# GENETICS

## PRINCIPLES OF INHERITANCE AND VARIATIONS

### INTRODUCTION

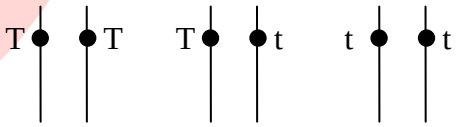
Genetics term was given by **W.Bateson.**
Genetics = Collective study of heredity & Variations.
**Heredity** = Transmission of genetic characters from parent to offsprings.
**Variation** = individuals of same species have some difference, these are called variation.

γ 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, lie Linkage, Sex linkage, Crossing over, Criss-cross inheritance, Linkage map on Drosophila.

γ **A.Garrod** = Father of human genetics & Biochemical genetics. Garrod Discovered fist human Metabolic genetic disorder which is called **alkaptonuria** (black urine disease).

### SOME GENETICAL TERMS

1. **Factors** :- Unit of heredity which is responsible for inheritance and appearance of characters. These factors were referred as genes by **Johannsen** (1909). Mendel used term **"element"** or **"factor"**. Morgan first used symbol to represent the **factor**. Dominant factors are represented by capital letter while recessive factor by small letter.
2. **Allele** :- Alternative forms of a gene which are located on same position [loci] on the homologous chromosome is called Allele. Term allele was coined by **Bateson**.




3. **Homozygous** :- A zygote is formed by fusion of two gametes having identical factors is called homozygous. **Ex.** TT, RR, tt
4. **Heterozygous :-** A zygote is formed by fusion of two gametes carrying different factors is called heterozygote (Tt, Rr) and individual developed from such zygote is called heterozygous. The term homozygous and heterozygous are coined by **Bateson**.
5. **Hemizygous** :- If individual contains only one gene of a pair then individual is said to be Hemizygous. Male individual is always Hemizygous for sex linked gene.
6. **Phenotype** :- It is the external and morphological appearance of an organism for a particual character.
7. **Genotype** :-The genetic constitution or genetic make-up of an organism for a particular character. Genotype & phenotype terms were coined by **Johannsen**.
8. **Phenocopy** :- If different genotypes are placed in different environmental conditions then they produce same phenotype. Then these genotypes are said to be Phenocopy of each other.
9. **Hybrid vigour/Heterosis** – Superiority of offsprings over it's parents is called as **Hybrid vigour** & it develops due to Heterozygosity.

- Hybrid vigour can be maintained for long time in vegetaively propagated crops.

- Hybrid vigour can be lost by invreeding (selfing) because inbreeding induces the Homozygosity in off springs. Loss of Hybrid vigour due to inbreeding, is called as **inbreeding depression**.

**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 1856-57. 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 "Experiment in plant Hybridization" was published in this journal.

Mendel was unable to get any popularity. No one understood of him. 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.

Rediscovery by three scientists independently.

1. **Carl Correns** - (Germany) - (Experiment on Maize)
2. **Hugo deVries** (Holland) (Experiment on Evening Primerose)
   He republished Mendel's results in 1901 in Flora magazine
3. **Erich von Tschermak Seysenegg**- (Austria) (Experiment on different flowering plants)

The credit of rediscovery of Mendelism goes to three scientists.

Correns gave two laws of Mendelism.

Law of Heredity /lnheritance/Mendelism

I$^{st}$ Law- Law of segregation.

II$^{nd}$ Law - 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 :**

**1.** Mendel studied the inheretance 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).

**2.** **Selection of Material-**

Selection of garden Pea plant is suitable for studies, which have the following advantages :

(i) 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.

(ii) It has many contrasting traits.

(iii) Natural5elf pollination is present in pea plant.

(iv) Cross pollination can be performed in it artificially so hybridization can be made possible.

(v) Pea plant is easy to cultivate.

(vi) Pea seeds are large. In addition to pea, Mendel worked on rajarna and hawk weed.

3. Mendel quantitatively analyse the inheritance of gyaljtatiye characters.

4. He maintained the statistical records of all the emertrnents.

5. His experiments had a large sampling size.

**Mendel's work :** Mendel studied 7 characters or 7 pairs of contrasting traits.

Actual data obtained by Mendel in $F_2$ progenies in garden pea

| S.No. | Character | Ch. No. | Dominant | Recessive | Ratio |
|---|---|---|---|---|---|
| 1. | Length of plant (Stem height) | $4^{th}$ | 787 (tall) | 277 (dwarf) | 2.84 : 1 |
| 2. | Flower position | $4^{th}$ | 651 (axial) | 207 (terminal) | 3.14 : 1 |
| 3. | Shape of pod | $4^{th}$ | 882 (inflated) | 299 (constricted) | 2.94 : 1 |
| 4. | Pod colour | $5^{th}$ | 428 (green) | 152 (yellow) | 2.82 : 1 |
| 5. | Seed shape | $7^{th}$ | 5.474 (round) | 1.850 (wrinkled) | 2.96 : 1 |
| 6. | Seed colour | $1^{st}$ | 6.022 (yellow) | 2.001 (green) | 3.01 : 1 |
| 7. | Flower colour | $1^{st}$ | 705 (violet) | 224 (white) | 3.15 : 1 |
| Average of all traits studied | | | | | 2.98 : (3 : 1) |

**Special Point :**

γ **S. Blixt** concluded that the genes studied by Mendel are located on four different pairs of chromosomes. These are chromosome $1^{st}$, $4^{th}$, $5^{th}$, $7^{th}$

- Two of the genes are on chromosome 1st and three are on chromosome 4th. These genes are located far apart on the chromosome except genes controlling plant height and pod shape.
- In Pea plant sead coat colour and Rower colour are regulated by same gene.
  Gene which controls more than one character is called as **pleiotropic gene.**
- Mendel obtained wrinkled seeds due to absence of **Starch Branching enzyme (SBE)**
  In Wrinkled seed free sugar is more in place of starch.

## Technique of Mendel

Mendel conducted such artificial pollination/cross pollination experiments using several true-breeding pea lines. A truebreeding line is one that. having undergone continuous self-pollination. shows the stable trait inheritance and expression for several generations: Mendel selected 14 true-breeding pea plant varieties, as pairs which were similar except for one character with contrasting traits.

He developed a technique **Emasculation and Bagging** for hybridization in plants.

Rowers of pea plant are bisexual. In this method one considered as male and another as female.

**Emasculation :** Removal of anther from a bisexual flower in immature stage is called Emasculation.

**Emasculation** is done to prevent self pollination.

**Bagging:** Emasculated flowers are covered by bags, this is called bagging.

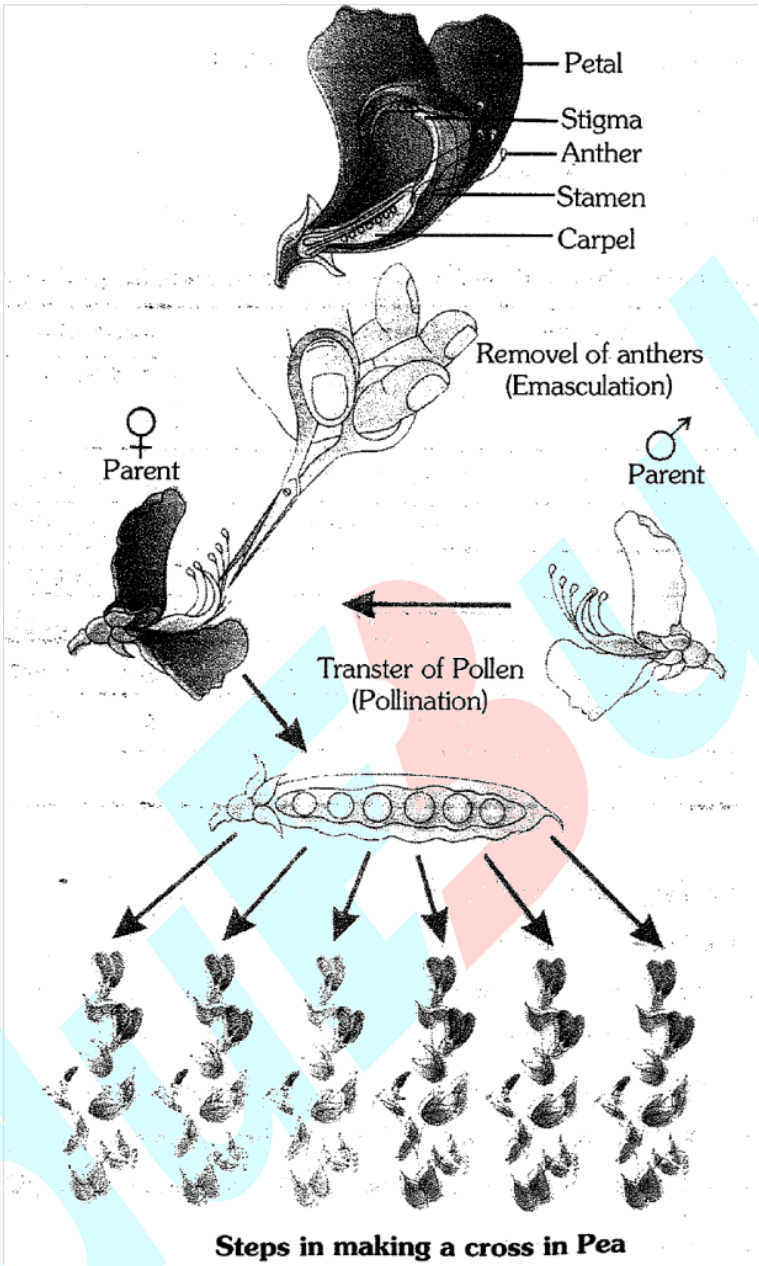
Bagging is only used to prevent undesirable cross pollination.

Mature pollen grains are collected from male plants and spread over emasculated flower.

Seeds are formed in the female flower after pollination.

The plants that are obtained from these seeds are called First Filial generation or $F_1$ generation according to Mendel.

The plants of $F_1$ generation are self pollinated & $F_2$ generation is produced.




**Steps in making a cross in Pea**

## MONOHYBRID CROSS

When we consider the inheritance of one character at a time in a cross this is called monohybrid cross. First of all, Mendel selected tall and dwarf plants.

**Parent** Tall (Pure) × Dwarf (Pure)

↓

**$F_1$-Generation** **All tall** (impure)

↓ **Self pollination**

| | **Tall** | | **Dwarf** (phenotypic ration or basic or Mendelian ratio) |
|---|---|---|---|
| **$F_2$-Generation** | 3 | : | 1 |

↓

1 tall (pure) | 2 tall (impure) | 1 Dwarf (Pure)

(selfing)↓ | (selfing)↓ | (selfing)↓

**$F_3$-Generation** All Tall | 3tall : 1 dwarf | All dwarf

**Checker board Method**

First time it was used by **Reginald. C. Punnett** (1875-1967)

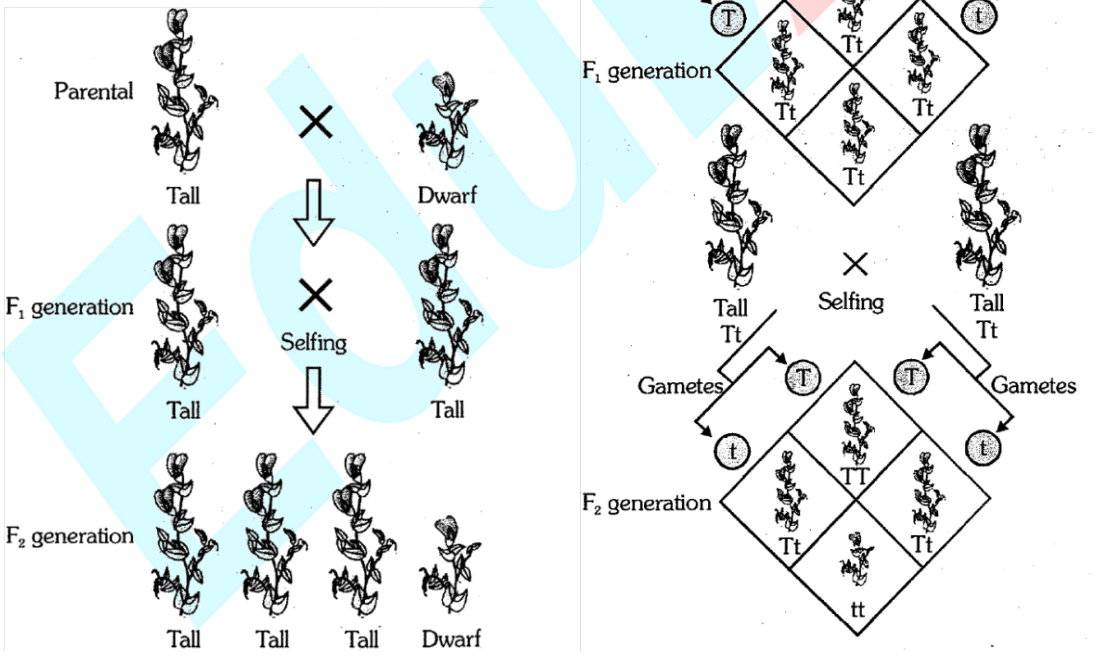
The representation of generations to analyse in the form of symbols of squares. Male gametes lie horizontally and female gametes lie vertically.

Phenotypic ratio : tall : draft

3 : 1

Genotypic ratio : TT : Tt : tt

1 : 2 : 1




**Conclusions (results) of Monohybrid Cross**

**I$^{st}$ Conclusion (Postulate of paired factors) :**

According to Mendel :-

1. Characters are controlled by discrete units called factors.
2. Factors occur in pairs.

**II$^{nd}$ Conclusion (Postulate of Dominance):**

This conclusion is based on $F_1$ - generation. When two different unit factors are present in single individual, only one unit factor is able to express itself and known as dominant unit factor. Another unit factor fails to express is the recessive factor. In the presence of dominant unit factor recessive unit factor can not express and it is known as conclusion of dominance.

| | | |
|---|---|---|
| Tall | × | Dwarf |
| TT | ↓ | tt |
| $F_1$-Generation | (Tt) | All tall |

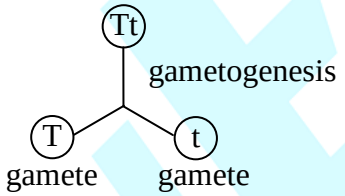
Law of dominance = I$^{st}$ Conclusion + II$^{nd}$ Conclusion.

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

**III$^{rd}$ Conclusion (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.

During gamete formation ; the unit factors 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 conclusiop. of purity of gametes or segregation.




γ There is no exception of Law of segregation. The segregation is essential during the meiotic division in all , sexually reproducing organisms. (Nondisjunction may be exception of this law).

**FORK UNE METHOD FOR GAMETES FORMATION**

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

AaBb = 4 types of gamete

$$A\begin{cases} B - AB = 1/4 = 25\% \\ b - Ab = 1/4 = 25\% \end{cases}$$

$$a\begin{cases} B - aB = 1/4 = 25\% \\ b - ab = 1/4 = 25\% \end{cases}$$

Type of gamete /phenotypic category= $2^n$

Type of genotype = $3^n$

Number of zygote produced by selfing of a genotype = $4^n$

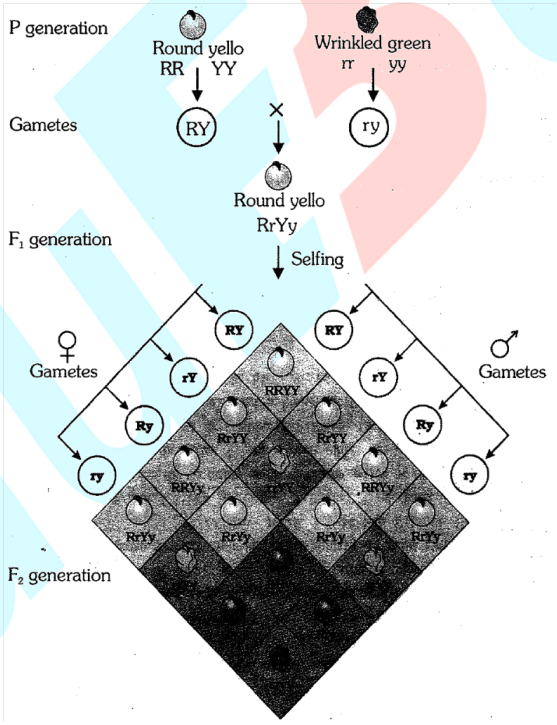
n = Number of hybrid character or heterozygous pair.

**DIHYBRID CROSS**

A cross in which there is study of inheritance of two pairs of contrasting traits.
Mendel wanted to observe the effect of one pair of heterozygous on other pair.
Mendel selected :-

**In garden Pea**
- Seed shape
  - Round [R]- Dominant
  - Wrinkled [r]- Recessive
- Seed colour
  - Round [Y]- Dominant
  - Wrinkled [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, there were in the ratio of 9 : 3 : 3 : 1. This ratio is known as dihybrid ratio.




**Phenotypic ratio :** round yellow : round green : wrinkled yellow : wrinkled green
9 3 3 1
Number of phenotype = 4

**Genotypic ratio :** RRYY RrYY RRYy RrYy rrYy rrYy RRyy Rryy rryy

1 2 2 4 1 2 1 2 1

Number of genotype = 9

| Parental combination | : | New combination |
|---|---|---|
| 9[Round yellow] + 1 [Green wrinkled] | : | 3[Round green] + 3 [Wrinkled yellow] |
| 10 | | 6 |

**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 trait [2 different characters] are combined in a hybrid, segregation of one pair of character is independent of the other pair of character."

This is known as **Conclusion of Independent Assortment.** It is based on F 2 -generation of dihybrid cross.

**Exception :** Linkage

## TRIHYBRID CROSS

A cross in which there is study of three pairs of contrasting traits.

RRYYTT × rryytt

↓

$F_1$ = RrYyTt

↓ Selfing

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

Genotype $3^3 \rightarrow$ 27 types

## BACK CROSS

A back cross is a cross in which F 1 individuals are crossed with any of their parents.

**(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 the any analysis can not possible in $F_1$ generation.

$F_1$ progeny (hybrid) Dominant parent

Tt × TT

↓

T TT

T Tt

All Tall

**(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% is dominant phenotypes and 50% is recessive phenotypes.

It can be represented in symbolic forms as follows.

$F_1$ progeny(hybrid) Recessive parent

Tt × tt

↓

T Tt Monohybrind test cross ratio = 1 : 1

T tt

**[B] Dihybrid Test Cross :-** The progeny is obtained from dihybrid test cross are four types and each of them is 25%

$F_1$ – Dihybrid Recessive parent

T t R r × t t r r

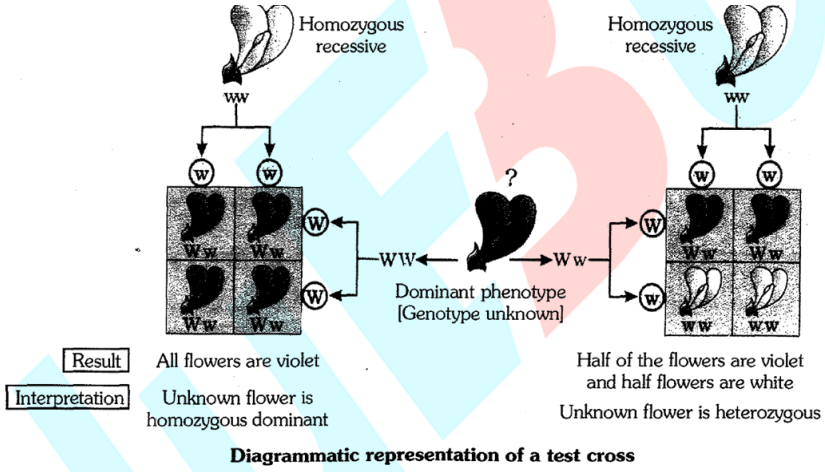
↓

| | RT | Tr | Rt | tr |
|---|---|---|---|---|
| tr | TtRr | Ttrr | ttRr | ttrr |

The ratio of Dihybrid test cross = 1: 1: 1: 1

**Conclusion:-** In test cross phenotypes and genotypes ratio are same.

**Test cross helps to find out the genotype of dominant individual.**




**Diagrammatic representation of a test cross**

## RECIPROCAL CROSS

When two parents are used in two experiments in such a way that in one experiment "A" is used as the female parent and "B" is used as the male parent, in the other experiment "A" will be used as the male parent and "B" as the female parent. such type of a set of two experiments is called Reciprocal cross.

Characters which are controlled by karyogene present on autosomes are not affected by Reciprocal cross. In case of cytoplasmic inheritance and sex linkage result will change by Reciprocal cross.

(a) TI × tt (b) TT × tt

(Female) (Male) (Male) (Female)

↓ ↓

All Tall All Tall

## BEGINNER'S BOX-1

1. If the cell of an organism heterozygous foe 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 postulate of Mendel is still universal in nature?
(1) Postulate I (2) Postulate II (3) Postulate III (4) Postulate N

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

## GENE INTERACTION

Gene interaction is of two types :

**(i) Allelic interaction/lntragenic interaction**

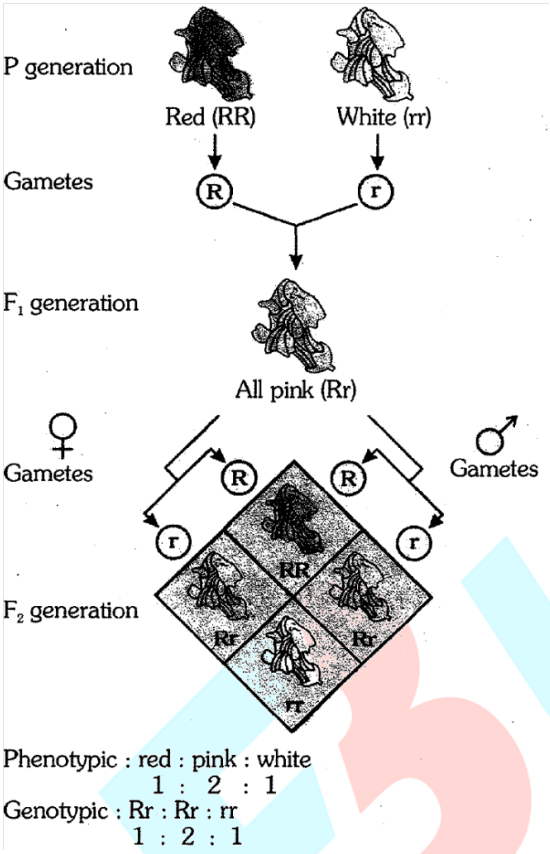
**(ii) Non allelic interaction/lntergenic interaction**

**(i) Allelic interaction/lntragenic interaction:** Allelic interaction takes place between allele of same gene which are present at same locus. Example of Allelic interaction are as follows :-

**(1) Incomplete' dominance :-** According to Mendel's law of dominance, dominant character must be present in $F_1$ generation. But in some organisms, $F_1$ generation is different from the both parents. Both factors such as dominant and recessive are present in incomplete dominance but dominant factors is unable to express its character completely, resulting Intermediate type of generation is formed which is different from the both parents. Some examples are -

**(a) Flower colour in Mirabilis jalapa :** Incomplete dominance was first discovered by **Correns** in Mirabilis jalapa. This plant is called as **'4 O' clock plant** or **'Gul-e-Bans'**. Three different types of plant are found in Mirabilis on the basis of flower colour, there are red, white and pink.

- When plants with red flowers is crossed with white flower, plants with pink flower are obtained in $F_1$ generation. The reason of this is that the genes of red colour is incompletely dominant over the genes of white colour.
- When $F_1$ generation of pink flower is self pollinated then the phenotypic ratio of $F_2$ generation is red, pink, white is 1:2:1 ratio in place of normal monohybrid cross ratio 3:1.
- The ratio of phenotype and genotype of $F_2$ generation in incomplete dominance is always same.




**(b) 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.

**(c) Feather colour in Andalusian Fowls :-** Incomplete dominance is present for their feather colour. When a black colour fowl is crossed with a white colour fowl, the colour of $F_1$ generation is blue.

**(d) Size of starch grain in pea plant :** When a large size is crossed with small size, the size of starch grain in $F_1$ generation is intermediate.

**[2]** **Co-dominance :-** In this phenomenon, both the allele of a gene express for a particular character in $F_1$ progeny. There is no blending of characters, where as both the characters expressed equally.

**Examples:-**

(i) Co-dominance is seen .in cattle for coat colour.

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

When we obtain $F_2$ generation from the $F_1$ generation, the ratio of black; black-white (Roan); white cattle is obtained in 1 : 2 : 1

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

BLACK × WHITE

$R_1R_2$ ↓ $R_1R_2$

$F_1$ generation $R_1R_2$ (Roan)

↓ Sib-mating cross

| | $R_1$ | $R_2$ |
|---|---|---|
| $R_1$ | $R_1R_2$ | $R_1R_2$ |
| $R_2$ | $R_1R_2$ | $R_2R_2$ |

$R_1R_2$ = Black-1 $R_1R_2$ = Roan-2 $R_2R_2$ = White – 1

It is obvious by above analysis that the ratio of phenotype as well as genotype is 1:2:1 in co-dominance.

**Sp. Note :-** In incomplete dominance, characters are blended phenotypically, while in co-dominace, both the genes of a pair exhibit both the characters side by side and effect of both the character is independent from each other.

Other Examples of Co-dominance :

(ii) AB blood group inheritance ($I^AI^B$)

(iii) Carrier of Sickle cell anaemia ($Hb^A$ $Hb^S$)

**[3] Multiple allele :-** More than 2 alternative forms of same gene are called as multiple allele. Multiple allele is formed due to mutation; Multile allele are located on same locus of homologous chromosome.

A diploid individual contains two alleles and gamete contains one allele for a character.

If n is the number of allele of a gene then number of different possible genotype = $\frac{n(n+1)}{2}$

**Example of multiple allele :**

**1. ABO blood group →** ABO blood group system are determined by three alleles - $I^A$, $I^B$, and i

$I^A$ = dominant

$I^B$ = dominant

i = recessive

Possible phenotypes- A, B, AB, O

| Blood group | Genotype | Antigen or agglutinogen | Antibody or agglutinin |
|---|---|---|---|
| A | $I^AI^A$, $I^Ai$ | A | b |
| B | $I^BI^B$, $I^Bi$ | B | a |
| AB | $I^AI^B$ | A & B | None |
| O | ii | None | a & b |

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

2. Coat colour in rabbits → 4 alleles

**[4] Lethal gene :-** Gene which causes death of individual when it comes in homozygous condition called lethal gene. Lethal gene may be dominant or recessive both, but mostly recessive for lethality.

1. Lethal gene was discovered by **L. Cuenot** in coat colour of mice.
   Yellow body colour(Y) was dominant over normal brown colour(y).
   Gene of yellow body colour is lethal.
   So homozygous yellow mice are never obtained in population. It dies in embryonal stage.
   When yellow mice were crossed among themselves segregation for yellow and brown body colour was obtained in 2 : 1 ratio.

   Yy × Yy

| | Y | y |
|---|---|---|
| Y | YY | Yy |
| y | Yy | yy |

   YY - death in embryonal stage modified ratio = 2 : 1

2. In plant lethal gene was first discovered by **E. Baur** in Snapdragon (Antirrhinum majus)

   Snapdragon ⟨ Golden leaves(G) / Green leaves(g)

   Golden × Golden
   Gg Gg

| | G | g |
|---|---|---|
| G | GG | Gg |
| g | Gg | gg |

   Modified ratio : 2 : 1

   Homozygous golden leaves are never obtained.

Other example of lethality :

(i) Gene of haemophilia [$X^hX^h$ female]

(ii) Gene of sickle cell anaemia [$Hb^S$ $Hb^S$]

**[4] 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 is the effect of a gene underlying mechanism of pleiotropy in most cases is the effect of a gene on metabolic patheays which contributes towards different phenotypes. An example of this is the disease phenylketonuria. which occurs in humans. The disease is caused by mutation in the

gne that codes for the enz.yme phenyl alanine hydroxylase (single gene mutation). This manifests itself through phenotypic expression characterised by mental retardation and a reduction in hair and skin pigmentation.

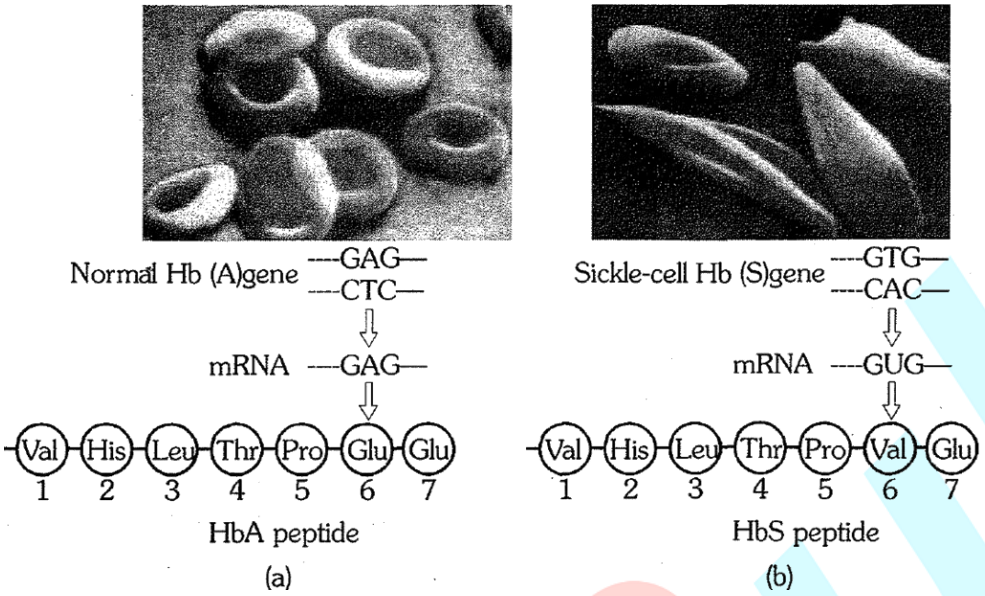
For example :

(1) In Pea plant : Seed shape and size of starch grain control by same gene located on 7th chromosome. Occasionally. a single gene product may produce more than orie effect. For example, starch synthesis in pea seeds is controlled by one gene. It has two alleles (B and b). Starch is synthesised effectively by BB homozygotes and therefore, large starch grains are produced. In contrast, bb homozygotes have lesser efficiency in starch synthesis and produce smaller starch grains. After maturation of the seeds. BB seeds are round and the bb seeds are wrinkled. Heterozygotes produce round seeds. and so B seems to be the dominant allele. But, the starch grains produced are of intermediate size in Bb seeds. So if starch grain size is considered as the phenotype, then from this angle, the alleles show incomplete dominance.

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 phehotype that we choose to examine, in case more than one phenotype is influenced by the same gene.

| | Seed shape | Size of starch grain |
|---|---|---|
| BB | Round | Large |
| Bb | Round | Medium |
| bb | Wrinkel | Small |

**(2) Sickle cell anaemia (Autosomal linked recessive) -** Gene $Hb^S_\beta$ provide a classical example of pleiotrophy. It not only causes haemolytic anaemia but also results increased resistance to one type of malaria that caused by the parasite Plasmodium falciparum. The sickle cell $Hb^S_\beta$ allele also has pleiotropic effect on the development of many tissues and organs such as bone, lungs, kidney, spleen, heart. 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.




**Micrograph of the red blood cells and the amino acid composition of the relevant portion of 13-chain of haemoglobin : (a) From a normal individual; (b) From an individual with sickle-cell anaemia**

**(ii) Non allelic interaction/Intergenic interaction**

When interaction takes place between non allele is called non allelic gene interaction. It changes or modifies other non allelic gene.

**Examples of nonallelic interaction.**

**1. Complementary Gene:-** Two pair of non allelic genes are essential in dominant form to produce a particular character.

Such genes that act together to produce an effect that neither can produce, it's effect separately are called complementary genes.

Both types of gene must be present in dominant form.

Example :- colour of flowers in Lathyrus odoratus :-

C - p Purple coloured .·

C - pp colourless

cc - p colourless

cc - pp colourless

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

CCPP × ccpp

(coloured) ↓ (Colourless)

$F_1$-Generation → CcPp (coloured)

| | CP | Cp | cP | cp |
|---|---|---|---|---|
| CP | CCPP Coloured | CCPp Coloured | CcPP Coloured | CcPp Coloured |
| Cp | CCPp | CCpp | CcPp | Ccpp |

| | Coloured | Colourless | Coloured | Colourless |
|---|---|---|---|---|
| cP | CcPP<br>Coloured | CcPp<br>Coloured | ccPP<br>Colourless | ccPp<br>Colourless |
| cp | CcPp<br>Coloured | Ccpp<br>Colourless | ccPp<br>Colourless | ccpp<br>Colourless |

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

**2.** **Epistasis :-** When, a gene prevents the expression of another non-allelic gene, then it is known as epistatic gene and this phenomenon is known as Epistasis.

Gene which inhibit the expression of another non alleleic gene is called epistatic gene and expression of gene which is suppressed by epistatic gene called hypostatic gene.

Epistasis is of two types:- (1) Dominant epistasis (2) Recessive epistasis

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

Y = Dominant allele for yellow colour of fruit

y = Recessive allele for green colour of fruit

W = Epistatic gene over Y and y gene and forms white colour of fruit.

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

White × Green

WWYY × wwyy

↓

$F_1$-generation WwYy White colour

↓ $F_2$ generation

| | WY | Wy | wY | wy |
|---|---|---|---|---|
| WY | WWYY<br>White | WWYy<br>White | WwYY<br>white | WwYy<br>white |
| Wy | WWYy<br>white | WWyy<br>white | WwYy<br>white | Wwyy<br>white |
| wY | WwYY<br>White | WwYy<br>white | wwYY<br>yellow | wwYy<br>yellow |
| wy | WwYy<br>white | Wwyy<br>white | wwYy<br>yellow | wwyy<br>green |

It is obviously clear by above analysis, the phenotypic ratio of $F_2$- generation in dorminant epistasis is – 12 : 3 : 1

**3.** **Supplementary gene and Recessive Epistasis :-** A pair of gene change the effect of another non allelic gene, is called supplementary gene.

**Example :-** Coat colour in Mice.

If alleles, C = Black coat colour

c = Albino (Colourless coat) or (It has no effect)

A = Supplementary gene

When black caot mice crossed with albino mice, the $F_1$ generation is Agouti.

It means, here the effect of non allelic gene is changed.

Black Albino

CCaa × ccAA

↓

$F_1$-Generation → CcAa [Agouti]

↓ Sibmateing

| | CA | Ca | cA | Ca |
|---|---|---|---|---|
| CA | CCAA<br>Agouti | CCAa<br>Agouti | CcAA<br>Agouti | CcAa<br>Agouti |
| Ca | CCAa<br>Agouti | CCaa<br>Black | CcAa<br>Agouti | Ccaa<br>Black |
| cA | CcAA<br>Agouti | CcAa<br>Agouti | ccAA<br>Albino | ccAa<br>Albino |
| ca | CcAa<br>Agouti | Ccaa<br>Black | ccAa<br>Albino | ccaa<br>Albino |

Thus, Recessive epistasis and supplementary gene ratio in $F_2$-Agouti : Black : Albino

9 : 3 : 4

## POLYGENIC INHERITANCE/QUANTITATIVE INHERTANCE

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. Such traits are generally controlled by three or more genes and are thus called as polygenic traits.

Besides the involvement of multiple genes polygenic inheritance also takes into account the influence of environment. Human skin colour is another 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.

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

lightest skin colour and that with all the recessive alleles (aabbcd will have the lightest skin colour. As expected the genotype with three dominant alleles and three recessive alleles will have an intermediatge skin colour. In this manner the number of each type of alleles in the enotype would determine the darkness or lightness of the skin in an individual.

Inheritance of characters in which one character is controlled by many genes and intensity of character depends upon the number of dominant allele.

Number of phenotype = (2n + 1) here n = number of polygene

**Example-1.** kernal colour of wheat is regulated by two polygene.

RRBB × rrbb
Red White
↓
$F_1$-generation RrBb (intermediate)

| Phenotype :- $F_2$-generation | 1 | : | 4 | : | 6 | : | 4 | : | 1 |
|---|---|---|---|---|---|---|---|---|---|
| | Full red | | light red | | intermediate red | | very light | | white |
| Number of dominant gene. | 4 | | 3 | | 2 | | 1 | | 0 |

1Red : 14 intermediate : 1 white

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

The inheritance of colour of skin in human was studied by **Devenport.**

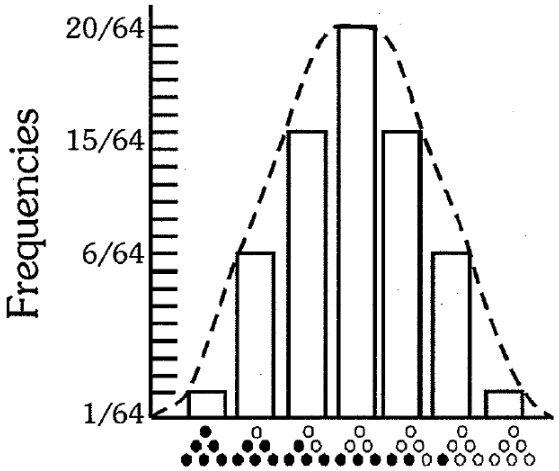
Human skin colour is regulated by three polygene.

When a Negro Black (AA BB CC) phenotype is crossed with white (aa bb cc) phenotype, intermediate phenotype is produced in $F_1$ generation.

Negro Black × White
AA BB CC ↓ aa bb cc
Aa Bb Cc → $F_1$-generation
[Intermediate brown/ mullato]
↓ (Inbreeding)
$F_2$-generation
↓

Phenotypic ratio will be :-

| Negro Black | : | Very Dark Brown | : | Dark Brown | : | Mullato | : | Light Brown | : | Very Light Brown | : | White |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1 | : | 6 | : | 15 | : | 20 | : | 15 | : | 6 | : | 1 |




## CYTOPLASMIC INHERITANCE

Discovered by Correns.

Inheritance of characters which are controlled by cytogene or cytoplasm is called cytoplosmic inheritance.

Genes which are present in cytoplasm called **'cytogene'** or **'plasniagene'** or extra nuclear gene.

Total cytogene present in cytoplasm is called **'Plasmon'.**

A gene which is located in the nucleus is called ' karyogene'.

Inheritance of cytogene in organisms occurs only through the female. Because female gamete has karyoplasm, simultaneously it has cytogene because of more cytoplasm.

The male gamete of higher plant is called male nucleus. It has very minute [equivelent to nil] cytoplasm. so male gamete only inherited karyogene.

Thus, inheritance of cytogene occurs only through female. (also called maternal inheritance)

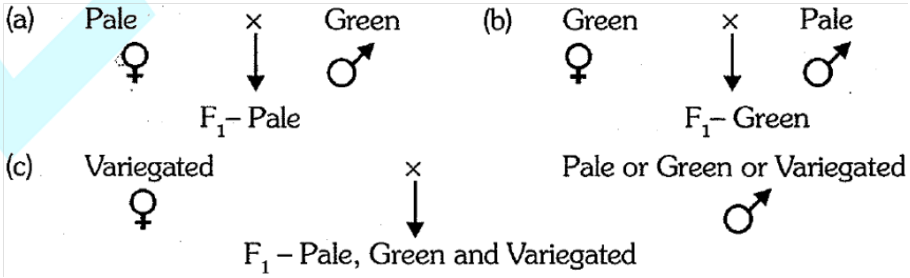
If there is a reciprocal cross in this condition, then results may be affected.

Cytoplasmic inheritance involving essential organelles like, Chloroplast and mitochondria called as **organellar inheritance.**

**Example of Organellar Inheritance:** (True examples of cytoplasmic inheritance)

**(a) 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.

Branch colour



**(b)** **Male sterility in maize plant :** Gene of male sterelity is present in mitochondria. If a normal male plant crossed with a female plant which has genes of male sterility then all the generation of male become sterile because a particular gene was present with female which was inherited by female.

**(c)** **Albinism in plant :** Gene of albinism found in chloroplast. Gene of albinism in Maize is lethal.

## BEGINNER'S BOX-2

**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 pairs of genes. 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

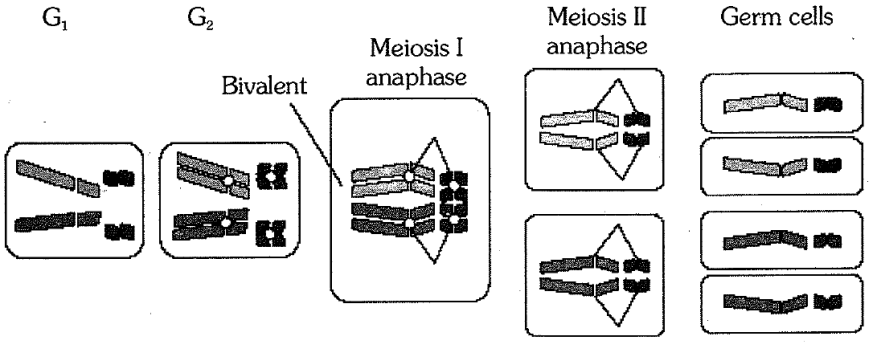
**5.** In mother has blood group AB, father has A group then Which of the following blood group will not found in the offspring.
(1) A (2) B (3)AB (4) O

## 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 Theodore Boveri noted that the behaviour of chromosomes was parallel to the behaviour of genes and used chromosome movement to explain Mendel's laws.** Recall that you have studied the behaviour of chromosomes during mitosis (equational division) and during meiosis (reduction division). 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 sites on homologous chromosomes.

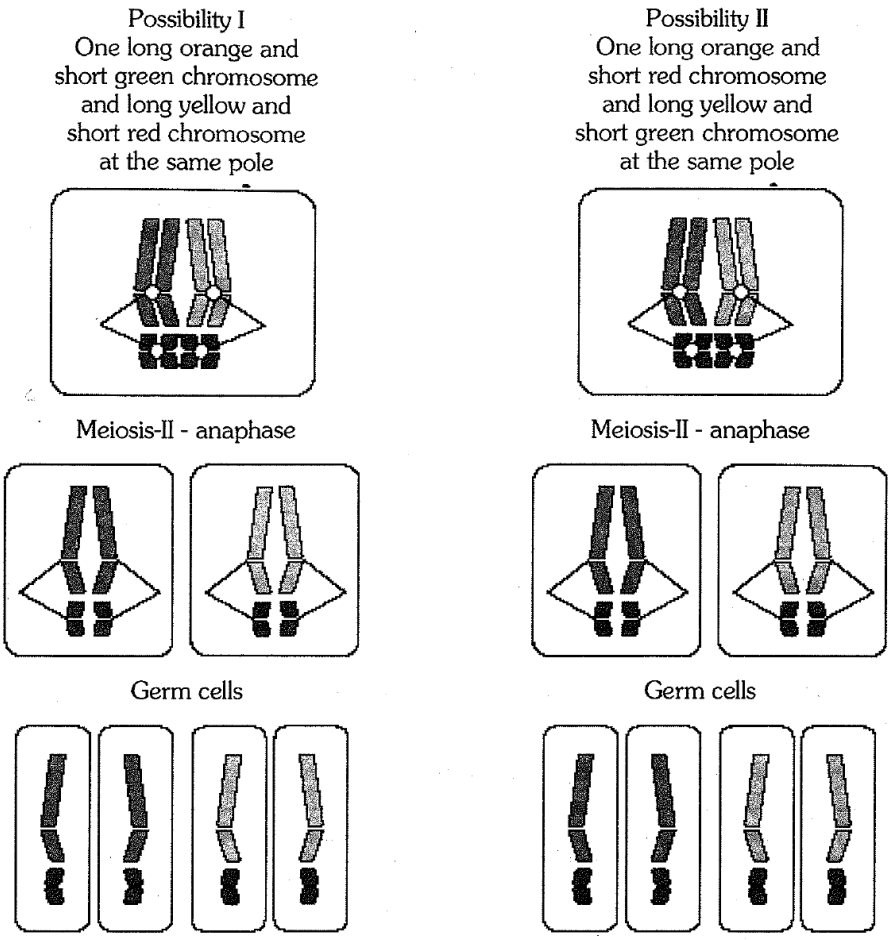



**Meiosis and germ cell formation in a cell with four chromosomes. You can see how chromosomes segregate when germ cells are formed**

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 D orange and green is segregating together. But in the right hand column (Possibility 10 the orange chromosome is segregating with the red chromosomes.

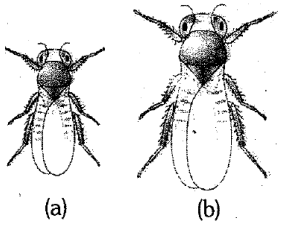
**A Comparison between the Behaviour of Chromosomes and Genes**

| A (Gene) | B(Chromosome) |
|---|---|
| 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 |




**Independent assortment of chromosomes**

Sutton and Boveri argued that the pairing and separation of a pair of chromosomes would lead to the segregation of a pair of factor 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*
(a) Male (b) Female

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

1. They could be grown on simple synthetic medium in the laboratory.
2. They complete their life cycle in about two weeks,

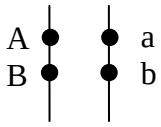
3. A single mating could produce a large number of progeny flies.
4. There was a clear differentiation of the sexes- the male and female flies are easily distinguisable. ·
5. It has many types of hereditary variations that can be seen with low power microscopes.

**LINKAGE :** The physical association of genes on a chromosome is called linkage.

Collective inheritanre of dlaracter is due to linkage. Sex linkage was first discoverd by **Morgan** in Drosophila and coined the term linkage. He proposed the theory of linkage.

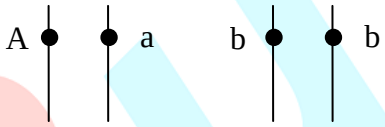
Linkage and independent assortment can be represented in dihybrid plant, as –

In case of linkage in dihybrid (AaBb)




It produces two types of gamete
AB : ab

In case of independent assortment in dihybrid (AaBb)




It produces two types of gamete
AB : ab : aB : Ab

**Theory of linkage**

1. Linked genes are linearly located on same chromosome. They get separated if exchange (crossing over), takes place between them.
2. 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 inherit together so we consider one linkage group.
   Number of Linkage group =Number of homologous chromosome pairs or Haploid no. of chromosomes.

| | 2n | n | Pair | Linkage group |
|---|---|---|---|---|
| Pea | 14 | 7 | 7 | 7 |
| Maize | 20 | 10 | 10 | 10 |
| Drosophila | 8 | 4 | 4 | 4 |
| Barley | 14 | 7 | 7 | 7 |
| Mouse | 42 | 21 | 21 | 21 |

**3.** Strength of linkage $\alpha$ 1/ distance between the genes. It means, if the distance between two genes is increased then strength of linkage is reduced and it proves that greater is the distance between genes, the greater is the probability of their crossing over.

Crossing over obviously disturbs or degenerates linkage. Linked genes can be separated by crossing over.

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

(1) Distance↑ = C.O.↑

(2) Temperature↑ = C.O.↑

(3) Age↑ = C.O.↓

(4) Sex- Male C.O.↓ (Crossing over is totally absent in male Drosophila)

**Arrangement of linked Genes on Chromosomes :-**

The arrangement of linked genes in any dihybrid is of two types.

**[a] Cis - Arrangement :-** When, two dominant genes are located on one chromosome and both recessive genes located on another chromosome, such type of arrangement is termed as cis-arrangement. Cis-arrangement is an original arrangement.


γ Two types of gamete can be produced in cis-arrangement→ (AB) and (ab).

**[b] Trans-arrangement :-** When a chromosome bears one dominant and one .recessive gene, and another chromosome also possess one dominant and one recessive gene, such type of arrangement is called trans-arrangement. Trans-arrangement is not an original form. It is due to crossing over. Two types of gamete also formed in trans-arrangement but it is different from cis-arrangement (Ab) and (aB).


**Types of Linkage :-** There are two types of linkage -

1. **COMPLETE LINKAGE :-** Linkage in which genes always show parental combination. It never forms new combination.

   Crossing over is absent in it. Such genes are located very close on the chromosomes. Such type of linkage is very rare in nature. e.g. male Drosophila, female silk moth.

2. **INCOMPLETE LINKAGE :-** When new combinations also appear along with parental combination in off springs, this type of linkage is called incomplete linkage, the new combinations are formed due to crossing over.

   **Application of Linkage :-**

   Distance between linked genes can be identified by the incomplete linkage. It's unit is centi Morgan.

$$\text{Strength of linkage} \propto \frac{1}{\text{Distansce b/w linked gene}} \propto \frac{1}{\text{Crossing Over}}$$

**Genetic map/Linkage map/chromosome map-** In genetic map different genes are linearly arranged according to% of recombination ($\propto$ Distance) between them.
With the help of genetic map we can find out the position of a particular gene on chromosome.
Genetic map is helpful in the study of genome.

Morgan's student Alfred Sturtevant used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and 'mapped' their position on the chromosome.

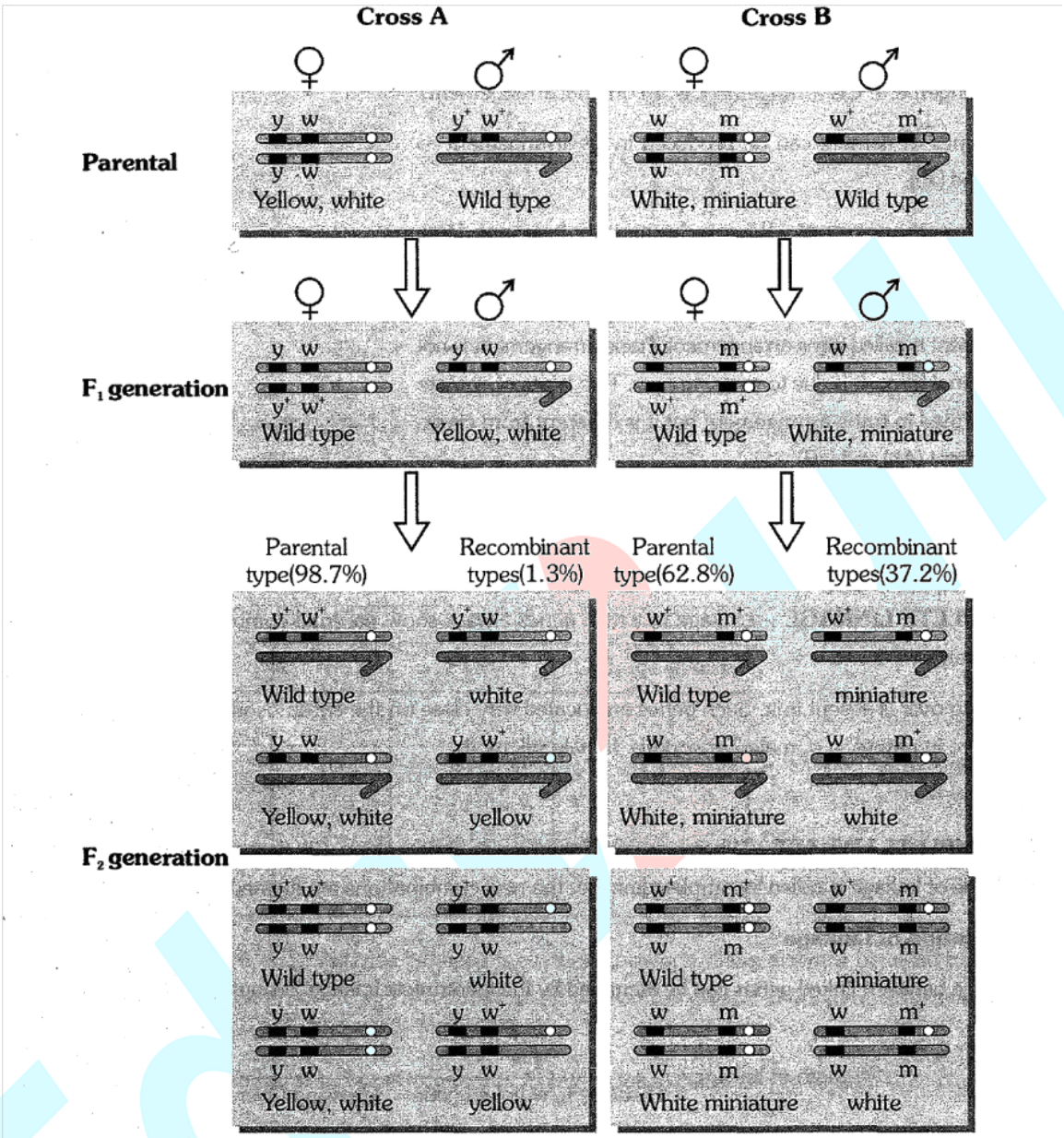



**Figure :** Results of two dihybrid crosses conducted by Morgan.Cross A shows crossing between gene y and w; Cross B shows crossing between genes w and m. Here dominant wild type alletes are represented with (+) sign in superscript. Note : The strength of linkage between y and w is higher than w and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

## SEX LINKAGE

When the genes are present on sex-chromosome is termed as sex linked gene and such phenomenon is known as sex-linkage.

**Types of sex linkage :-**

**1. X-linkage.**

Genes of many somatic characters are found on x-chromosome. The inheritance of x-linked character may be through the males and females.

**Example of X-linkage :-**

**[i] Eye colour in Drosophila :-** Eye colour in Drosophila is controlled by aX-linked gene.

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

Gene for red eye is dominant (+) and white colour of eye is recessive (w)

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

Heterozygous red eyed female = $X^{+}X^{W}$

Homozygous white eyed female = $X^{W}X^{W}$

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

Hemizygous white eyed male = $X^{W}Y$

It is dear by above different types of genotype that female is either homozygous or heterozygous for eye colour. But, for the male eye colour, it is always hemizygous.

**[ii] Haemophilia :-**

Haemophilia is also called **"bleeder's disease"** and first discovered by **John Otto** (1803). The gene of haemophilia is recessive.

On the basis of x-linked, following types of genotype are found.

$X^{h}X$ = Carrier female

$X^{h}X^{h}$ =Affected female

$X^{h}Y$ =Affected male.

Haemophilia –A$\rightarrow$ due to lack of factor -Vlll (Antihaemophilic globulin AHG). It shows ethality in homogygous condition.

Haemophilia B or Christmas disease - due to lack of factor - IX (Plasma thromboplastin component)

Haemophilia - C (Autosomal disorder) ~ due to lack of factor - XI (Plasma Thromboplastin antecedent)

**[iii] Colour Blindness :-** The inheritance of colour-blindness is alike as haemophilia, but it is not a lethal disease so it is found in male and female.(discovered by **Horner**)

**Types of X-linked colour blindness are :-**

**[a] Protanopia :-** It is for red colour.

**[b] Deuteranopia :-** It is for green colour

Colour b1indness is checked by ishihara - chart.

It is a sex-linked recessive disorder due to defect in either red or green cone of eye resulting in failure to discriminate between red and green colour. This defect is doe to mutation in certain genes present in the X chromosome. it occurs in about 8 per cent of males and only about 0.4 per cent of females. This is because the genes that lead to red-green colour blindness are on the X chromosome. Males have only one X chromosome and females have two. The son of a woman who carried the gene has a 50 per cent change of being coour blind. The mother is not herself colour blind because the gene is recessive.That means that its effect is suppressed by her

matching dominant normal gene. A daughter will not normally be colour blind. unless her mother is a carrier and her father is colour blind.

**Other examples of X-linkage**

[iv] Diabetes insipidus (recessive).
[v] Duchenne muscular dystrophy (recessive).
[vi] Pesudoricketes (Dominant)
[vii] Defective enamel of teeth (Dominant)

**2. Y-linkage -** The genes of some somatic characters are located on Y- chromosome. The inheritance of such type of character is only through the males. Such type of character is called Holandric character. These characters are found only in male.

e.g. (1) Gene which forms TDF /sry-gene
(2) Hypertrichosis (excessive hair on ear pinna)

Gene which is located on differential region of Y - chromosome is known as Holandric gene.

**Types of Inheritance of sex linked characters :-**

**1. Criss cross inheritance (Morgan) :-** In criss-cross inheritance male or female parent transfer a 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)

## Sex-Limited Character

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.

For example - genes of beard-moustache express their effects only in the presence of male hormone - testosterone.

## 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.

## 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.

**Sex Determination on the basis of fertilization.**

**Three types -**

1. **Progamic -** Sex is determined before fertilization.
   **eg.** - drone in honey bee
2. **Syngamic** - Sex is determined during fertilization.
   **eg. -** most of plants& animals
3. **Epigamic-** Sex is determined after fertilization.
   **eg.** - Female in honey bee.

**Mechanism of sex determination :**

**[1] Allosomic determination of sex-**

Chromosomes are of two types -

(a) Autosomes or somatic chromosomes -
These regulate somatic characters.

(b) Allosomes or Heterosomes or Sex chromosomes -

These chromosomes are associated with sex determination. Term **"Allosome"** & **"Heterosome"** were given by Montgomery~

Sex chromosomes were discovered by **"Me Clung"** in grass hopper

X- Chromosome was discovered by **"Henking"** and called it 'x-body'.

**Wilson & Stevens** proposed chromosomal theory for sex determination.

**(1) XX - XY type or Lygaeus type :-** This type of sex determination first observed by **Wilson** & **Stevens** in Lygaeus insect. Two types-

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

2A + XX (Female) → gametes → A + X, A + X

Male is heterogametic (male produce two types of gamete)

2A + XY (Male) → gametes → A + X, A + Y

**eg.** Man

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

It is found in birds.

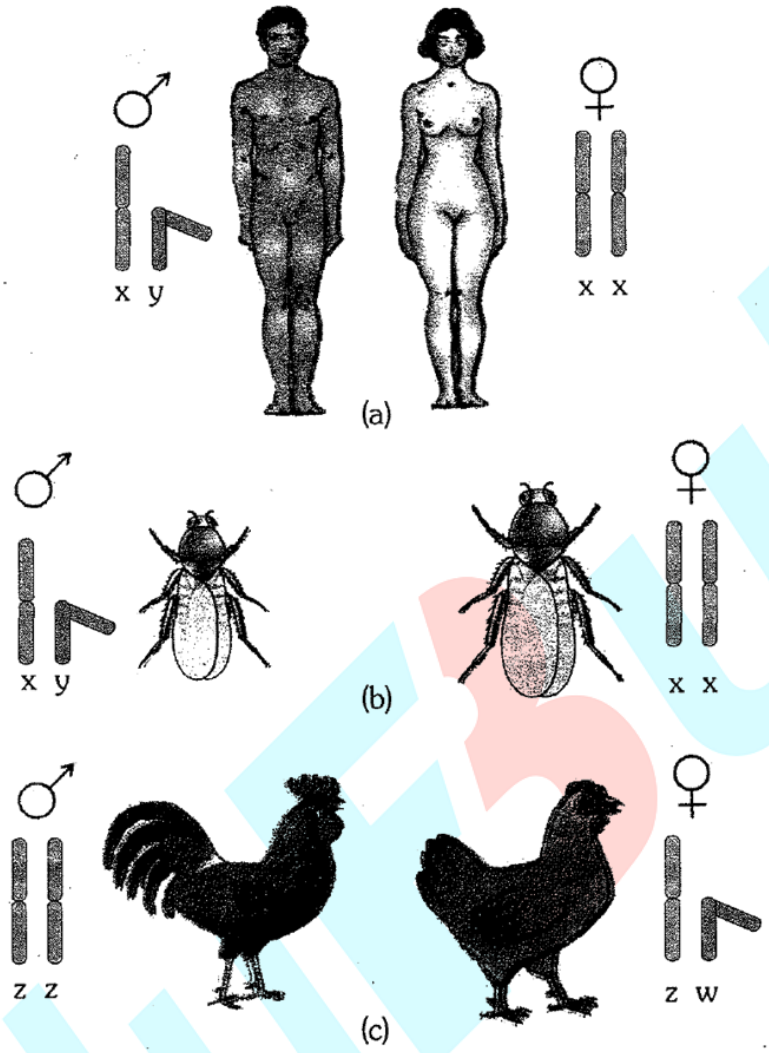



**Figure :** Determination of sex by chromosomal differences: (a,b) Both in humans and in *Drosophila*, the female has a pair of XX chromosomes (homogametic) and the male XY (heterogametic) composition; (c) In many birds, female has a pair of dissimilar chromosomes ZW and male two similar ZZ chromosomes 0

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

Female (2A + XX) → A + X, A + X → homogametic

Male (2A + XO) → A + O, A + X → homogametic

**Example :-**

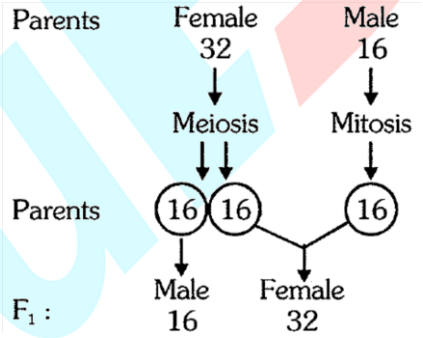
γ Grass hopper
γ Cockroch

**Haploid- diploid mechanism (Sex determination in Honey Bee)-**

Sex determination takes place by sets of chromosomes.
Diploid (two sets) → Female
Haploid (One set) → Male
In honey bee, male individual (Drone) develops from unfertilized eggs (Haploid). Male develop by parthenogenesis.
Queen and worker bees develop from diploid eggs i.e. fertilized egg.

Fertilzed egg → diploid larva 2n
- Feeds on Royal jelly → Queen (Fertile female )
- Bee bread → Worker (Sterile female )

The sex determination in honey bee is based on the number of sets of chromosomes an individual receives. 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 chromomes than that of a female. The females are diploid having 32 chromosomes as haplodipoloid sex-determination system and has special characteristic features such as the males produce sperms by mitosis (figure), they do not have father and thus cannot have sons, but have a grandfather and can have grandsons.




**[2] Genic balance theory:-** C.B. Brides proposed genic balance theory for sex determination in Drosophila.

γ According to Brides in Drosophila Y chromosome is heterochromatic so it is not active in sex determination In Drosophila sex determination takes place by sex index ratio.

$$\text{Sex index ratio} = \frac{\text{No. of x chromosomes}}{\text{No. of set of Autosomes}} = \frac{X}{A}$$

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 and development of male reproductive organ, so y-chromosome is essential for the production of fertile male.

Sex index ratio (a) $\frac{X}{A} = 1 \rightarrow$ female (2A + XY)

(b) $\frac{X}{A} = 0.5 \rightarrow$ male $\begin{cases} (2A+ XY) = \text{Fertile male} \\ (2A+ XO) = \text{Sterile male} \end{cases}$

(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)

**Cytological basis of sex identification -**

Barr body technique or **Lyon's hypothesis**-

Interphasic nucleus of human female contains two X- chromosomes. Out of two, one X-chromosome becomes heterochromatin and other X- chromosome is euchromatin. By staining X- heterochromatin, it appears as a dense body which is called Barr body. (Facultative hetrochromatin)

No. of Barr body $\Rightarrow$ (No. of X chromosomes - 1)

So in a Normal female (2A + XX) $\rightarrow$ One Barr body

Normal male (2A + XY) $\rightarrow$ Barr body absent

Turner syndrome (Sterile female) (2A + XO) $\rightarrow$ No. Barr body

Klinefelter syndrom (Sterile male)(2A + XXY) $\rightarrow$ One Barr body

Drum stick which occurs in blood of female of mammals, is also a type of Barr body.

## BEGINNER'S BOX-3

**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) Mullar (3) Mendel (4) Johannsen

**3.** X-linked recessive gene easily express in :
(1) male (2) female
(3) equal in male and female (4) not easily express

**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

# HUMAN GENETICS

The study (analysis) of genetic characters and aspects like genetic improvements among humans are included in **human genetics.** This is also known as **eugenics** (well born).

Eugenics is a term derived from Greek language **'Eugenes'** meaning "Well born". First of all, **Sir Francis Galton,** 1883 proposed the idea of improvement in human species through change in hereditary characters in a scientific manner and named it Eugenics. Because of this Sir Francis Galton is known as "father of Eugenics". To find out various facts, scientists have to perform a number of experiments, but it is not possible to do so in humans. The following problems are faced in studying human genetics -

1. Cells of human body are relatively smaller and number of chromosomes present in them is more.
2. It is not possible to perform various experiments on humans in the laboratory.
3. Due to greater life span of humans, a lot of time is required to study their genetic characteristics.
4. Rate of reproduction is slow in humans.
5. Individuals of human species are generally heterozygous for various characters and it is very difficult to find homozygous individuals.
6. Due to controlled hybridization and long lived life among humans, it is not possible to study many generations in easy way.

Despite of above mentioned problems humans are considered suitable for genetical experiments due to-

1. Longer life span of humans, the abnormalities that require relatively long period to express themselves can be easily studied.
2. By pedigree study and analysis of families, many genetic characters of man can be traced out.
3. To study many diseases (like haemophilia, colour blindness etc.) and intelligence quotient (I.Q.) humans are more preferable than any other organisms.

**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 tounge | Rollar | Non Rollar |
| Rh Factor | $Rh^+$ | $Rh^-$ |
| PTC (Phenyl thiocarbamide) faster | Taster | Non Taster |
| Skin pigmentation | Normal | Albino |

## Devices Used in Human Genetical Studies

The study and analysis of human genetics is performed by many methods like pedigree analysis. Statistical analysis and human karyotyping of these the important ones, that is, pedigree analysis and human karyotype is being described here.

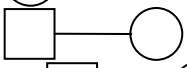
### 1. Pedigree Analysis

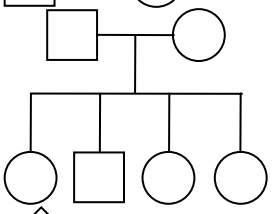
Study of ancestoral history of man of transmission of genetic characters from one generation to next, is pedigree analysis. Dwarfism, albinism, colour blindness, haemophilia etc. are genetically transmitted characters. To study and analyse them a pedigree of genetic facts/data and following symbols are used.

**Symbol used in Pedigree :**

**1.**  - Normal Male

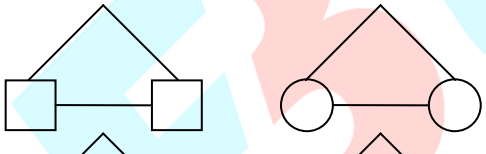
**2.**  - Normal Female

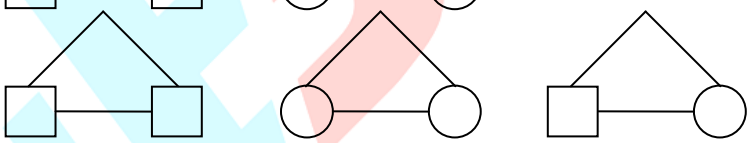
**3.**  - Mating (marriage)

**4.**  - The siblings are indicated in chronological order of birth

**5.**  - Sex unspecified

**6.** Twin

If monozygotic 

If dizyotic 

**7.**  Affected male and female individual

**8.**  Heterozygous for autosomal recessive

**9.**  Carrier female of sex linked recessive character or disease

**10.**  Death of individual

**11.** Abortion or still birth (sex unspecified)

**12.** Consanguineous marriage

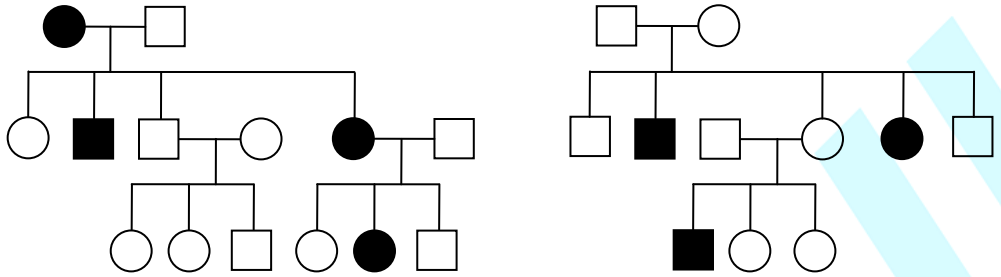
**13.** Parent with male child affected with disease

**14.**  Five unaffected off springs

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.

A representative pedigree is shown in Figure for dominant and recessive traits, discuss with your teacher and design pedigrees for characters linked to both autosomes and sex chromosome.

**Example:-**



(a) (b)

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

## POPULATION GENETICS

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

1. **Gene pool -** A gene pool is the sum total of genes in reproductive gametes of a population.
2. **Gene-flow** - Migration of gene from one population to another population by cross fertilization.
3. **Genetic load** - The existance within the population of disadvantegeous allele in heterozygous genotype is known as genetic load
4. **Gene frequency** - Gene frequency is defined as proportion of different alleles of a gene in a population.

**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 gene - $50 \times 2 + 20 = 120$
Total N gene - $30 \times 2 + 20 = 80$
Total gene - 200

Frequency of M gene $P = \frac{M}{M+N} = \frac{120}{200} = 0.6$

Freq. of N gene $\frac{N}{M+N} = \frac{120}{200} = 0.4$

$0.6 + 0.4 = 1$
$P + q = 1$

**Hardy Weinberg Law-**

1908 G.H.Hardy (English methematician) & German Physician, W. Weinbergh independently discovered that an equilibrium is established between frequencies of allele in random mating population and these gene frequency remain constant from generation to generation.

This law is applicable when factors like mutation, selection & migration, are absent.

**Hardy Weinberg theorem or Hardy weinbergh law**

$p^2 + 2pq + q^2 = 1$ $(A \rightarrow p, a \rightarrow q, p + q = 1)$

AA Aa aa

In this equation frequency $\rightarrow$ P or the Frequency of Homozygous dominant will be AA-$P^2$

In this equation frequency of a $\rightarrow$ q the frequency of homozygous recessive - $q^2$

The frequency of Aa = 2pq

The frequency of different genotype produced due to random mating will depend upon the gene frequency and equilibrium is stablished after one single generation of random mating.

**Q.** In a random 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 x 0. 7 x 0.3 = 0.42 = 42%

## BEGINNER'S BOX-4

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

(1)  = unaffacted female

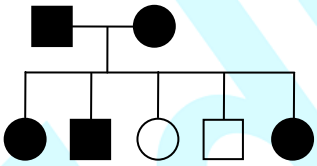
(2)  = affacted male

(3)  = affacted female

(4)  = affacted 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%

## 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

♦ Complementary gene = 9 : 7
Dominant epJstasis = 12 : 3 : 1
Recessive epistasis = 9 : 3 : 4

♦ Coupling = 7 : 1 : 1 : 7
Repulsion = 1 : 7 : 7 : 1

♦ 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

♦ Trihybrid phenotypic ratio= 27 : 9: 9: 9: 3: 3: 3: 1

♦ Type of phenotype= $2^n$

♦ Type of gametes = $2^n$

♦ Type of genotype = $3^n$

♦ Total possible zygotic combination = $4^n$

♦ Possible genotype number On multiple alleles) = $\frac{n(n+1)}{2}$

♦ Modified ratio of lethal gene in monohybrid cross = 2 : 1

♦ Polygenic inheritance
- 1 : 4 : 6 : 4 : 1 (For two gene)
- 1 : 6 : 15 : 20 : 15 : 6 : 1 (For three gene)

♦ Type of phenotype in polygenic intheritance = (2n + 1)

♦ Contribution of each dominant allele = $\frac{\text{Maximum expression} - \text{Minimum expression}}{\text{Total number of dominant allele}}$

## ANSWER KEY

### BEGINNER'S BOX-1

**1.** (3) **2.** (3) **3.** (1) **4.** (4) **5.** (2) **6.** (1) **7.** (1)
**8.** (2) **9.** (4) **10.** (3)

### BEGINNER'S BOX-2

**1.** (2) **2.** (3) **3.** (3) **4.** (1) **5.** (4)

### BEGINNER'S BOX-3

**1.** (4) **2.** (1) **3.** (1) **4.** (2) **5.** (3)

## BEGINNER'S BOX-4

**1.** (4) **2.** (1) **3.** (1) **4.** (1) **5.** (1)

## Build Up Your Understanding