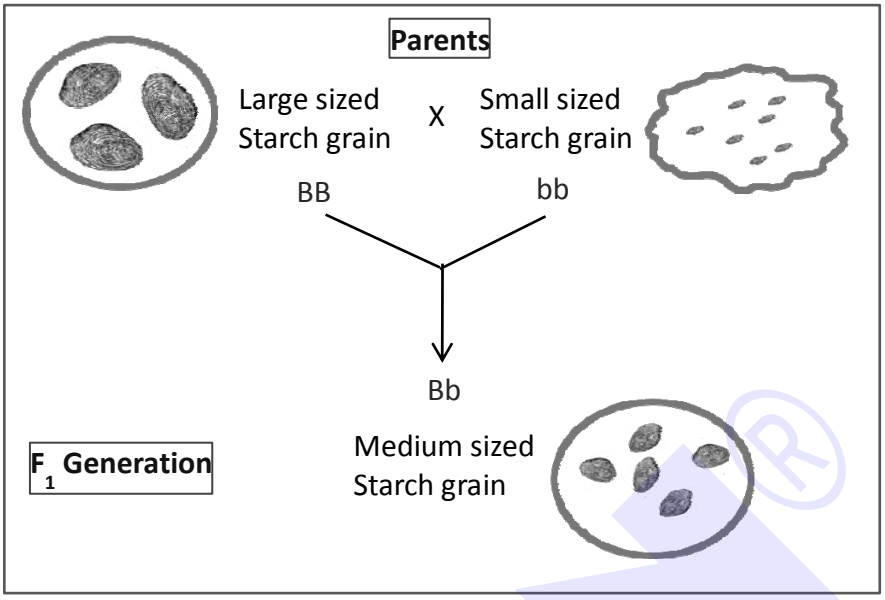
- When plant with red flowers is crossed with plant with white flowers, plants with pink flowers are obtained in F_1 generation. The reason of this is that the gene for red colour is incompletely dominant over the gene for white colour.



(ii) Flower colour in *Antirrhinum majus* :-

- Incomplete dominance is also seen in flower colour of this plant. This plant is also known as '**Snapdragon**' or '**Dog flower**'. Incomplete dominance is found in this plant which is the same as *Mirabilis.*

(iii) Size of starch grains in pea plant :-



(B) Co-Dominance :-

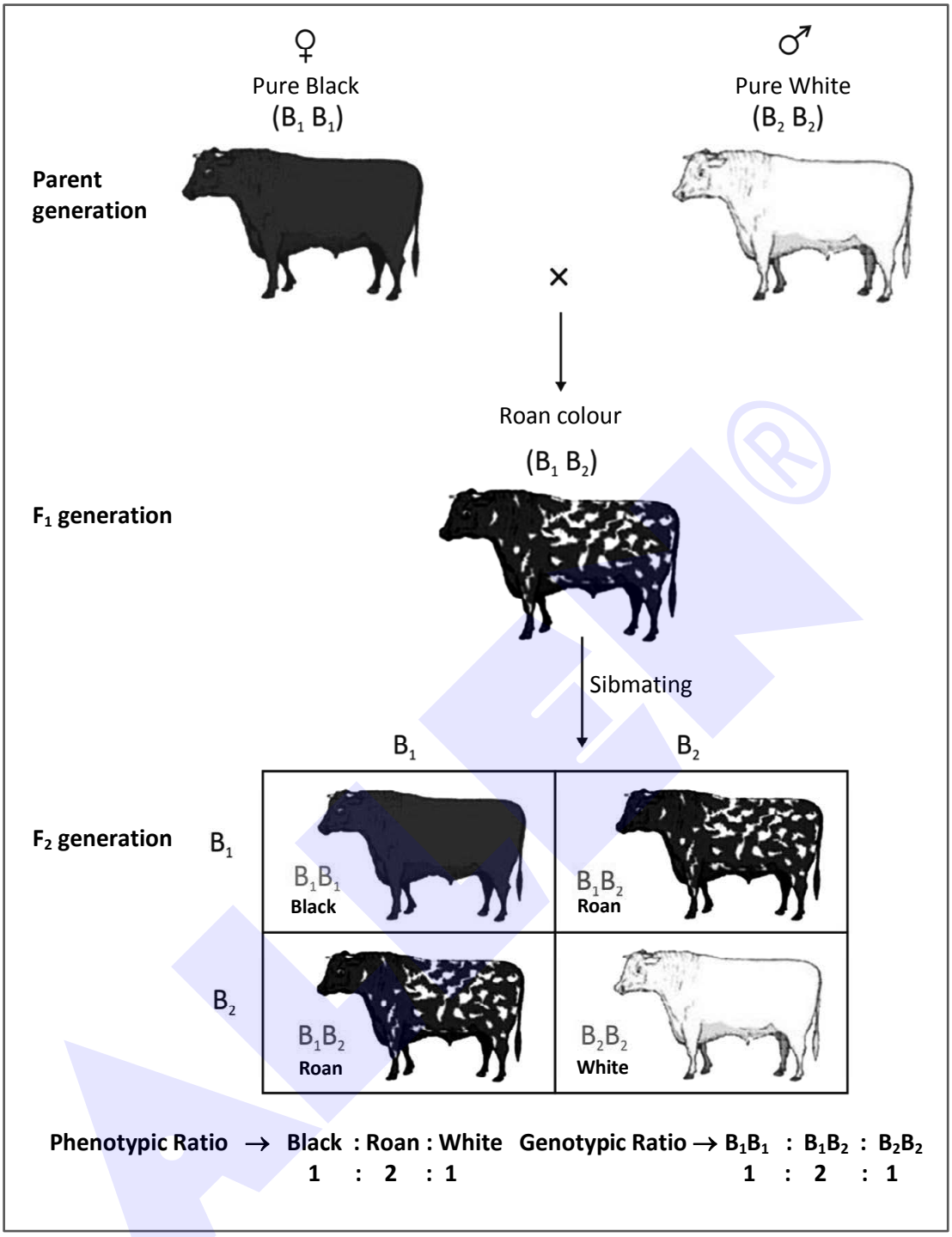
- Exception to Mendel's law of dominance.
- In this interaction in heterozygous condition both alleles are equally dominant and show their independent expression so in heterozygous condition an intermediate phenotypes does not appear.
- Thus, F_1 hybrid resemble to both of the parent.

Examples :-

(i) Coat Colour in Cattles:-

When a black parent is crossed with white parent, a roan coloured F_1 progeny is produced.

Note :- F_2 generation is obtained in animals by sib-mating cross.



Phenotypic Ratio → Black : Roan : White
1 : 2 : 1

Genotypic Ratio → B_1B_1 : B_1B_2 : B_2B_2
1 : 2 : 1

Other Examples of Co-dominance :

(i) AB blood group inheritance (I^AI^B)

(ii) Carrier of Sickle cell anaemia (Hb^A Hb^S)

- In case of dominance, F_1 resembles to either of the two parents.
- In case of incomplete dominance, F_1 is intermediate of the two parents.
- In case of co-dominance, F_1 resembles to both of the parents.

(C) Multiple Alleles :–

- More than 2 alternative forms of same gene are called multiple alleles.
- Multiple alleles are formed due to mutation.
- Multiple alleles are located on same locus of homologous chromosomes.
- A diploid individual contains two alleles and gamete contains one allele for a gene.
- Since a diploid individual can have only two alleles of a gene and other alleles can be seen in other members of population, so multiple allele can be studied in a **population** but not in an individual.

- If n is the number of alleles of a gene then types of different possible genotypes = $\boxed{\frac{n(n+1)}{2}}$

  n = total number of alleles

Example of multiple alleles :

ABO blood group

- ABO blood groups are controlled by gene I
- The gene (I) has 3 alleles :- I^A, I^B & i

  I^A = dominant

  I^B = dominant

  i = recessive
- The plasma membrane of the red blood cells has sugar polymers that protrude from its surface and the kind of sugar controlled by the gene I.

Possible phenotypes - A, B, AB, O

Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Blood types of offspring
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	i	I^A i	A
I^B	I^A	$I^A I^B$	AB
I^B	I^B	$I^B I^B$	B
I^B	i	I^B i	B
i	i	ii	O

NCERT XII Page No. 77, Table No. 5.2

Possible number of genotype $= \frac{3(3+1)}{2} = 6$ genotypes

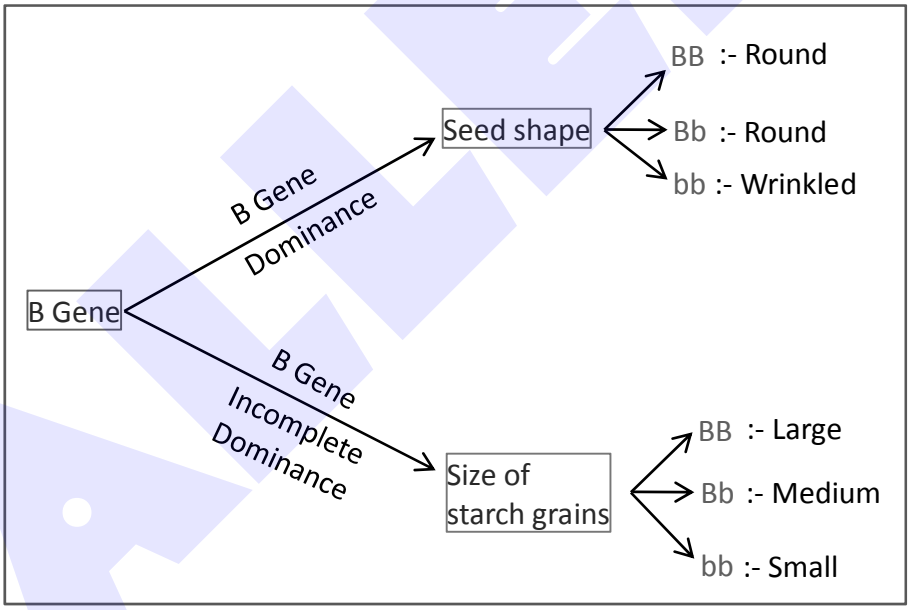
(D) Pleiotropic Gene :-

- Gene which controls more than one character is called pleiotropic gene.
- This gene shows multiple phenotypic effect.
- We have so far seen the effect of a gene on a single phenotype or trait. There are however instances where a single gene can exhibit multiple phenotypic expression. Such a gene is called pleiotropic gene. The underlying mechanism of pleiotropy in most cases is the effect of a gene on metabolic pathways which contributes towards different phenotypes.

For example :

(i) In Pea plant :

- Seed shape and size of starch grains are controlled by the same gene located on 7^{th} chromosome.
- Occasionally, a single gene product may produce more than one effect. For example, starch synthesis in pea seeds is controlled by one gene. It has two alleles (B and b).



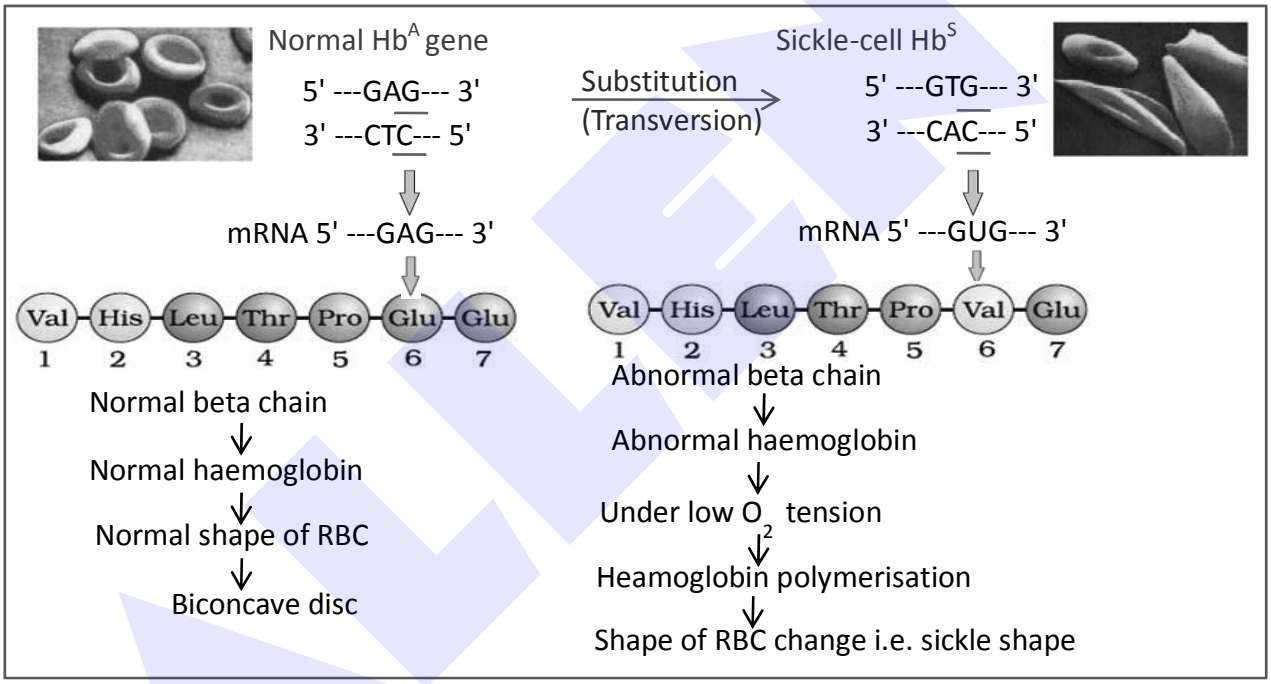
- Therefore, dominance is not an autonomous feature of a gene or the product that it has information for. It depends as much on the gene product and the production of a particular phenotype from this product as it does on the particular phenotype that we choose to examine, in case more than one phenotype is influenced by the same gene.

(ii) Most of the human genetic disorders are pleiotropic :-

(a) Phenylketonuria

(b) Sickle cell anaemia (Autosome linked recessive disorder) :

- The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule.
- The substitution of amino acid in the globin protein results due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG.
- The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure.



- Gene Hb^S provide a classical example of pleiotropy. It not only causes haemolytic anaemia but also results in increased resistance to one type of malaria that is caused by the parasite *Plasmodium falciparum.*
- The sickle cell Hb^S allele also has pleiotropic effect on the development of many tissues and organs such as bone, lungs, kidney, spleen, heart.

(2) NON ALLELIC INTERACTION/INTERGENIC INTERACTION

- When interaction takes place between alleles of two different genes or non alleles then it is called non allelic gene interaction. It changes or modifies the phenotype.

(A) Complementary Gene :-

- When two non allelic genes are not able to express the dominant trait when present separately.
- At least one dominant allele of both the genes must be present to express the dominant trait. Thus both the genes complement each other to express the dominant trait.

Example :- Colour of flowers in *Lathyrus odoratus* (Sweet pea) :-

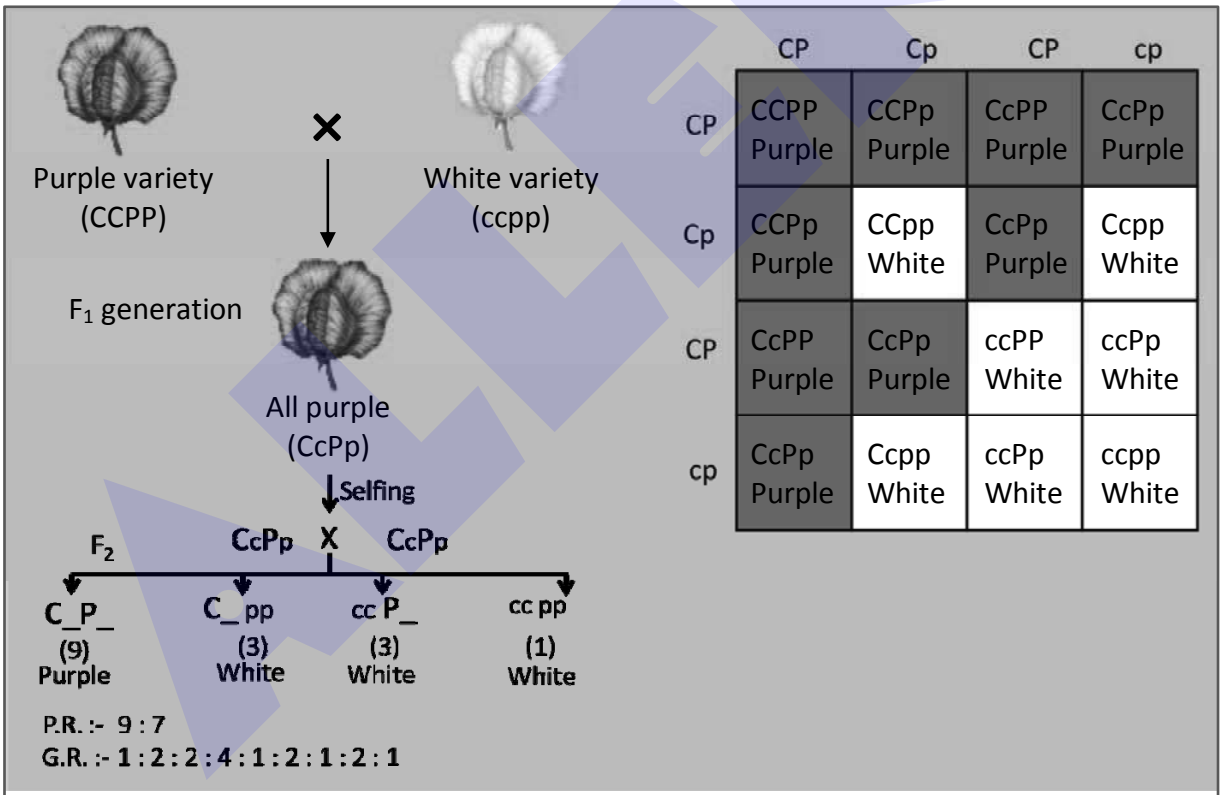
C _ P _ Purple coloured

C _ pp colour less

cc P _ colour less

ccpp colour less

Raw material (Colourless) $\xrightarrow{\text{Gene C}}$ Chromagen (Colourless) $\xrightarrow{\text{Gene P}}$ Anthocyanin (Purple)



	CP	Cp	CP	cp
CP	CCPP Purple	CCPp Purple	CcPP Purple	CcPp Purple
Cp	CCPp Purple	CCpp White	CcPp Purple	Ccpp White
CP	CcPP Purple	CcPp Purple	ccPP White	ccPp White
cp	CcPp Purple	Ccpp White	ccPp White	ccpp White

Thus phenotypic ratio of complementary genes = Coloured : Colourless 9 : 7

(B) Epistasis :-

- When, a gene prevents the expression of another non-allelic gene, this phenomenon is known as Epistasis.

- Gene which inhibits or suppresses the expression of another non allelic gene is called epistatic gene and gene whose expression is suppressed by epistatic gene is called hypostatic gene.
- Epistasis is of two types :- (1) Dominant epistasis (2) Recessive epistasis

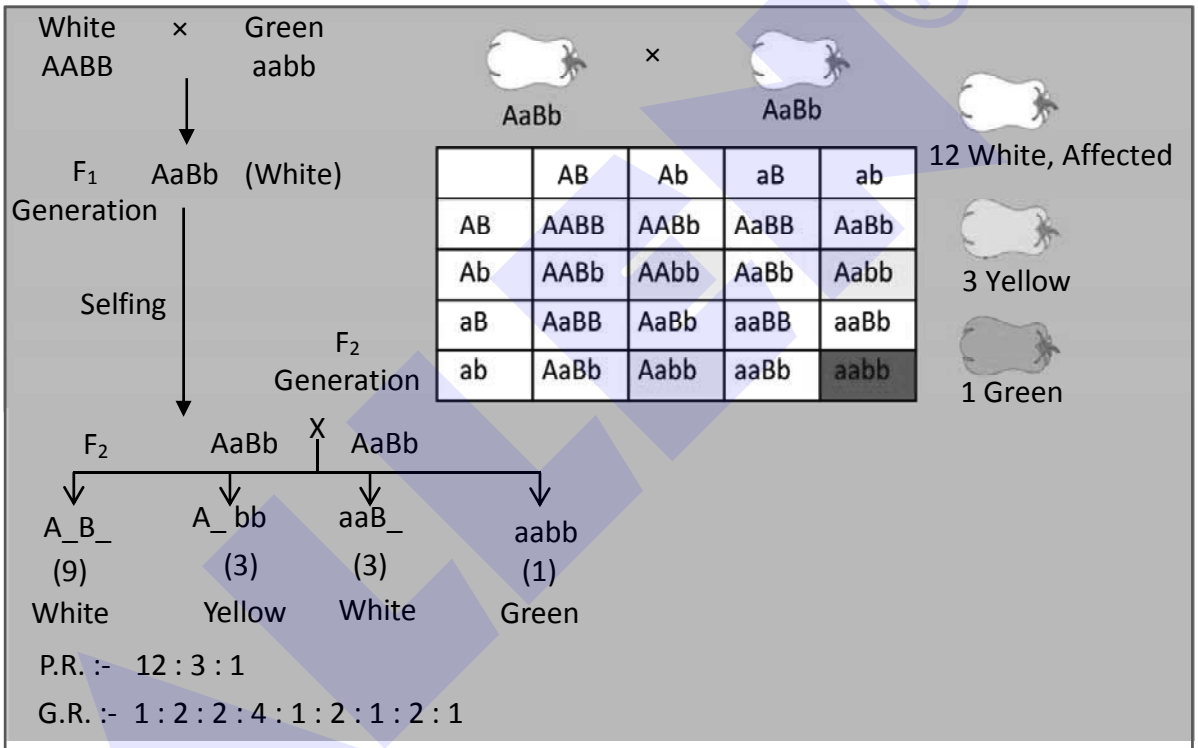
Dominant epistasis : Example - Fruit colour in summer squash *(Cucurbita pepo)*

A = Dominant allele for yellow colour of fruit

a = Recessive allele for green colour of fruit

B = Epistatic gene over A and a gene and forms white colour of fruit.

Following types of offsprings will be obtained in a Mendelian pattern of cross-



The phenotypic ratio of F_2-generation in dominant epistasis is- 12 : 3 : 1

10. POLYGENIC INHERITANCE/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 don't just have tall or short people as two distinct alternatives but a whole range of possible heights.
- Polygenic traits are generally controlled by two or more genes and each gene shows quantitative effect.

- Besides the involvement of multiple genes, polygenic inheritance also takes into account the influence of environment. Human skin colour is classic example for this.
- In a polygenic trait the phenotype reflects the contribution of each allele, i.e., the effect of each allele is additive.
- Polygenic inheritance is inheritance of characters in which one character is controlled by many genes and intensity of phenotype depends upon the number of dominant alleles/quantity of product.
- Types of phenotype = (2n + 1),
  Types of genotype = 3^n
  Zygotic combinations = 4^n (here n = number of polygenes)

Example-1. Kernel colour of wheat :- Regulated by two polygenes (A and B).

Parent Max. expression AABB Red X Min. expression aabb White

F_1 AaBb Intermediate red

Selfing

F_2 AaBb X AaBb

AABB	AABb	AaBB	AaBb
AABb	AAbb	AaBb	Aabb
AaBB	AaBb	AAbb	aaBb
AaBb	Aabb	aaBb	aabb

No. of Dominant alleles	Genotype	Phenotype	Ratio
4	AABB -1	Red	1
3	AABb -2 AaBB -2	Light red	4
2	AaBb -4 AAbb -1 aaBB -1	Intermediate red	6
1	Aabb -2 aaBb -2	Very light red	4
0	aabb -1	White	1

In F_2 generation :-

(1) P.R. :- 1 : 4 : 6 : 4 : 1

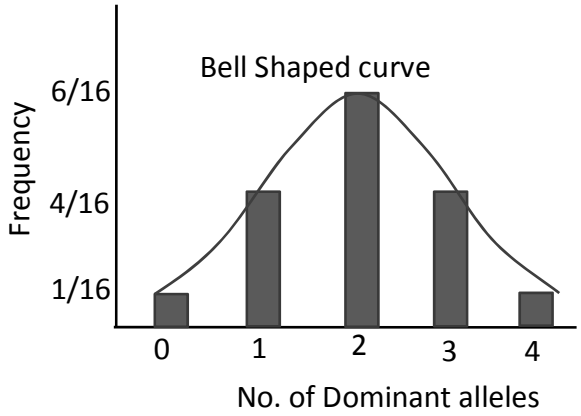
(2) G.R. :- 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

(3) Types of phenotype :- 2n + 1 = (2 x 2 + 1) = 5

(4) Type of Genotype :- $3^n = 3^2 = 9$

(5) Zygotic combination :- $4^n = 4^2 = 16$

(6) % of parental plant :- $\frac{2}{16} \times 100 = 12.5\%$



Example-2. :- **Colour of the skin in Human :-**

- The inheritance of colour of skin in human was studied by **Davenport**.
- Human skin colour is regulated by three polygenes (A, B and C).
- To understand with the dominant forms A, B and C control skin colour in human with the dominant forms A, B, and C responsible for dark skin colour and the recessive forms a, b, and c for light skin colour.
- The genotype with all the dominant alleles (AABBCC) will have the darkest skin colour and that with all the recessive alleles (aabbcc) will have the lightest skin colour.
- As expected the genotype with three dominant alleles and three recessive alleles will have an intermediate skin colour. In this manner the number of each type of alleles in the genotype would determine the darkness or lightness of the skin in an individual.

Parent Max. expression AABBCC Negro Black X Min. expression aabbcc White

F_1 AaBbCc Intermediate Brown/ Mulatto

↓ In breeding

F_2 AaBbCc X AaBbCc

↓

Checker board of 64

No. of Dominant alleles	Phenotype	Ratio
6	Negro Black	1
5	Very Dark Brown	6
4	Dark Brown	15
3	Mulatto	20
2	Light Brown	15
1	Very Light Brown	6
0	White	1

In F_2 generation :-

(1) P.R. :- 1 : 6 : 15 : 20 : 15 : 6 : 1

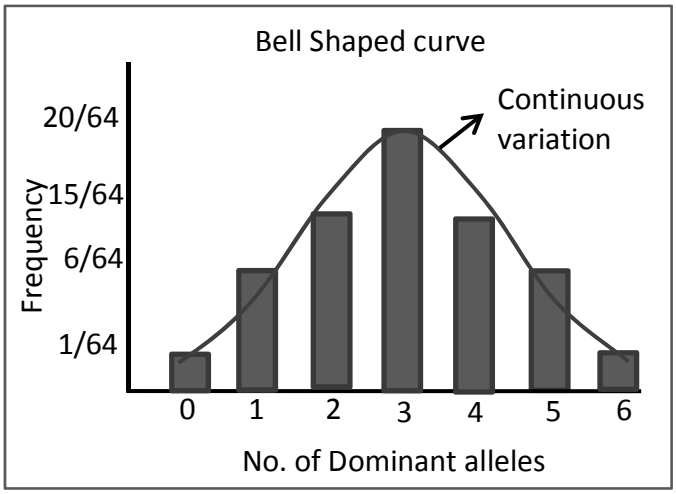
(2) G.R. :- (1 : 2 : 1) X (1 : 2 : 1) X (1 : 2 : 1)

(3) Types of phenotype :- 2n + 1= (2x3+1) = 7

(4) Type of Genotype :- $3^n = 3^3 = 27$

(5) Zygotic combination :- $4^n = 4^3 = 64$

(6) % of parental offsprings :- $\frac{2}{64}$ X 100 = 3.125%



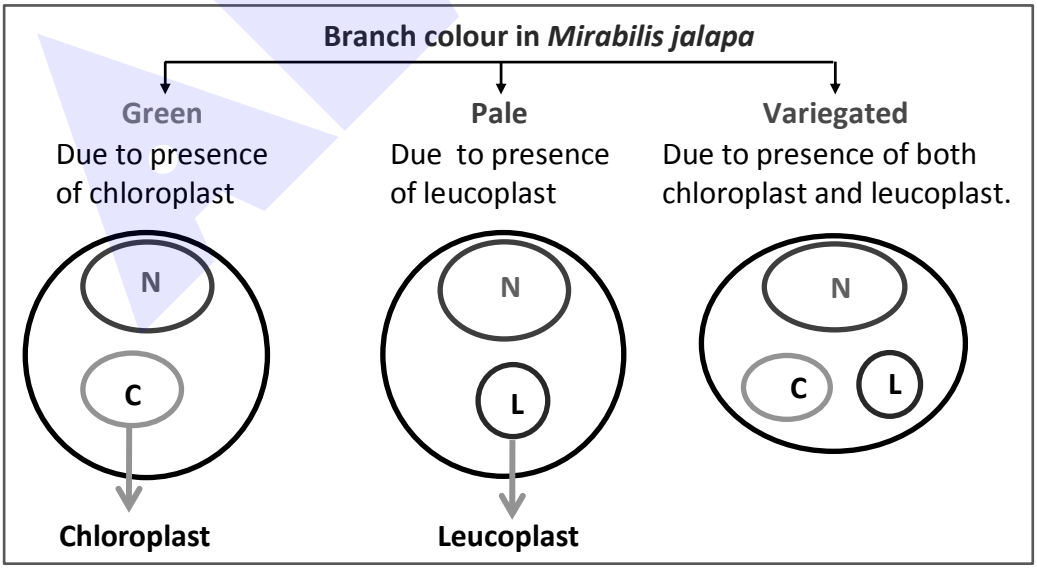
11. CYTOPLASMIC INHERITANCE

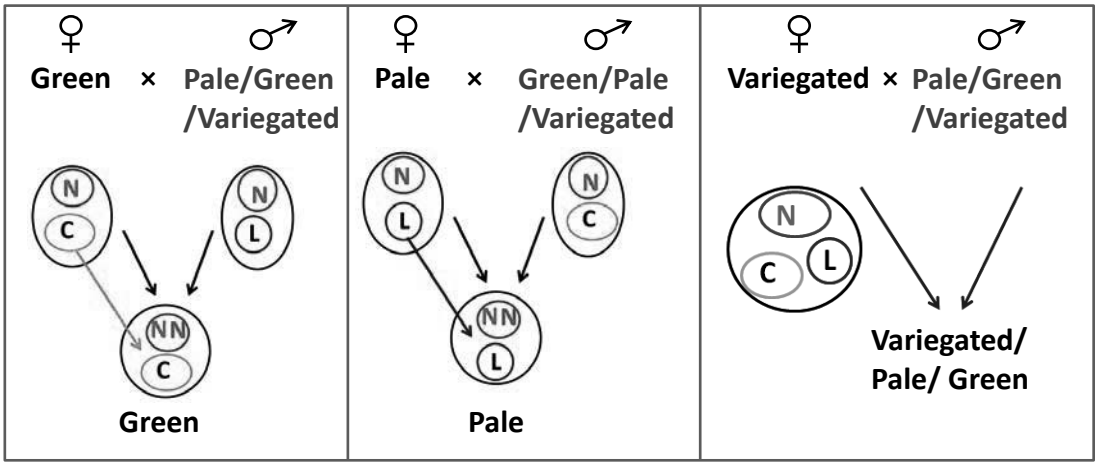
- Discovered by Correns.
- Inheritance of characters which are controlled by cytogene or cytoplasmic gene is called cytoplasmic inheritance. Genes which are present in cytoplasm are called **'cytogene'** or **'plasmagene'** or **extra nuclear gene.**
- Total cytogenes present in cytoplasm is called **'Plasmon'**.
- A gene which is located in the nucleus is called '**karyogene**'.
- Inheritance of cytogene occurs only through female. (so it is also called as maternal inheritance)
- If there is a reciprocal cross in this condition, then results may be affected or different.
- Cytoplasmic inheritance involving organelles like, Chloroplast and mitochondria is called as **organellar inheritance.**

Example of Organellar Inheritance :

(i) Plastid inheritance in *Mirabilis jalapa* –

- Cytoplasmic inheritance was first discovered by **Correns** in *Mirabilis jalapa*.
- In *Mirabilis jalapa* branch (leaf) colour is decided by type of plastid present in leaf cells. So it is an example of cytoplasmic inheritance





(ii) Male sterility in maize plant :– Gene of male sterility is present in mitochondria.

Golden Key Points

- Incomplete dominance (Monohybrid cross)
  - Phenotypic ratio = 1 : 2 : 1
  - Genotypic ratio = 1 : 2 : 1
- Co-dominance (Monohybrid cross)
  - Phenotypic ratio = 1 : 2 : 1
  - Genotypic ratio = 1 : 2 : 1
- Possible genotype number (In multiple alleles) = $\frac{n(n+1)}{2}$
- Complementary gene = 9 : 7
- Dominant epistasis = 12 : 3 : 1
- Polygenic inheritance
  - 1 : 4 : 6 : 4 : 1 (For two genes)
  - 1 : 6 : 15 : 20 : 15 : 6 : 1 (For three genes)
- Types of phenotype in polygenic inheritance = (2n + 1)
- Contribution of each dominant allele = $\frac{\text{Maximum expression} - \text{Minimum expression}}{\text{Total number of dominant alleles}}$

BEGINNER'S BOX — GENE INTERACTION TO CYTOPLASMIC INHERITANCE

1. In case of co-dominance the monohybrid ratio of phenotypes in F_2 generation is :-

(1) 3 : 1 (2) 1 : 2 : 1 (3) 1 : 1 : 1 : 1 (4) 2 : 2

2. Which cross yields red, white and pink flower variety of Snapdragon flower?

(1) RR × Rr (2) Rr × RR (3) Rr × Rr (4) Rr × rr

3. In polygenic inheritance, a trait is controlled by two polygenes. Two individuals which are heterozygous for both genes, crossed each other, such type of cross produces what phenotypic ratio ?

(1) 1 : 2 : 1 (2) 9 : 3 : 3 : 1

(3) 1 : 4 : 6 : 4 : 1 (4) 1 : 6 : 15 : 20 : 15 : 6 : 1

4. If dominant C and P genes are essential for the development of purple colour in *Lathyrus odoratus* flowers. What would be the ratio of purple and white colour in a cross between CcPp × cc Pp.

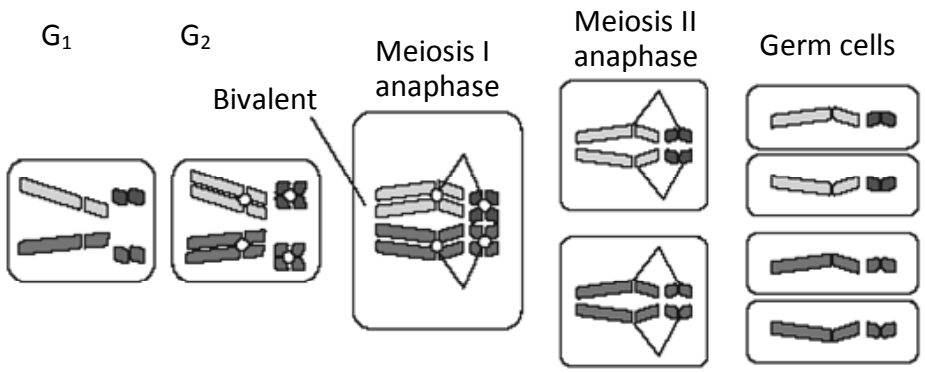
(1) 3 : 5 (2) 9 : 7 (3) 2 : 6 (4) 4 : 4

5. If mother has blood group AB, father has A group then Which of the following blood group will not be found in the offspring?

(1) A (2) B (3) AB (4) O

12. CHROMOSOMAL THEORY OF INHERITANCE

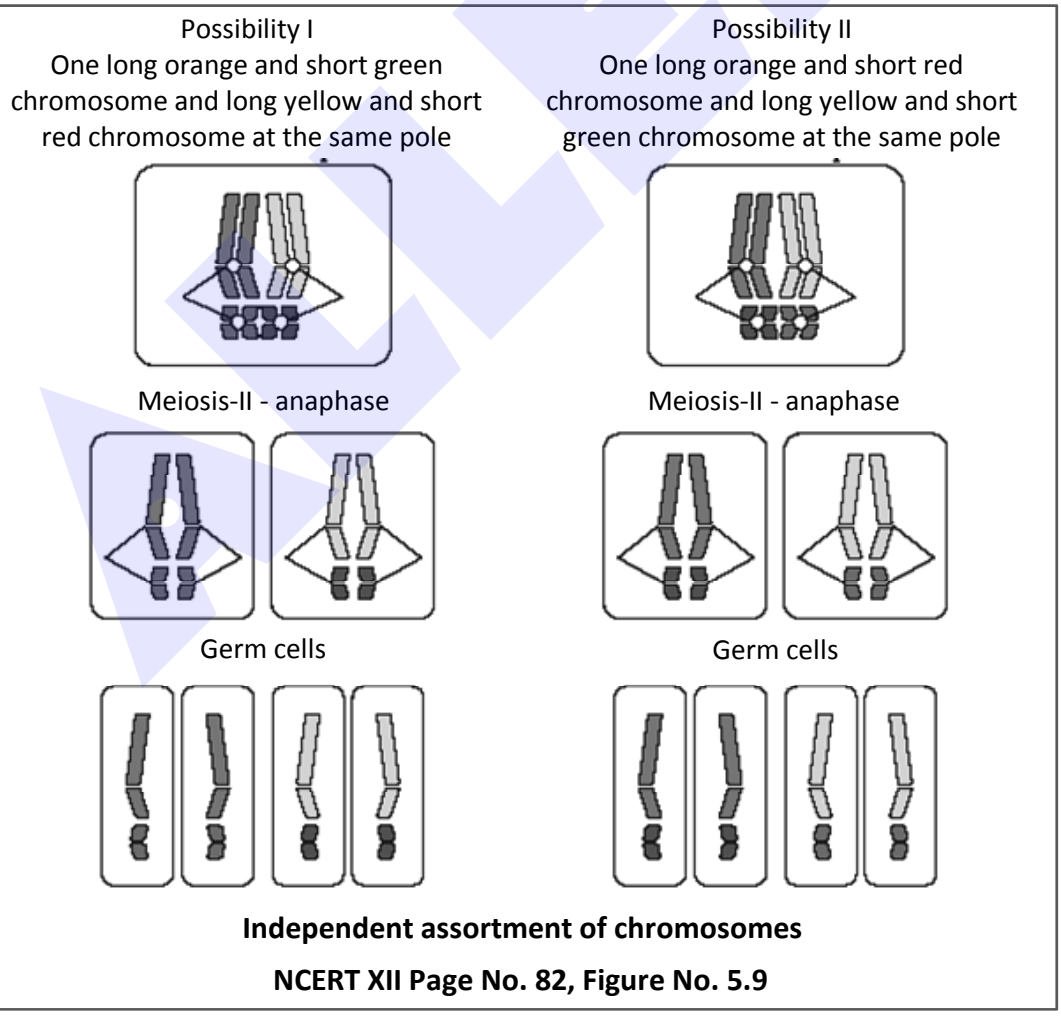
- This theory was proposed by **Walter Sutton** and **Theodor Boveri** (1902).
- In 1900, three Scientists (de Vries, Correns and von Tschermak) independently rediscovered Mendel's results on the inheritance of characters.
- Also, by this time due to advancements in microscopy that were taking place, 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 called **chromosomes** (*colored bodies*, as they were visualised by staining).
- By 1902, the chromosome movement during meiosis had been worked out. **Walter Sutton and Theodor Boveri noted that the behaviour of chromosomes was parallel to the behaviour of genes and used chromosome movement to explain Mendel's laws.**
- The important things to remember are that chromosomes as well as genes occur in pairs. The two alleles of a gene pair are located on homologous site on homologous chromosomes.



Meiosis and germ cell formation in a cell with four chromosomes. You can see how chromosomes segregate when germ cells are formed

NCERT XII Page No. 81, Figure No. 5.8

- During metaphase of meiosis I, the two chromosome pairs can align at the metaphase plate independently of each other. To understand this, compare the chromosomes of four different colour in the left and right columns. In the left column (Possibility I) orange and green is segregating together. But in the right hand column (Possibility II) the orange chromosome is segregating with the red chromosomes.



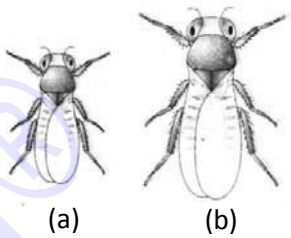
Independent assortment of chromosomes

NCERT XII Page No. 82, Figure No. 5.9

A Comparison between the behaviour of Chromosomes and Genes

A (Genes)	B (Chromosomes)
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

NCERT XII Page No. 82, Table No. 5.3



(a) (b)

Drosophila melanogaster
(a) Male (b) Female

NCERT XII Page No. 83, Figure No. 5.10

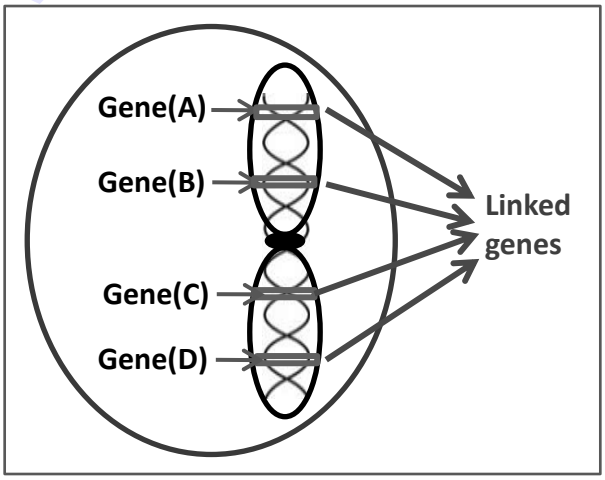
- Sutton and Boveri argued that the pairing and separation of a pair of chromosomes would lead to the segregation of a pair of factors they carried. Sutton united the knowledge of chromosomal segregation with Mendelian principles and called it the **chromosomal theory of inheritance.**
- Following this synthesis of ideas, experimental verification of the chromosomal theory of inheritance by Thomas Hunt Morgan and his colleagues, led to discovering the basis for the variation that sexual reproduction produced. Morgan worked with the tiny fruit files,

Drosophila melanogaster, which were found very suitable for such studies as :-

- They could be grown on simple synthetic medium in the laboratory.
- They complete their life cycle in about two weeks.
- A single mating could produce a large number of progeny flies.
- There was a clear differentiation of the sexes – the male and female flies are easily distinguishable.
- It has many types of hereditary variations that can be seen with low power microscopes.

13. LINKAGE AND RECOMBINATION

- Exception to Mendel's law of independent assortment.
- The physical association between genes present on a chromosome and their tendency to be inherited together is called linkage.
- Collective inheritance of characters is due to linkage.

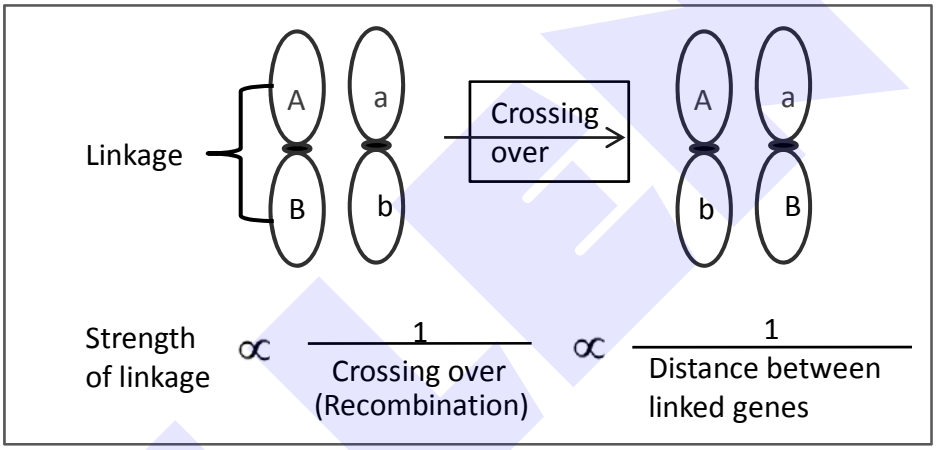


- Linkage first time observed by Bateson and Punnett.
- Linkage term and detail study by T.H. Morgan.
- He performed experiments on *Drosophila melanogaster* (fruit fly) and gave theory of linkage.

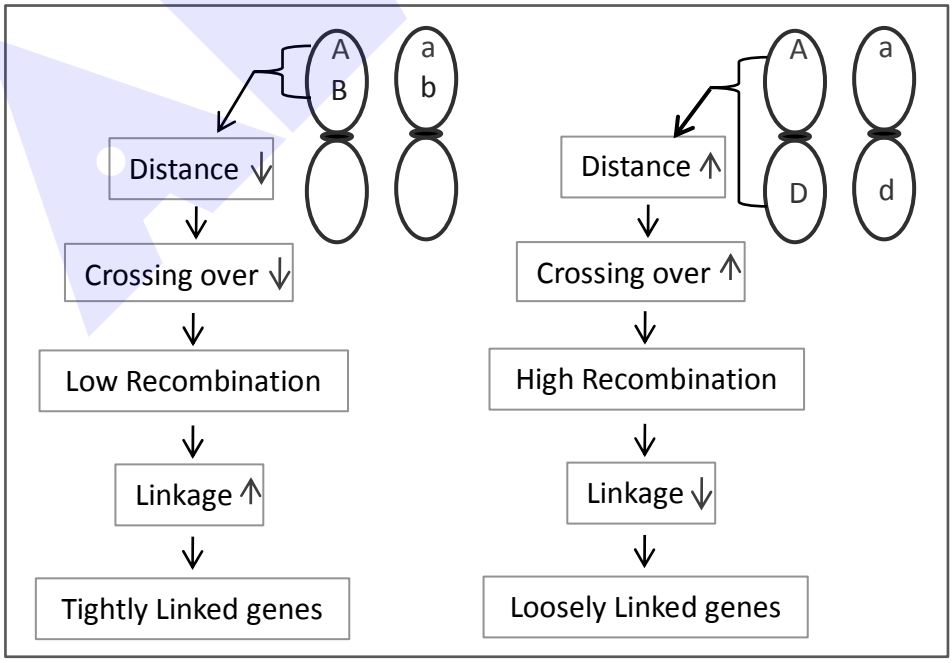
(1) THEORY OF LINKAGE

- According to this theory :-
  - (a) Genes showing linkage are called linked genes
  - (b) Linked genes are nonallelic
  - (c) Linked genes are present on same chromosome
  - (d) Linked genes can be separated by crossing over

    Crossing over :- exchange of chromosomal segments between non - sister chromatids of homologous chromosomes



(A) Factors affecting crossing over (C.O) :-



Factors	Crossing over	linkage
Distance ↑	↑	↓
Euchromatin	↑	↓
Heterochromatin	↓	↑
Age ↑	↓	↑
Sex → Female	↑	↓
Sex → Male	↓	↑

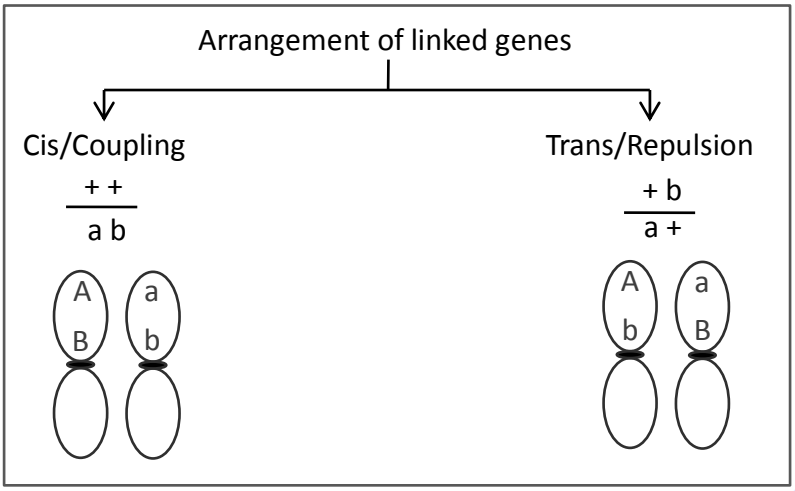
(B) Linkage Group :-

- All the genes which are located on one pair of homologous chromosome form one linkage group. Genes which are located on homologous chromosomes are inherited together so we consider them as one linkage group.
- Number of Linkage groups = Number of homologous chromosome pairs or Haploid no. of chromosomes.

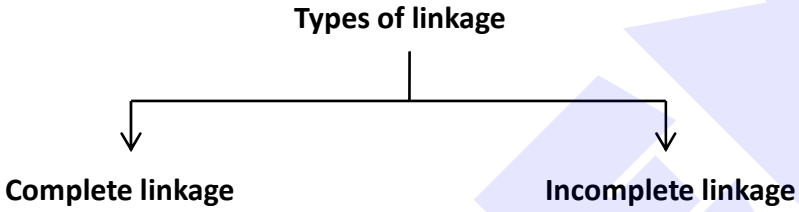
Organisms	2n	n	Pairs	Linkage groups
Pea	14	7	7	7
Maize	20	10	10	10
Onion	16	8	8	8
Drosophila	8	4	4	4
Human	46	23	23	23
Bacteria				1

Human → Male = 24 linkage groups (XY)
Human → Female = 23 linkage groups (XX)

(2) ARRANGEMENT OF LINKED GENES ON CHROMOSOMES



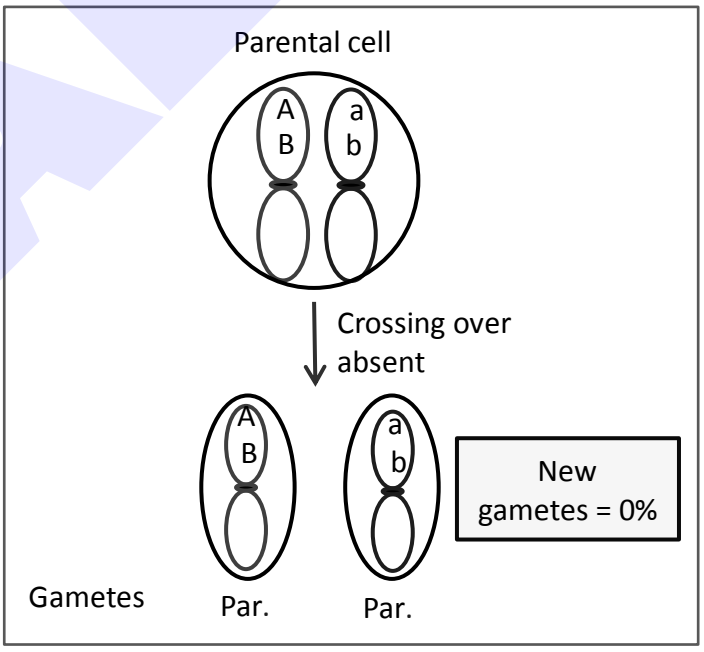
(3) TYPES OF LINKAGE



(1) Complete linkage :-

- Very rare
- In this linkage, crossing over does not occur between the genes

Linkage :- 100%
Crossing over :- 0%
New combination :- 0 %
Parental combination :- 100%
E.g. :- Few genes of male Drosophila and female Silkworm



- Complete linkage behave just like monohybrid cross.

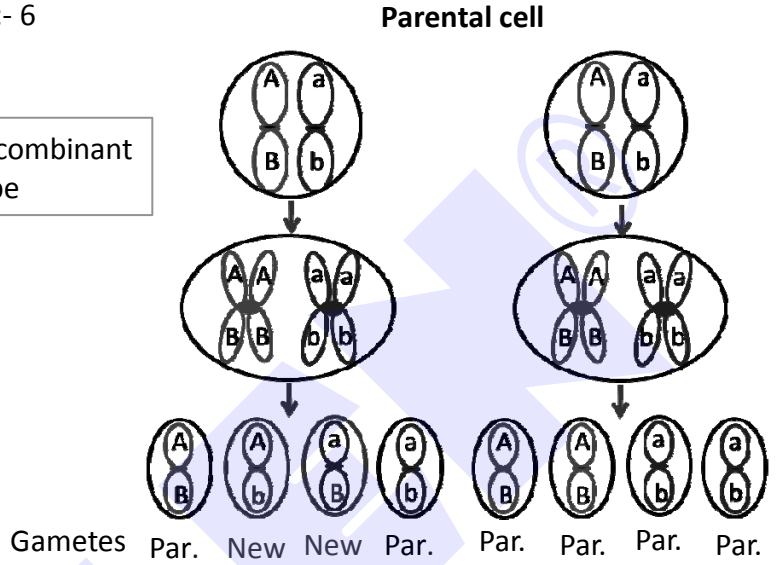
(2) Incomplete linkage :-

- Most common type of linkage.
- In this linkage crossing over occur between the genes.
- So new combinations are formed but parental combinations are greater than new combinations.

Parental type gametes :- 6

New type gametes :- 2

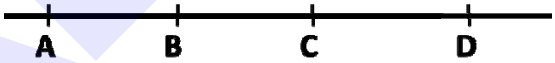
Parental type > Recombinant type



- Maximum frequency of recombination is 50%.
- It happens when crossing over occur in all cells with respect to desired genes.

Genetic map/Linkage map/chromosomal map –

- It is a straight line in which different genes are linearly arranged in a specific sequence and separated by some distance.



- Distance $\propto$ % of recombination
- 1 % recombination = 1 map unit distance (1 mu)
  or
  1 centi Morgan (1 cM)
- It is based on results of test cross
- Max. recombination $\rightarrow$ upto 50%

  Recombination Frequency or Cross over value [COV]

$$= \frac{\text{No. of recombinant offsprings}}{\text{Total no. of offsprings}} \times 100$$

- Alfred Sturtevant prepared 1st chromosomal map of Drosophila by use of recombination frequency.

Chromosomal map provides information about :-

1. Sequence of different linked genes on a chromosome.
2. Distance between the two linked genes.

MORGAN'S EXPERIMENT :-

Cross - A

1st character – Body Colour
- Brown → y^+ (wild)
- Yellow → y (mutant)

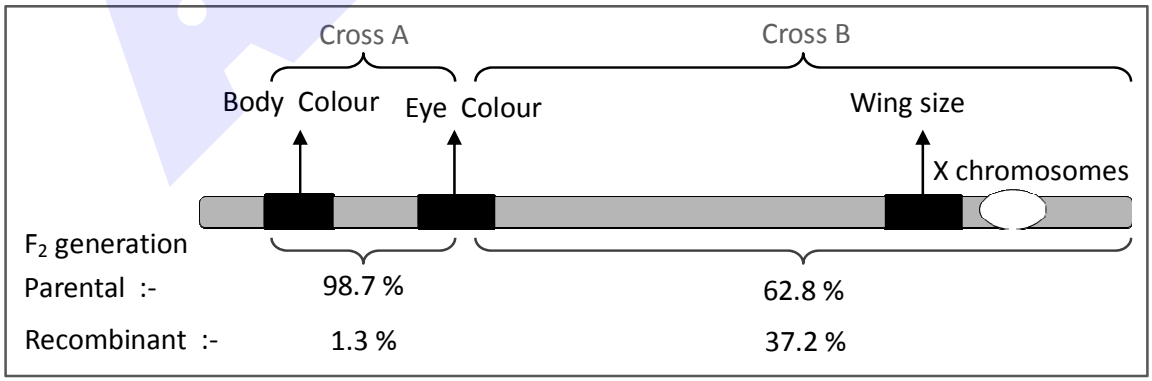
2nd character – Eye Colour
- Red → w^+ (wild)
- White → w (mutant)

Cross - B

1st character – Eye Colour
- Red → w^+ (wild)
- White → w (mutant)

2nd character – Wing size
- Normal → m^+ (wild)
- Miniature → m (mutant)

- Genes of all these characters are located on X-chromosome



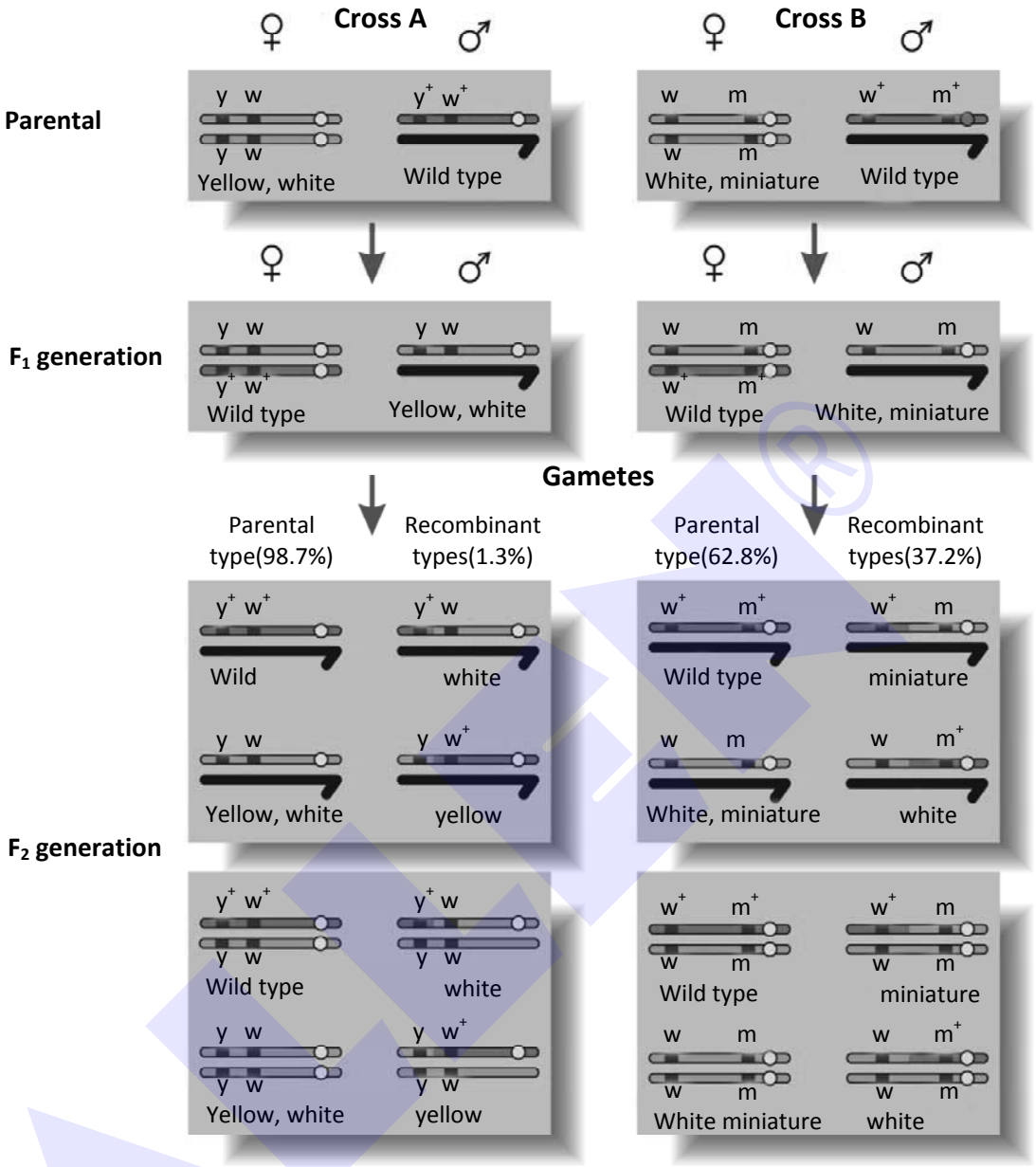


Figure : Results of two dihybrid crosses conducted by Morgan. Cross A shows crossing between gene y and w; Cross B shows crossing between genes w and m. Here dominant wild type alleles are presented with (+) sign in superscript. Note : The strength of linkage between y and w is higher than w and m.

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14. SEX LINKAGE

- Genes that are located on sex chromosome are called sex linked genes and their inheritance is called sex linked inheritance.
- Sex linkage was discovered by Morgan.
- He studied eye colour of *Drosophila* and performed reciprocal and test cross and found that gene of eye colour is located on X chromosome.

Examples :-

Eye Colour in *Drosophila* :-

If a red eye colour gene is represented as '+' and white eyed colour represented as 'w', then on this basis, different types of genotype are found in *Drosophila*.

Eye colour of drosophila

Red X^{w+} or X^{+} White X^{w}

Genotype :- X^{+} X^{w}

♀

$X^{+}X^{+}$ = Homozygous red eyed

$X^{+}X^{w}$ = Heterozygous red eyed

$X^{w}X^{w}$ = Homozygous white eyed

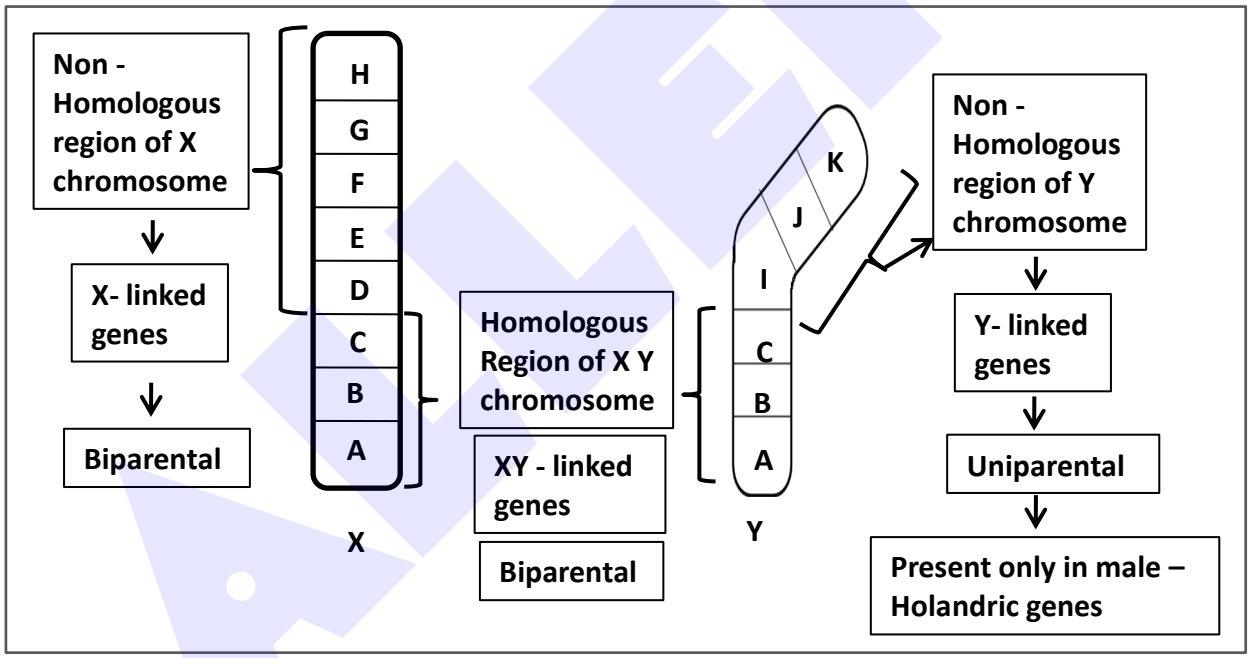
♂

$X^{+}Y$ = Hemizygous red eyed

$X^{w}Y$ = Hemizygous white eyed

It is clear from above genotypes that female is either homozygous or heterozygous for eye colour. But, for eye colour, male is always hemizygous.

Sex linkage in human being :-



Y-LINKAGE

- Genes present on non homologous region of Y chromosomes.
- Uniparental
- Present only in male (Holandric gene).

Example :

(i) SRY (Sex determining region on Y- Chr.) gene / TDF (Testis determining factor) gene :-

- Synthesises a protein- TDF.

(ii) Hypertrichosis – Excessive hairs on ear pinna.

X-LINKAGE

- Genes present on non homologus region of X chromosomes.
- Biparental

TWO TYPES

X – linked Recessive	**X – linked Dominant**
(a) Glucose -6- phosphate dehydrogenase deficiency (G6PD)	(a) Pseudorickets (Vitamin D resistant rickets)
(b) DMD(Duchenne muscular dystrophy) • Due to non- synthesis of dystrophin protein. • Gene for dystrophin synthesis is largest gene of human (2.4 million bps).	(b) Defective enamels of teeth
(c) Diabetes insipidus	
(d) Haemophilia	
(e) Colourblindness	

(d) Haemophilia :-

- X- linked recessive disease.
- Blood clotting time is delayed due to the absence of some blood clotting factor and person die due to excessive bleeding.
- Blood clotting time → Normal person → 1 – 8 min.
  Haemophilic person → 30 min. – 24 hr.

Genotype :-

Haemophilia

X^+ — Wild allele (Normal)

X^h — Mutant allele (Haemophilic)

♀

X^+X^+ = Normal

X^+X^h = Normal but carrier

X^hX^h = Haemophilic

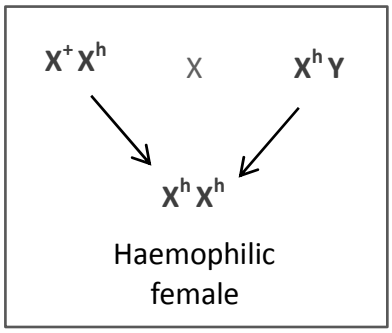
♂

X^+Y = Normal

X^hY = Haemophilic

- Haemophilic female is very rare as :-

  Mother of such female has to be at least carrier and father should be haemophilic (unviable in the later stage of life)



1. Haemophilia – A / Royal disease :-

- Most common type of haemophilia.
- First seen in royal family of England/ Queen Victoria family. She was carrier of this disease.
- Due to absence of blood clotting factor VIII – Anti-Haemophilic Globulin (AHG).

2. Haemophilia – B / Christmas disease :-

- Due to absence of blood clotting factor IX – Christmas factor/Plasma thromboplastin component.

3. Haemophilia – C :-

- Autosomal recessive disease
- Due to absence of blood clotting factor XI – Plasma Thromboplastin Antecedent.

(e) Colour Blindness :-

- X- linked (sex – linked) recessive disease.
- Colourblind person is unable to differentiate some basic colours like red and green due to defect in cone cells.

  Types:-

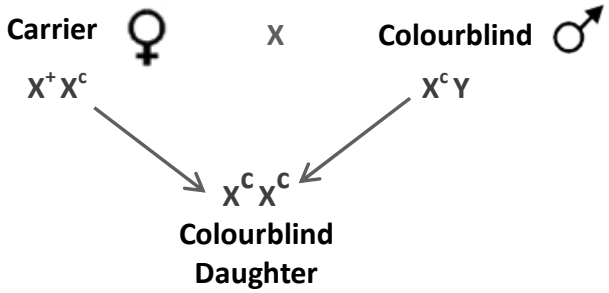
  1) Protanopia – Red colourblindness
  2) Deuteranopia – Green colourblindness

Genotype :-

COLOUR BLINDNESS

X^+	X^C
Wild allele (Normal)	Mutant allele (colorblind)
♀	♂
X^+X^+ = Normal	X^+Y = Normal
X^+X^C = Normal but carrier	X^C = colourblind
X^CX^C = colourblind	

- Colour blindness is checked by :- Ishihara chart.
- It is not a lethal disease, so found in both males and females.
- This disease is more common in males due to hemizygous condition.
- It occurs in about 8% of males and about 0.4% of females.
- A daughter will not normally be colourblind unless her mother is at least carrier and father is colourblind.



Types of Inheritance of sex linked characters :-

(1) CRISS CROSS INHERITANCE (MORGAN)

- In criss-cross inheritance male or female parent transfer an X-linked character to grandson or grand daughter through the offspring of opposite sex.

(A) Diagenic (Diagynic) :- Inheritance in which characters are inherited from father to the daughter and from daughter to grandson.

Father → daughter → grand son.

(B) Diandric :- Inheritance in which characters are inherited from mother to the son and from son to grand daughter. Mother → Son → Grand-daughter.

(2) NON CRISS-CROSS INHERITANCE

- In this inheritance male or female parent transfer sex linked character to grand son or grand daughter through the offspring of same sex.

(A) Hologenic (Hologynic) :- Mother → Daughter → Grand-daughter (female to female)

(B) Holandric :- Father → Son → Grand-son (male to male)

15. SEX-LIMITED CHARACTERS

- These characters are present in one sex and absent in another sex. But their genes are present in both the sexes and their expression is depend on sex hormone.
- Example :- Secondary sexual characters → these genes are **located on the autosomes** and these genes are present in both male and female, but effect of these are dependent upon presence or absence of sex-hormones.
- **Genes of beard-moustache** express their effects only in the presence of male hormone - testosterone.

16. SEX INFLUENCED CHARACTERS

- Genes of these characters are also **present on autosomes** but they are influenced differently in male and female. In heterozygous condition their effect is different in both the sexes.
- Example :- **Pattern baldness** : Gene of pattern baldness is dominant (B).

Genotype	Male	Female
BB	Baldness present	Baldness present
bb	Baldness absent	Baldness absent
Bb	Baldness present	Baldness absent

- Gene Bb shows partiality in male and female, Baldness is found in male due to effect of this gene, but baldness is absent in female with this genotype.

17. SEX DETERMINATION

- Establishment of sex through differential development in an individual at an early stage of life, is called sex determination.
- There are different methods for sex determination in organisms like allosomic sex determination, haplodiploidy, genic balance etc.

(1) SEX DETERMINATION ON THE BASIS OF FERTILIZATION

(A) Progamic - Sex is determined before fertilization.

eg. - drone in honey bee

(B) Syngamic - Sex is determined during fertilization.

eg. - most of the plants & animals

(C) Epigamic - Sex is determined after fertilization.

eg. - Crocodile, Turtle

(2) MECHANISM OF SEX DETERMINATION

(A) Allosomic Determination of Sex –

Chromosomes are of two types -

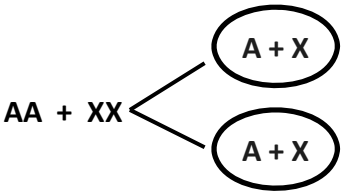
- Autosomes or somatic chromosomes :-

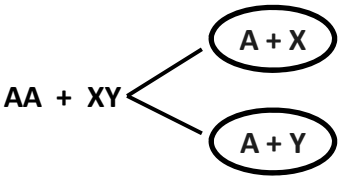
  These regulate somatic characters.

- Allosomes or Heterosomes or Sex chromosomes :-

- These chromosomes are associated with sex determination.
- X-Chromosome was discovered by ***"Henking"*** (1891) and called it 'x-body'.

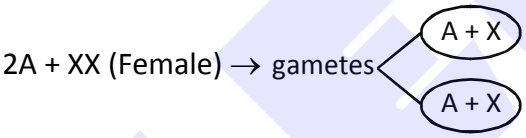
Homogametic Parent	Heterogametic Parent
➢ Both sex chromosomes are similar and produce one type of gamete. ➢ Ex – Human female	➢ Both sex chromosomes are dissimilar and produce 2 types of gametes. ➢ Decide sex of the offspring. ➢ Ex – Human male





(i) **XX - XY type or Lygaeus type :-** This type of sex determination was first observed by ***Wilson*** & ***Stevens*** in Lygaeus insect. It is of two -

(a) XX female and XY male :- In this type of sex determination female is Homogametic i.e produces only one type of gametes



Male is heterogametic (male produces two types of gametes)

eg. Human, Drosophila and many mammals

(b) ZW female and ZZ male :- In this type of sex determination female is Heterogametic i.e produces two types of gametes and male individual is homogametic i.e produces one type of gametes.

It is found in most of the **birds** (Fowl)**.**

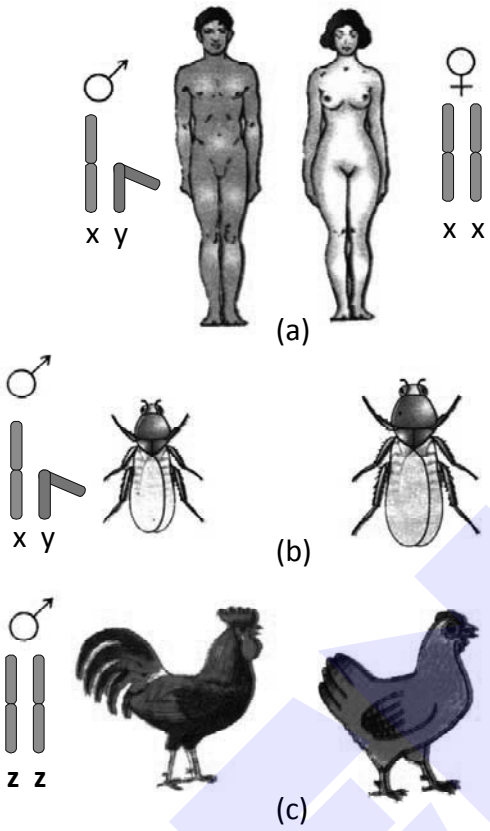
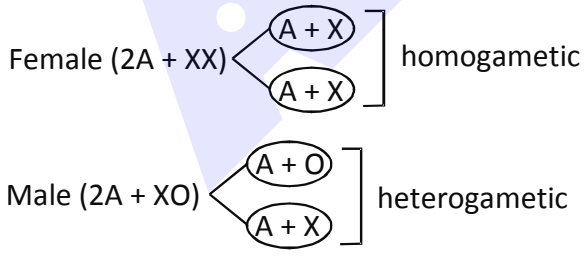
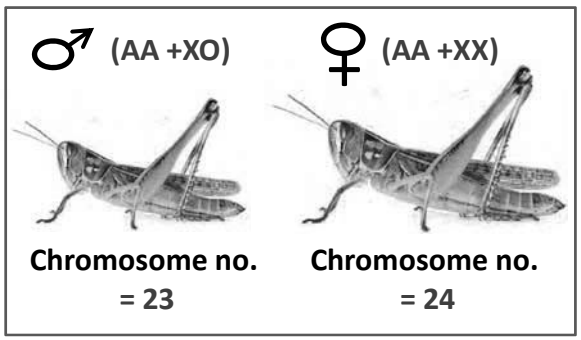


Figure : Determination of sex by chromosomal differences: (a,b) Both in humans and in *Drosophila*, the female has a pair of XX chromosomes (homogametic) and the male XY (heterogametic) composition; (c) In many birds, female has a pair of dissimilar chromosomes ZW and male has two similar ZZ chromosomes

NCERT XII Page No. 86, Figure No. 5.12

(ii) XX female and XO male or "Protenor type" :- In this type of sex determination there is deficiency of one chromosome in male. In this type, female is homogametic and male is heterogametic.





Example :-

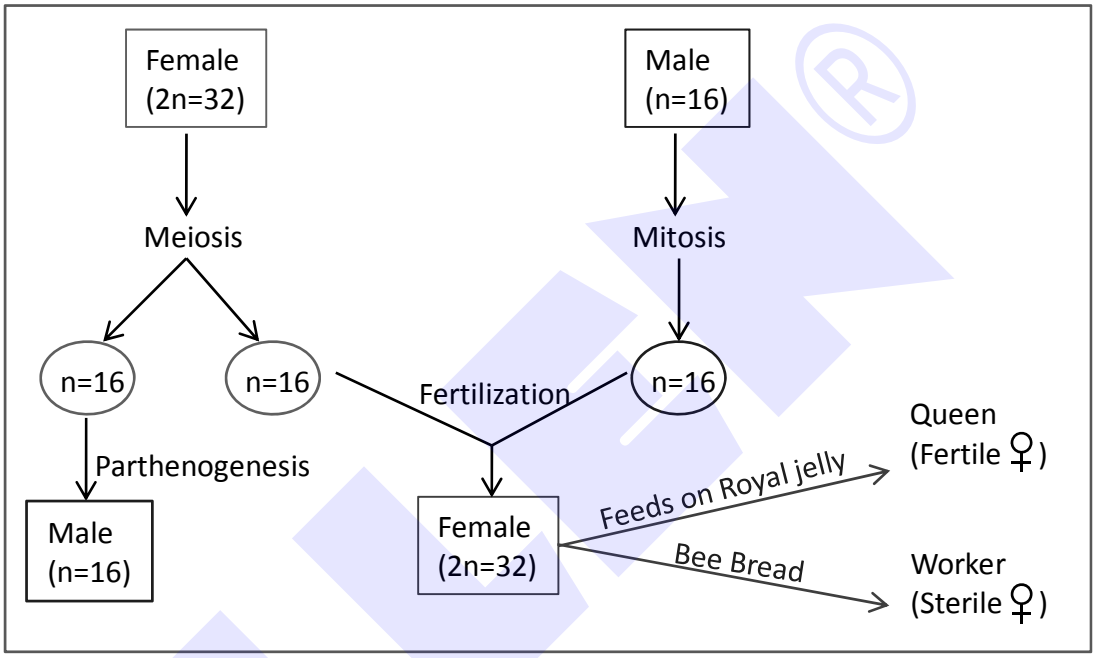
Most of the insects like :

– Grass hopper

– Cockroach

(B) Haploid - Diploid Mechanism (Sex Determination in Honey Bee) –

- Sex determination occurs on the basis of number of sets of chromosomes.
- Diploid (Two sets of chromosome) → Female
- Haploid (One set of chromosome) → Male
- In honey bee, male individual (Drone) develops from unfertilized eggs (Haploid). Male develops by parthenogenesis.
- Queen and worker bees (females) develop from diploid eggs i.e. fertilized egg.



- An offspring formed from the union of a sperm and an egg develops as a female (queen or worker), and an unfertilized 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 has special characteristic features such as the males produce sperms by mitosis, they do not have father and thus cannot have sons, but have a grandfather and can have grandsons.

(C) Genic Balance Theory :-

- **C.B. Bridges** proposed genic balance theory for sex determination in *Drosophila*.
- According to Bridges in *Drosophila* Y-chromosome is heterochromatic so it is not active in sex determination.

- In *Drosophila* sex determination takes place by sex index ratio.

$$\text{Sex Index Ratio} = \frac{\text{No. of X – chromosomes (X)}}{\text{No. of sets of autosome (A)}}$$

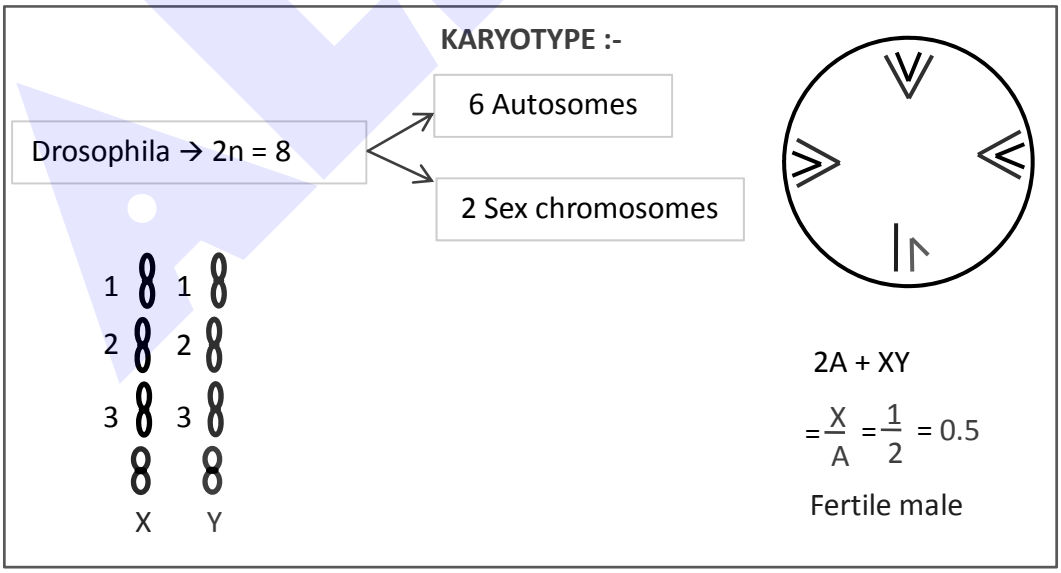
- In *Drosophila* gene of femaleness is located on x-chromosome and gene of maleness is located on autosome.
- Gene of male fertility is located on y-chromosome and in Drosophila, y-chromosome plays additional role in spermatogenesis so y-chromosome is essential for the production of fertile male.
- Sex index ratio (a) $\frac{X}{A} = 1 \rightarrow$ female (2A + XX)

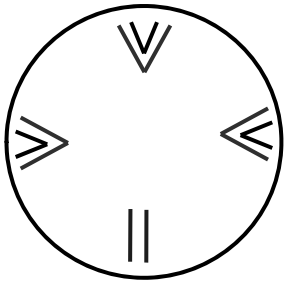
(b) $\frac{X}{A} = 0.5 \rightarrow$ male < (2A + XY) = Fertile male; (2A + XO) = Sterile male

(c) $\frac{X}{A} = 1.5 \rightarrow$ Super female or meta female (sterile) (2A + XXX)

(d) $\frac{X}{A}$ = less than 0.5 $\rightarrow$ Super male or meta male (Sterile) (3A + XY)

(e) $\frac{X}{A}$ = In between 0.5 and 1 $\rightarrow$ Intersex (Sterile) (3A+XX)

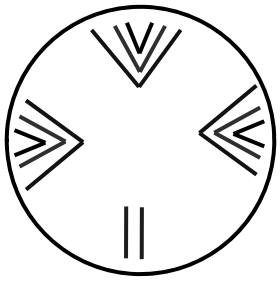




2A + XX

$= \frac{X}{A} = \frac{2}{2} = 1$

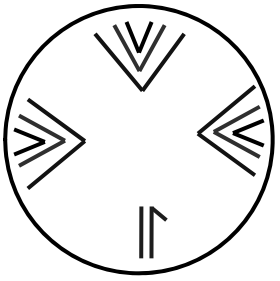
Fertile female



3A + XX

$= \frac{X}{A} = \frac{2}{3} = 0.66$

Inter sex



3A + XY

$= \frac{X}{A} = \frac{1}{3} = 0.33$

Super male

BEGINNER'S BOX — CHROMOSOMAL THEORY OF INHERITANCE TO SEX DETERMINATION

1. An exception to the law of independent assortment is :-

(1) Dominance (2) Incomplete dominance

(3) Segregation (4) Linkage

2. Experimental proof for sex-linked gene was given by :-

(1) Morgan (2) Muller

(3) Mendel (4) Johannsen

3. X-linked recessive gene is easily expressed in :-

(1) male

(2) female

(3) equal in male and female

(4) not easily expressed

4. Maize has 10 pairs of chromosomes. How many linkage groups are present.

(1) 05 (2) 10 (3) 20 (4) 40

5. The mechanism of sex determination in birds shows :-

(1) Male heterogamety

(2) Both heterogametic

(3) Female heterogamety

(4) Both homogametic

18. HUMAN GENETICS

- Study or analysis of genetics characters in human is called human genetics.

Eugenics :-

- Improvement of human beings by applying principles of genetics.
- Father of eugenics – Francis Galton.

Examples of some autosomal characters in human

Character	Dominant	Recessive
Eye colour	Brown / Black	Blue
Ear lobes	Free	Fused
Hair	Curly hair	Straight
Cheek	Dimple cheek	Normal
Rolling of tongue	Roller	Non Roller
Rh Factor	Rh^+	Rh^-
PTC (Phenyl thiocarbamide) taster	Taster	Non Taster
Skin pigmentation	Normal	Albino

Genetic disorders

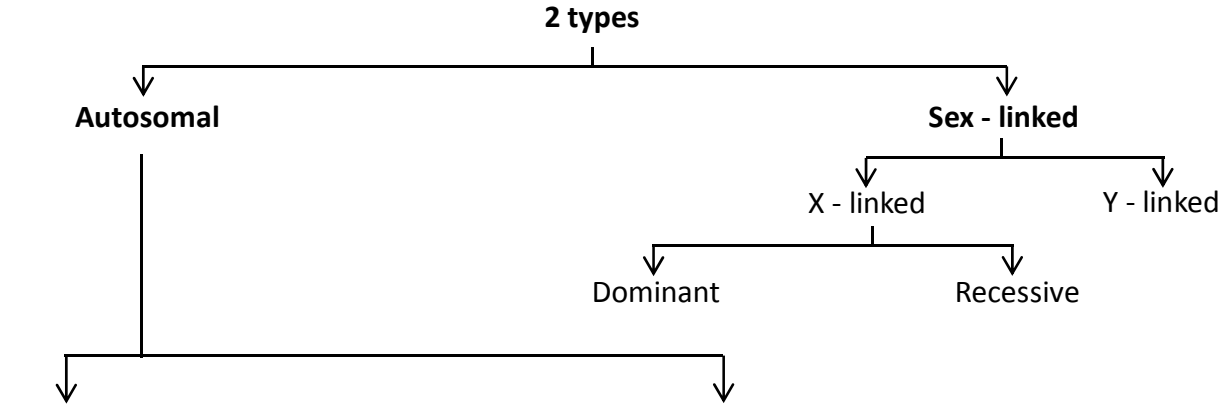
Chromosomal disorder
- Change in no. or structure of chromosomes
- Studied by karyotype analysis

Examples :-
- Down syndrome – 21st chr.
- Klinefelter syndrome – (XXY) – Male
- Turner syndrome – (XO) – Female

Mendelian disorder

Mendelian disorder :

- Change in structure and function of gene
- Studied by pedigree analysis



Dominant
AA / Aa → Diseased
aa → Normal
Ex-
- Huntington's chorea
- Myotonic dystrophy
- Polydactyly

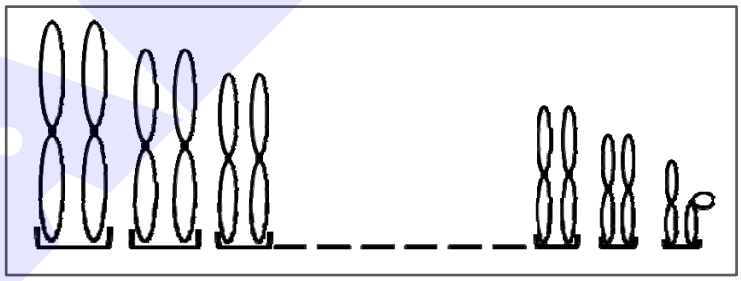
Recessive
AA → Normal
Aa → Normal (Carrier)
aa → Diseased
Ex-
- Sickle cell anaemia
- Thalassemia
- Phenylketonuria
- Alkaptonuria
- Albinism
- Cystic fibrosis

Indirect methods to study human genetics :-

1) Pedigree analysis

2) Population genetics

3) Karyotype analysis



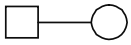
19. PEDIGREE ANALYSIS

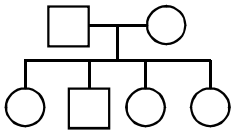
Pedigree = Family tree

- It is a record of some genetic characters or diseases for two or more generation in a family, which is represent by some specific symbols.

Symbol used in Pedigree :

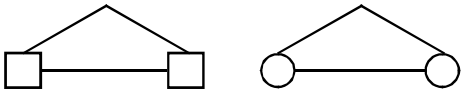
1. □ Normal Male
2. ◯ Normal Female

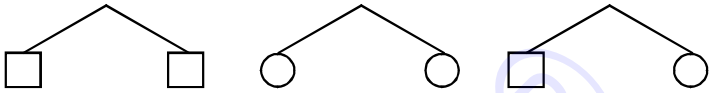
3.  Mating (marriage)

4.  The siblings are indicated in chronological order of birth

5.  Sex unspecified

6. Twins

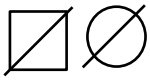
If monozygotic 

If dizygotic 

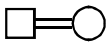
7.  Affected female and male individuals

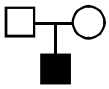
8.  Heterozygous for autosomal recessive character

9.  Carrier female of sex linked recessive character

10.  Death of individual

11.  Abortion or still birth (sex unspecified)

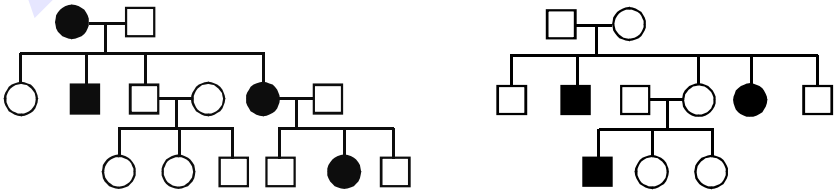
12.  Consanguineous marriage/mating

13.  Parent with male child affected with disease

14.  Five unaffected offsprings

- Pedigree analysis provides valuable informations regarding genetical make up of human beings.
- If any genetic disease is occuring in a family, then pedigree analysis provides guidance to forthcoming parents about their future progenies, for example- polydactyly in humans.

Examples :-



(a) (b)

Representative pedigree analysis of (a) Autosomal dominant trait (Myotonic Dystrophy)
(b) Autosomal recessive trait (Sickle Cell Anaemia)

20. POPULATION GENETICS

Study of gene frequency in a population is called population genetics.

- **Gene pool –** A gene pool is the sum total of all the genes and their alleles present in (reproductive gametes of) a population.
- **Gene-flow –** Migration of genes from one population to another population by cross fertilization.
- **Genetic load –** The existence of disadvantageous/harmful recessive alleles in heterozygous condition in a population is known as genetic load.
- **Gene frequency –** Gene frequency is defined as proportion of a given alleles out of total alleles of a gene in population.

$$\text{Frequency of a given allele} = \frac{\text{No. of given alleles}}{\text{Total no. of alleles}}$$

Ex. In a population of 100 individuals of MN blood group 50 MM, 20 MN, 30 NN. Find out the frequency of M & N.

MM – 50

MN – 20

NN – 30

Total M genes – $50 \times 2 + 20 = 120$

Total N genes – $30 \times 2 + 20 = 80$

Total genes– 200

Freq. of M gene $p = \frac{M}{M+N} = \frac{120}{200} = 0.6$

Freq. of N gene $q = \frac{N}{M+N} = \frac{80}{200} = 0.4$

$0.6 + 0.4 = 1$

$p + q = 1$

Hardy Weinberg Law

Proposed by

- G.H.Hardy (English mathematician) & W.Weinberg (German Physician).
- **According to this law :-**

  In an ideal population frequency of alleles remain constant generation after generation. Ideal population means, in this population :-
  - Random mating occurs
  - No mutation
  - No migration
  - No natural selection
  - Population is large

$\boxed{p + q = 1}$

Here ,

p = frequency of dominant allele

q = frequency of recessive allele

p + q = 1

Square on both side

$(p + q)^2 = (1)^2$

$\boxed{P^2 + 2pq + q^2 = 1}$

$p^2 + 2pq + q^2 = 1$

Here,

p^2 = frequency of homozygous dominant genotype/phenotype/character/organism.

2pq = frequency of heterozygous dominant genotype/phenotype/character/organism/carrier.

q^2 = frequency of homozygous recessive genotype/phenotype/character/organism.

p^2 + 2pq = frequency of dominant genotype/phenotype/character/organism

$p^2 + q^2$ = frequency of homozygous genotype/phenotype/character/organism

Q. In a random mating population, frequency of recessive phenotype is 0.09. What is the frequency of heterozygous genotype?

Sol. $q^2 = 0.09$

$q = 0.3$

$p = 1 - q$

$p = 1 - 0.3 \Rightarrow 0.7$

Frequency of heterozygote = $2pq = 2 \times 0.7 \times 0.3 = 0.42 = 42\%$

Q. In a random mating population of 1000 individuals , frequency of recessive phenotype is 0.16, then find out number of homozygous organisms ?

Sol. Recessive phenotype = 0.16

$q^2 = 0.16$

$q = 0.4$

$p + q = 1$

$p + 0.4 = 1$

$p = 0.6$

Homozygous organism = $p^2 + q^2$

$= (0.6)^2 + (0.4)^2$

$= 0.36 + 0.16$

$= 0.52 \times 1000$

$= 520$

BEGINNER'S BOX — HUMAN GENETICS TO POPULATION GENETICS

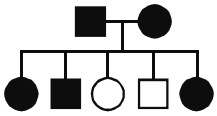
1. Which of the following symbols and its representation is correct :-

(1) □ = unaffected female (2) ◇ = affected male

(3) ■ = affected female (4) ● = affected female

2. The presence of recessive trait in a large population is found to be 16%. The frequency of dominant trait in that population is :-

(1) 0.84 (2) 0.42 (3) 0.56 (4) 0.96

3.  The pedigree shows

(1) Dominant inheritance

(2) Recessive inheritance

(3) Sex linked recessive inheritance

(4) Cytoplasmic inheritance

4. In a random mating population frequency of dominent allele is 0.7. What will be the frequency of homozygous dominant phenotype.

(1) 0.49 (2) 0.09 (3) 0.3 (4) 0.21

5. If a couple has four girls, the probability of fifth child being male, is

(1) 50% (2) 25% (3) 75% (4) 100%

BEGINNER'S BOX — ANSWERS KEY

INTRODUCTION TO RECIPROCAL CROSS

Que.	1	2	3	4	5	6	7	8	9	10
Ans.	**3**	**3**	**1**	**4**	**2**	**1**	**1**	**2**	**4**	**3**

GENE INTERACTION TO CYTOPLASMIC INHERITANCE

Que.	1	2	3	4	5
Ans.	**2**	**3**	**3**	**1**	**4**

CHROMOSOMAL THEORY OF INHERITANCE TO SEX DETERMINATION

Que.	1	2	3	4	5
Ans.	**4**	**1**	**1**	**2**	**3**

HUMAN GENETICS TO POPULATION GENETICS

Que.	1	2	3	4	5
Ans.	**4**	**1**	**1**	**1**	**1**

Parents characters → (Inheritance of characters) → Progeny

Mendelism

Mendel worked on – garden pea

Monohybrid

Genotype – 1:2:1
Phenotype – 3:1

Dihybrid

Genotype
1:2:2:4:1:2:1:2:1
Phenotype
9:3:3:1

Backcross

- **Outcross**
  - F_1 × Dominant parent
- **Test cross**
  - F_1 × Recessive parent

Reciprocal cross

- **Karyogene/ Autosome characters**
  - Result not changed
- **Cytoplasmic & Sex linked characters**
  - Result changed

Post mendelism

Gene interaction

Allelic

(1) Incomplete dominance
- F_1 different from both parents
  Genotype – 1:2:1
  Phenotype – 1:2:1

(2) Co-dominance
- In heterozygous condition both the alleles of a gene express for a particular character in F_1
  Genotype – 1:2:1
  Phenotype – 1:2:1

(3) Multiple alleles
- More than two alternate forms of same gene.
- Types of different possible genotype= $\frac{n(n+1)}{2}$

(4) Pleiotropic gene
- Gene which controls more than one character.

Non-allelic

(1) Complementry gene
- A pair of non-allelic genes are essential in dominant form to produce a character.
  Phenotype – 9:7

(2) Epistasis
- A gene prevents expression of another non-allelic gene
- Dominant epistasis
  Phenotype – 12:3:1

Polygenic Inheritance

- Inheritance of character in which one character is controlled by many genes
- Intensity of character depends upon number of dominant alleles.
- Types of phenotype- (2n+1)

Cytoplasmic Inheritance

- Inheritance of characters which are controlled by cyto gene or plasma gene or cytoplasm.
- Inheritance occurs only through female.

Linkage → Physical association of genes on a chromosome

- Linkage group
  - All the genes which are located on one pair of homologous chromosomes form one linkage group.
  - Number of linkage groups = Haploid no. of chromosomes
  - Linked genes can separate by crossing over.
  - Linkage $\propto \frac{1}{\text{Distance between genes}} \propto \frac{1}{\text{Crossing over}}$

Arrangement of linked gene

Cis | Trans

Types of Linkage

Complete	Incomplete
• Always parental combinations appear	• New combinations also appear

- **Genetic map / Linkage map**
  - Genes are linearly arranged according to % of recombination between them.
  - Helpful in study of genome.
- **Sex linkage** → X linkage
  - Eye colour in Drosophila

  Y linkage
  - TDF/Sry gene
  - Hypertrichosis
  - Genes located on Y chromosomes are known as holandric genes
- **Types of inheritance of sex linked character**

Criss cross | Non criss cross

Diagenic | Diandric | Hologenic | Holandric

- **Sex limited character**
  - Genes located on autosomes
  - Effect depends upon presence or absence of sex hormone.
    e.g. Gene of beard-moustache
- **Sex influenced character**
  - Genes present on autosomes
  - Influenced differently in male and female
    e.g. Baldness

- **Sex determination** → On the basis of fertilization
  - Progamic ↓ Before fertilization
  - Syngamic ↓ During fertilization
  - Epigamic ↓ After fertilization

- **Mechanism of Sex determination**
  - Allosomic
    - XX–XY type e.g. Human
    - ZW–ZZ type e.g. Birds
    - XX–XO type e.g. grass hopper
  - Haploid diploid e.g. Honey bee, Wasp, Ants
  - Genic balance theory e.g. *Drosophila*

Genetic disorders

Chromosomal disorders
- Change in chromosomal number
- Studied by karyotype analysis

e.g. Turner is syndrome
Down is syndrome
Klinefelter is syndrome

Mendelian disorders
- Change in structure and function of gene
- Studied by pedigree analysis

Autosomal
- Dominant
  - Myotonic dystrophy
  - Polydactyly
- Recessive
  - Sickle cell anaemia
  - Thalassemia
  - Phenylketonuria
  - Alkaptonuria
  - Albinism
  - Cystic fibrosis

Sex linked
- X linked
  - Dominant
    - Pseudorickets
    - Defective enamels
  - Recessive
    - Haemophilia
    - Colour blindness
    - G-6-PD
    - DMD
- Y linked