01 Principles of Inheritance and Variation

01. INTRODUCTION OF GENETICS

- Genetics term was given by **W. Bateson. (Genetics, Gr = to become or to grow into).**
- **Genetics:** Collective study of heredity and variations.
- **Heredity:** Transmission of genetic characters from parents to offspring.
- **Variation:** Individuals of same species have some differences; these are called variation.
- **Inheritance:** The process by which character is passed from parents to progeny; it is the basis of heredity.

History of researches in genetics :

- **G.J. Mendel:** Father of Genetics, born in Austria.
- **W. Bateson:** Father of Modern Genetics.
- **Morgan:** Father of Experimental genetics, He performed experiments on *Drosophila* & proposed various concepts, like Linkage, Sex linkage, Crossing over, Criss - cross inheritance, Linkage map on *Drosophila*.
- **Garrod:** Father of human genetics & biochemical genetics. Garrod discovered first human metabolic genetic disorder, which is called **alkaptonuria** (black urine disease).

Humans knew from as early as 8000-1000 B.C. that one of the causes of variation was hidden in sexual reproduction. They exploited the variations that were naturally present in the wild populations of plants and animals to selectively breed and select for organisms that possessed desirable characters. For example, through artificial selection and domestication from ancestral wild cows, we have well-known Indian breeds, e.g., Sahiwal cows in Punjab. We must, however, recognise that though our ancestors knew about the inheritance of characters and variation, they had very little idea about the scientific basis of these phenomena.

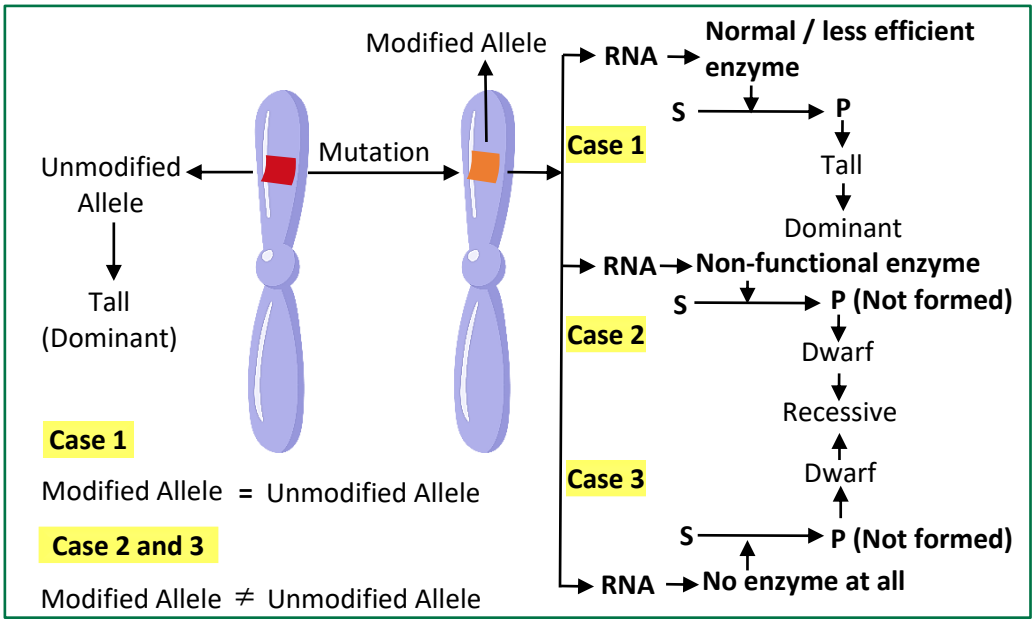
02. SOME GENETICAL TERMS

(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 are called Allele. Term allele was coined by **Bateson**.
- 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.

NCERT Question :

Differentiate between the following :-

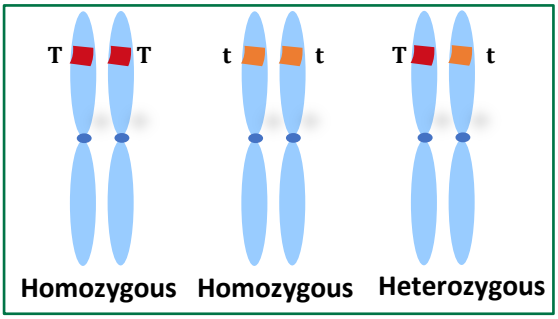
(b) Homozygous and Heterozygous

(3) HOMOZYGOUS :

A zygote is formed by fusion of two gametes having identical factors is called homozygote and organism developed from this zygote is called homozygous. e.g. TT, RR, tt.

(4) HETEROZYGOUS :

A zygote is formed by fusion of two different types of gamete carrying different factors is called heterozygote (**Tt, Rr**) and individual developed from such zygote is called heterozygous. The term homozygous and heterozygous were coined by **Bateson**.



(5) HEMIZYGOUS :

If any diploid 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 particular character.

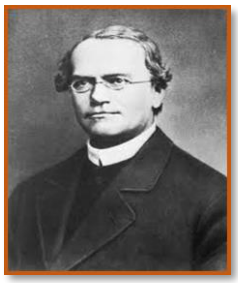
(7) GENOTYPE :

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 are said to be Phenocopy.

03. MENDELISM

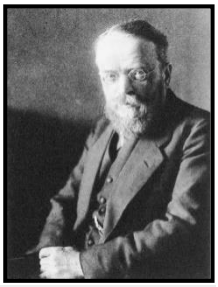


Gregor Johann Mendel (1822 - 1884)

- Experiments performed by Mendel on genetics and description of mechanisms of heredity 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 22 July 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 "**Experiments in plant Hybridization**" was published in a reputed 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.

  The credit of rediscovery of Mendelism goes to three scientists.



Carl Correns
(1864 - 1933)
Germany
(Experiment on Maize)

Hugo de Vries
(1848 - 1935)
Holland
(Experiment on Evening Primrose)

Erich von Tschermak
(1871 - 1962)
Austria
(Experiment on different flowering plants)

Hugo de Vries (Holland) Experiment on Evening Primrose. He republished Mendel's results in 1901 in **Flora** magazine.

Carl Correns converted two postulates of Mendel into Law of heredity/inheritance/Mendelism :

I Law - Law of segregation.

II Law - Law of independent assortment.

- Mendel published his work on inheritance of characters in 1865 but for several reasons, it remained unrecognised till 1900. Mendel experiments remain hidden for 34 years.
- **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.

NCERT Question :

Mention the advantages of selecting pea plant for experiment by Mendel.

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.

(b) 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 number of offspring can be analysed within a short period of time.

(ii) It has many contrasting traits.

(iii) Natural self 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 rajma and hawk weed.

(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. During Mendel's investigations into inheritance patterns it was for the first time that statistical analysis and mathematical logic were applied to problems in biology.

(f) The confirmation of his inferences from experiments on successive generations of his test plants, proved that his results pointed to general rules of inheritance rather than being unsubstantiated ideas.

Mendel's work :

Gregor Mendel, conducted **hybridisation experiments** on **garden peas** for **seven years** (1856-1863) and proposed the laws of inheritance in living organisms.

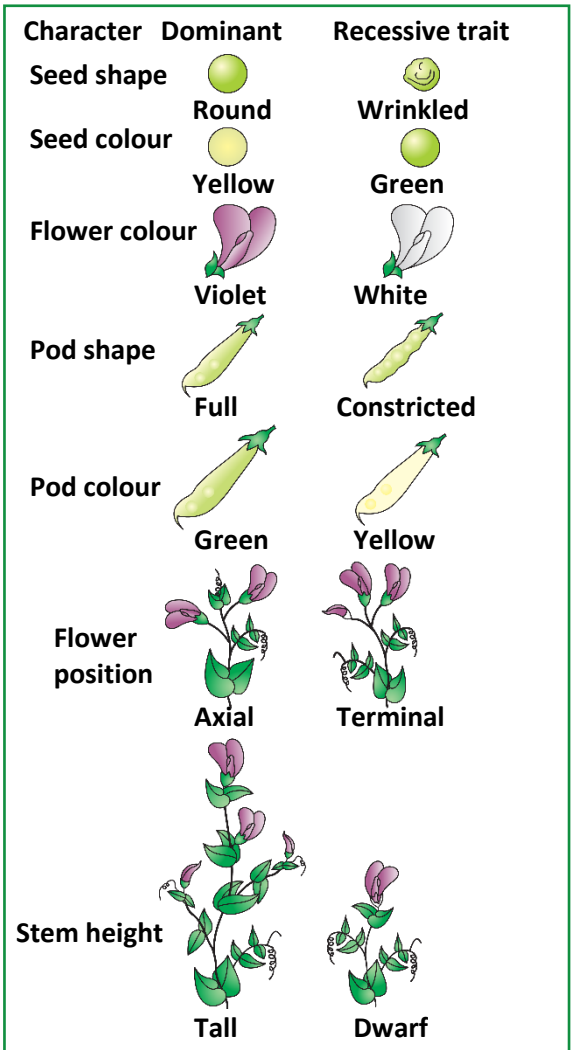


Fig. 4.1 Seven pairs of contrasting traits In pea plant studied by Mendel

Table 4.1 Mendel studied 7 characters or 7 pairs of contrasting traits in pea (*Pisum sativum*)

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

Technique of Mendel :

- Mendel conducted such **artificial pollination/cross pollination** experiments using several true-breeding pea lines.
- A true breeding 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.
- Flowers of pea plant are bisexual. In this method one is 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.

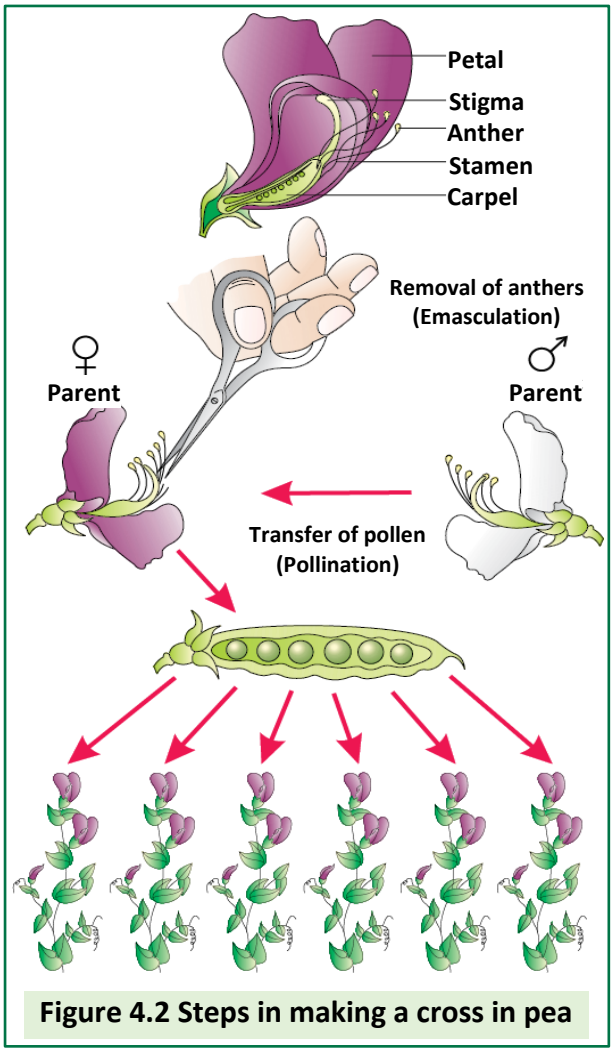


Figure 4.2 Steps in making a cross in pea

- Mature pollen grains are collected from male plants and spread over emasculated flower.
- Seeds are formed in the female flower after pollination.
- Seeds collected and grow them to generate plants.
- According to Mendel the plants that are obtained from these seeds are called First **Filial progeny** or the **F_1**.
- The plants of F_1 generation are self-pollinated & F_2 generation is produced.

04. MONOHYBRID CROSS (INHERITANCE OF ONE GENE)

- 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.

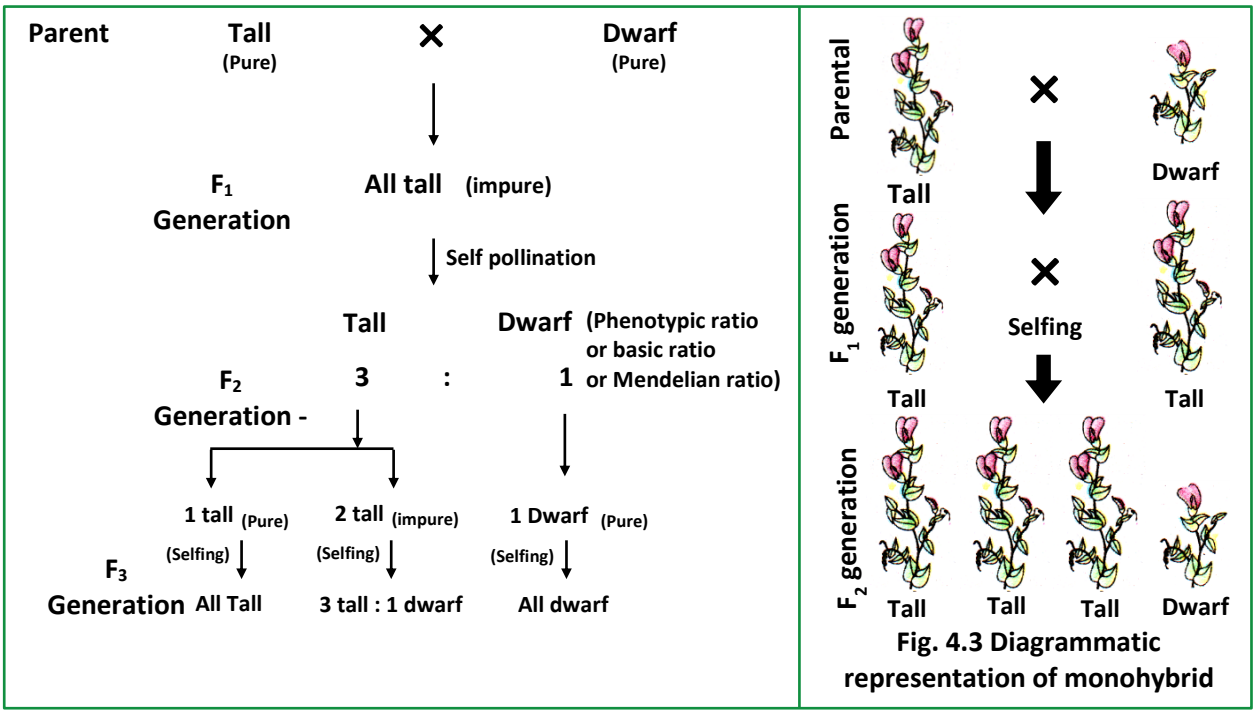
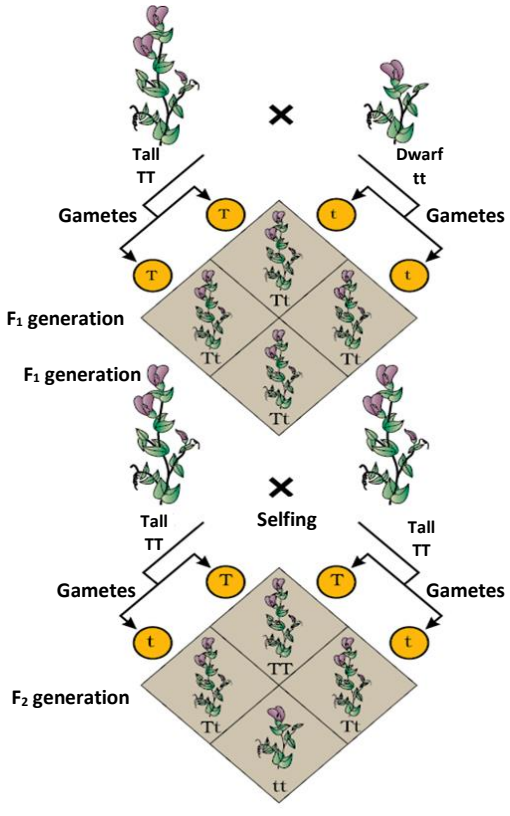


Fig. 4.3 Diagrammatic representation of monohybrid

Checker Board Method / Punnett Square :

- First time, it was used by **Reginald. C. Punnett (1875 - 1967) a British geneticist.**
- 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 : Dwarf
3 : 1
Genotypic ratio : TT : Tt : tt
1 : 2 : 1



Phenotypic ratio : tall : dwarf = 3 : 1
Genotypic ratio : TT : Tt : tt = 1 : 2 : 1

Fig 4.4 A Punnett square used to understand a typical monohybrid cross conducted by Mendel between true-breeding tall plants and true-breeding dwarf plants

NCERT Question :

Explain the Law of Dominance using a monohybrid cross.

Conclusions (results) of Monohybrid Cross :

(I) Conclusion (Postulate of paired factors)

(II) Conclusion (Postulate of Dominance)

(III) Conclusion (Law of segregation)

(I) Conclusion (postulate of paired factors) :

According to Mendel :

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

(II) 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 cannot express and it is known as conclusion of dominance.

Law of Dominance

(i) Characters are controlled by discrete units called factors.

(ii) Factors occur in pairs.

(iii) 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.

Law of dominance = I-Conclusion + II-Conclusion.

There are two exceptions of law of dominance.

[A] Incomplete dominance

[B] Co-dominance.

(III) Conclusion (Law of Segregation) :

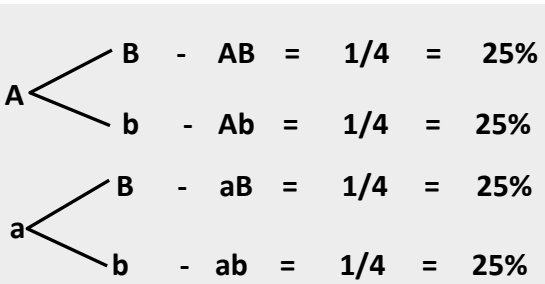
- This law is based on the fact that the alleles do not show any blending and that both the characters are recovered as such in the F_2 generation though one of these is not seen at the F_1 stage.

- During gamete formation; the unit factors of a pair segregate randomly and transfer inside different gametes. Each gamete receives only one factor of a pair; so, gametes are pure for a particular trait. It is known as conclusion of purity of gametes or segregation.
- There is no exception of Law of segregation. The segregation is essential during the meiotic division in all sexually reproducing organisms.

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



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

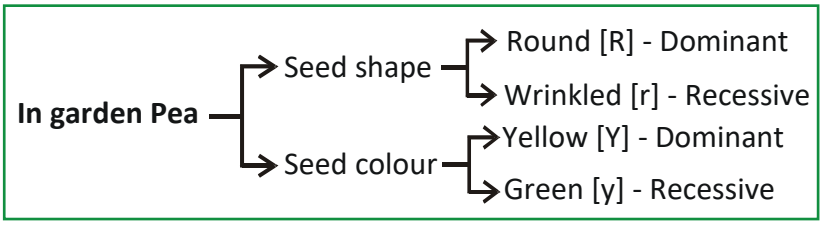
Types of genotype in $F_2 = 3^n$

Number of zygotes produced in $F_2 = 4^n$

n = Number of hybrid character or heterozygous pairs of genes.

05. DIHYBRID CROSS (INHERITANCE OF TWO GENES)

- A cross in which there is study of inheritance of **two pairs of contrasting traits**.
- Mendel wanted to observe if there any influence of inheritance of one character on the inheritance pattern of other character.
- Mendel selected two characters :



- Mendel crossed, pure 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.**

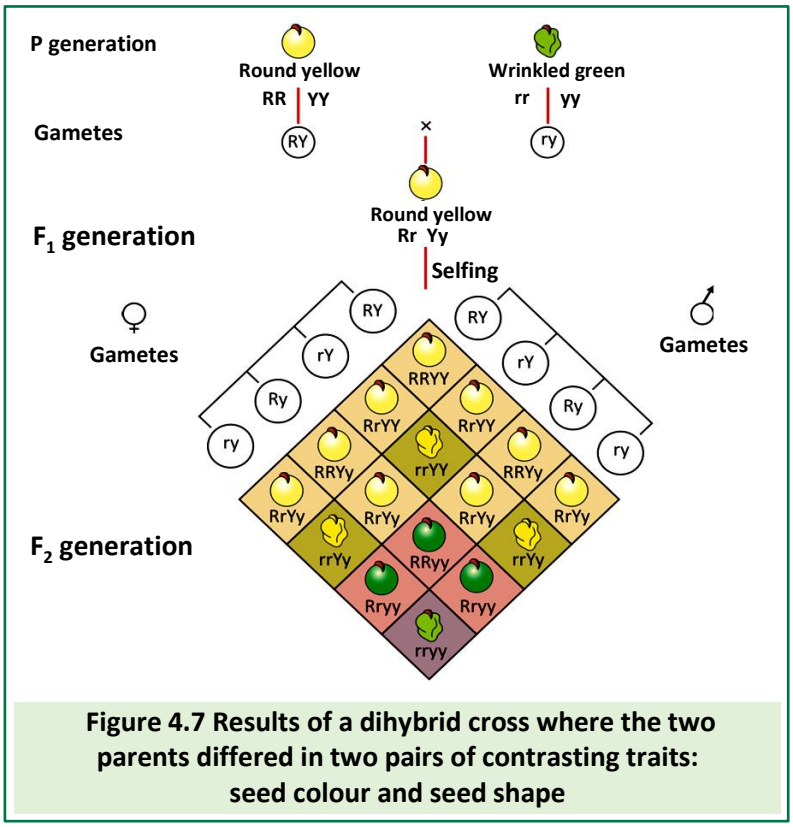


Figure 4.7 Results of a dihybrid cross where the two parents differed in two pairs of contrasting traits: seed colour and seed shape

- **Phenotypic ratio :**

  Round, yellow : Round, green : Wrinkled, yellow : Wrinkled, green

  9 3 3 1

  Types of phenotype (2^2) = 4

- **Genotypic ratio :**

  RRYY : RRYy : RRyy : RrYY : RrYy : Rryy : rrYY : rrYy : rryy

  1 2 1 2 4 2 1 2 1

  Types of genotypes (3^2) = 9

Parental combinations	:	**New combinations**
9 [Round, yellow] + 1 [Wrinkled, green]	:	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) : -

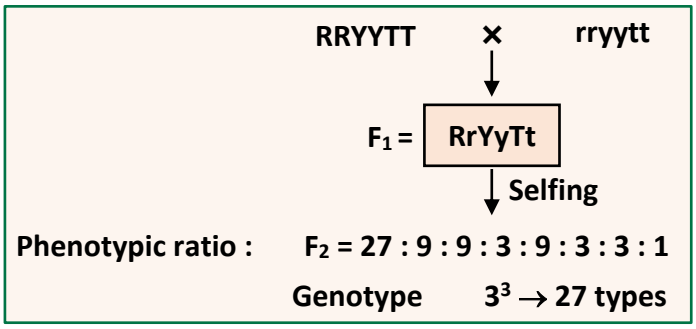
- The law states that "When two pairs of trait [2 different characters] are combined in a hybrid, segregation of one pair of character is independent of the other pair of character.
- "This is known as **Conclusion of Independent Assortment**. It is based on F_2 - generation of dihybrid cross.
- Exception : **Linkage.**

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 vegetatively propagated crops.
- Hybrid vigour can be lost by inbreeding (selfing) because inbreeding induces the Homozygosity in offsprings. Loss of Hybrid vigour due to inbreeding, is called as **inbreeding depression**.

06. TRIHYBRID CROSS

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

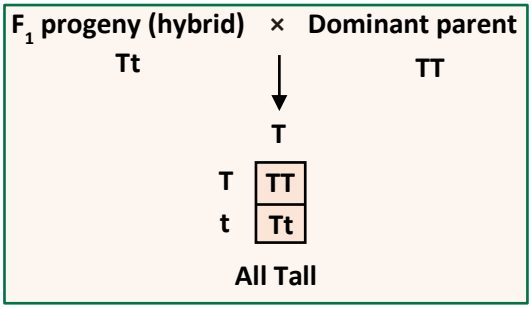


07. BACK CROSS

- 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**.
- All the progeny obtained from this cross possess dominant character. So, any analysis is not possible in F_1 generation.

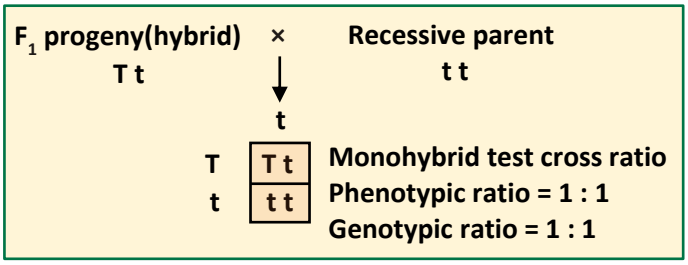


NCERT Question :

Define and design a test-cross.

(2) TEST CROSS :

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

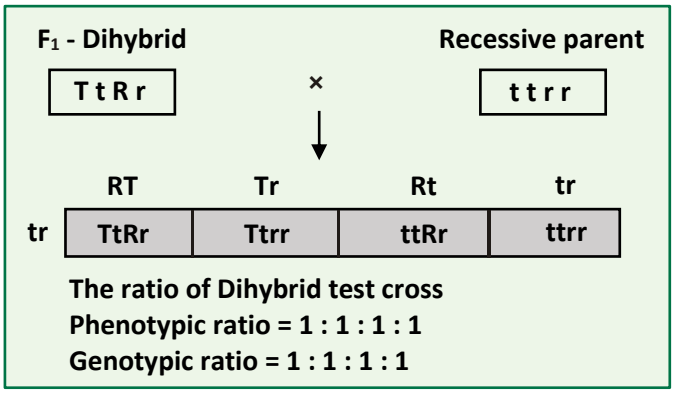


(A) Monohybrid Test Cross :

- 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.

(B) Dihybrid Test Cross :

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



Conclusion : In test cross phenotypes and genotypes ratio are same. **Test cross helps to find out the genotype of individual with dominant trait, because by simply looking at the phenotype of a dominant trait, it is not possible to know the genotypic composition.**

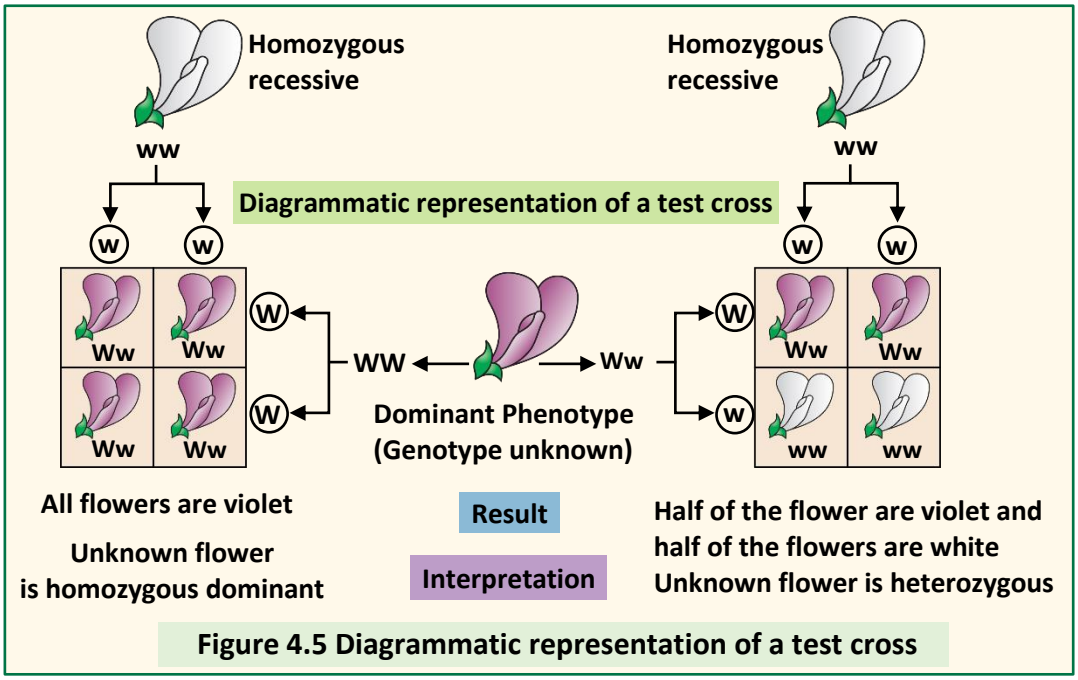
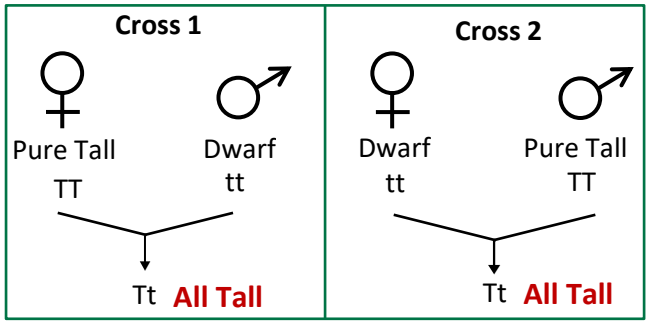


Figure 4.5 Diagrammatic representation of a test cross

08. 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.



- **Result :** Mendel obtained similar result in both cross.
- **Conclusion :** Inheritance of characters is not dependent on parental sex.
- In case of cytoplasmic inheritance and sex linkage, result will change by reciprocal cross.

★ 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-1 — 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

BPV499

2. Which law of Mendel is universal in nature?

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

(3) Law of segregation (4) Linkage

BPV500

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

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

BPV501

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.

BPV502

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

BPV503

6. A character which is expressed in a hybrid is called :-
(1) Dominant (2) Recessive (3) Co-dominant (4) Epistatic

BPV504

7. Alleles are :-
(1) Alternate forms of a gene (2) Homologous chromosome
(3) Pair of sex chromosome (4) None of these

BPV505

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

BPV506

9. How many types of gametes are produced if genotype is ttRryy :-
(1) 8 (2) 6 (3) 4 (4) 2

BPV507

10. In a dihybrid cross between RRYY and rryy the ratio of rryy, rrYy, RrYy, RRYY :-
(1) 9 : 3 : 3 : 1 (2) 4 : 2 : 1 : 1 (3) 1 : 2 : 4 : 1 (4) 1 : 4 : 2 : 1

BPV508

09. GENE INTERACTION

- Mendal's work allowed him to set up a basic framework of rules governing inheritance, which was expanded on by later scientists to account for all the diverse natural observations and the complexity inherent in them.

ALLELIC INTERACTION OR INTRAGENIC INTERACTION :

- Allelic interaction takes place between allele of same gene which are present at same locus. Examples of Allelic interaction are as follows :-

NCERT Question :

Explain the following terms with example :

(a) Co-dominance

(b) Incomplete dominance

(A) 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 factor is unable to express its character completely, resulting in intermediate type of generation which is different from the both parents. Some examples are: -

(i) 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.**

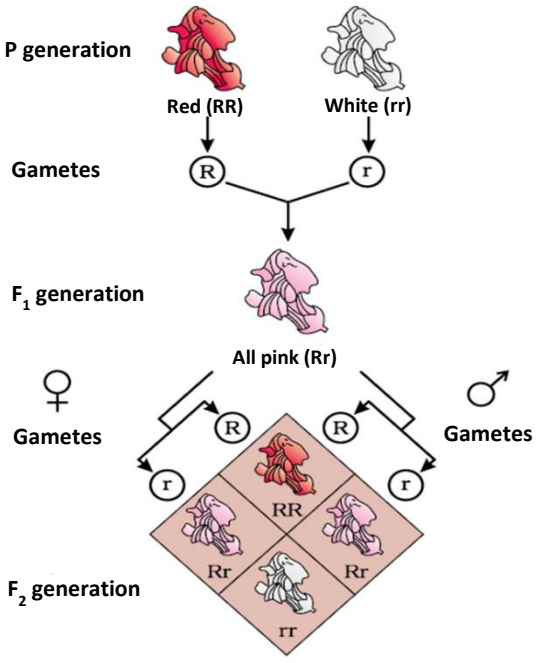


Figure 4.6 **Results of monohybrid cross in the plant Snapdragon, where one allele is incompletely dominant over the other allele**

(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 grain in pea plant :-

- When a plant with large size starch grain, is crossed with a plant with small size starch grain, the size of starch grain in F_1 generation is intermediate.

(B) Co-Dominance :

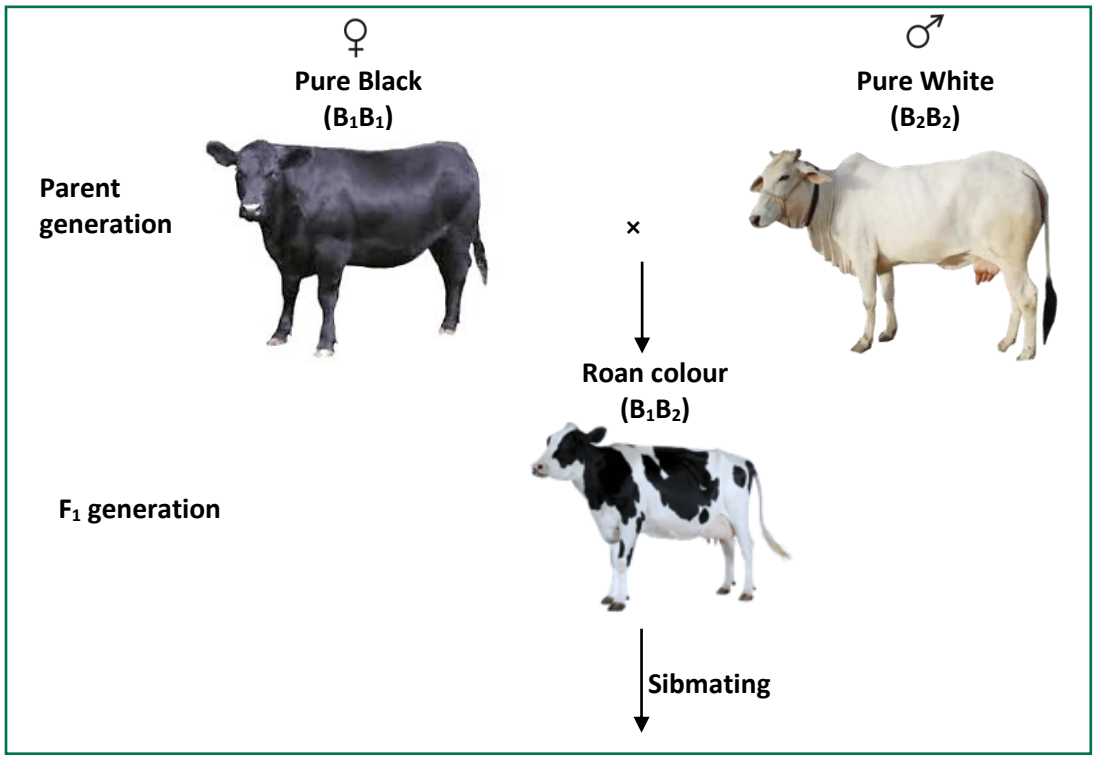
- In this phenomenon, both the alleles of a gene express for a particular character in F_1 progeny. There is no blending of characters, whereas both the traits of a character expressed equally.

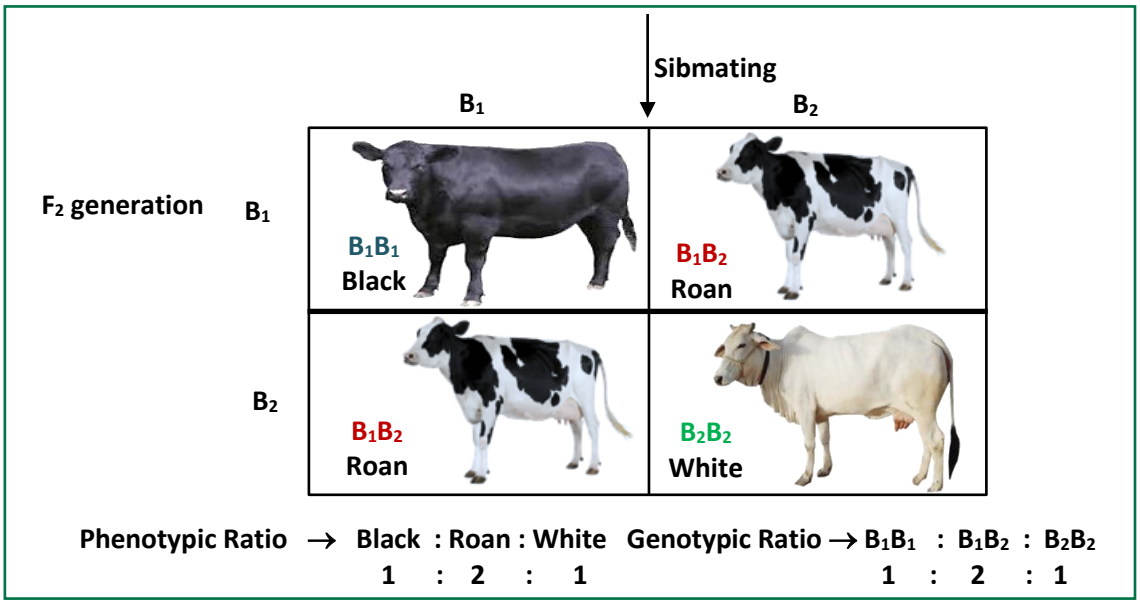
Examples :-

(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.**





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

Note : In incomplete dominance, characters are **blended phenotypically**, while in co-dominance, 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 ($Hb^A\ Hb^S$)

(C) Multiple Alleles:

- More **than 2 alternative forms of same gene** are called as multiple alleles.
- Multiple alleles are formed due to mutation. **Multiple alleles** 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 alleles of a gene then number of different possible genotype = $\frac{n(n+1)}{2}$

Example of multiple alleles :

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

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

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

(D) Pleiotropic Gene :

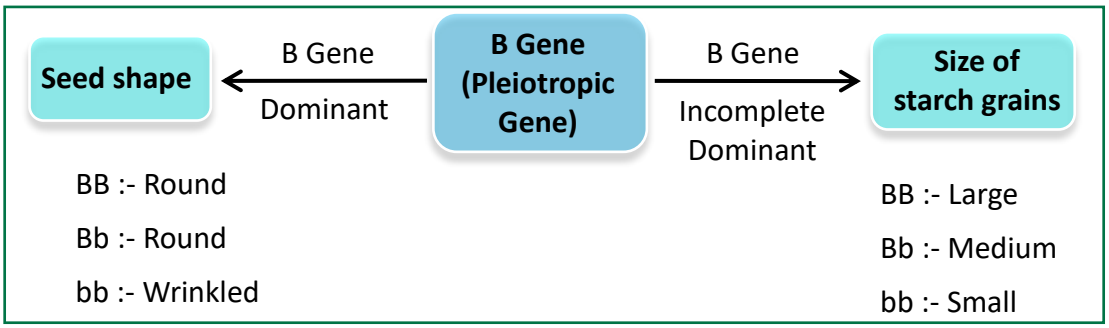
- Gene which controls more than one character is called pleiotropic gene. This gene shows **multiple phenotypic effect**.
- We have so far seen the effect of a gene on a single phenotype or trait. There are however instances where a single gene can exhibit multiple phenotypic expression. Such a gene 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. An example of this is the disease phenylketonuria, which occurs in humans. The disease is caused by mutation in the gene that codes for the enzyme phenyl alanine hydroxylase (single gene mutation). This manifest itself through phenotypic expression characterised by mental retardation and a reduction in hair and skin pigmentation.

Example :

(i) In Pea plant :-

- Seed shape and size of starch grain are controlled by same gene located on 7^{th} chromosome.
- Occasionally, a single gene product may produce more than one effect. For example, starch synthesis and seed shape in pea are controlled by one gene (B). 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.



(ii) Sickle cell anaemia :-

- This is an autosome linked recessive trait that can be transmitted from parents to the offspring when both the partners are carrier for the gene (or heterozygous).
- Gene Hb^S provide a classical example of **pleiotropy**. 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 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**.

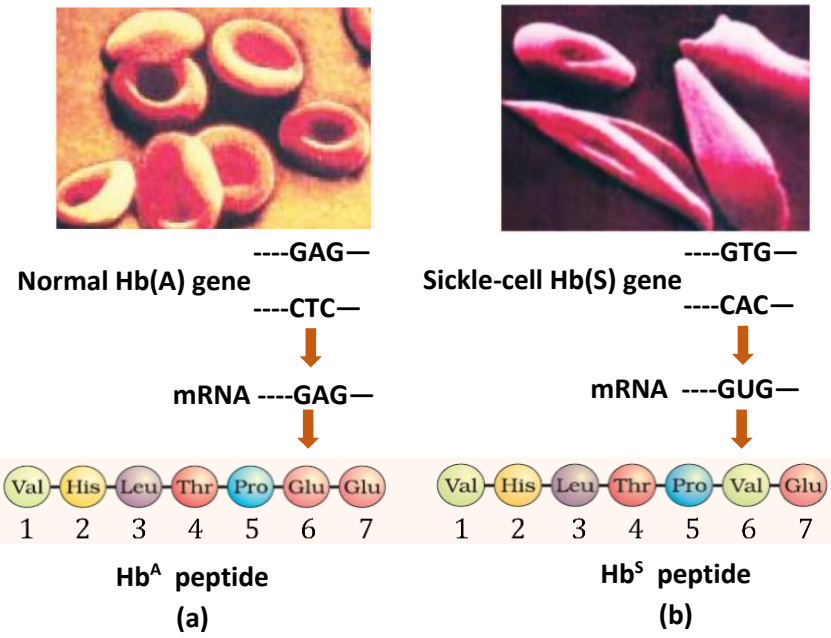


Fig. 4.15 Micrograph of the red blood cells and the amino acid composition of the relevant portion of β-chain of haemoglobin
(a) From a normal individual;
(b) From an individual with sickle-cell anaemia

10. POLYGENIC INHERITANCE OR 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. 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.
- Inheritance of characters in which one character is controlled by many genes and intensity of character depends upon the number of dominant alleles.
- Types of phenotypes (phenotypic categories) in F_2 generation = 2n + 1
  Here n = number of polygenes

Example-1:- Kernel (grain) colour of wheat is regulated by two polygenes.

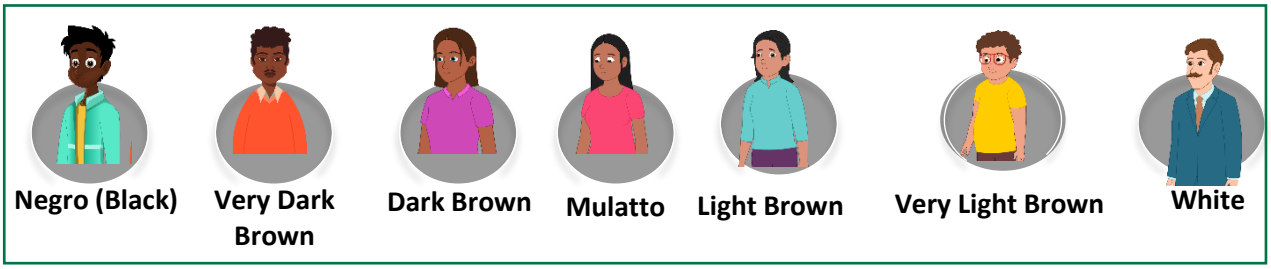
AABB (Red) X aabb (White)

F_1 → Aa Bb (Intermediate)

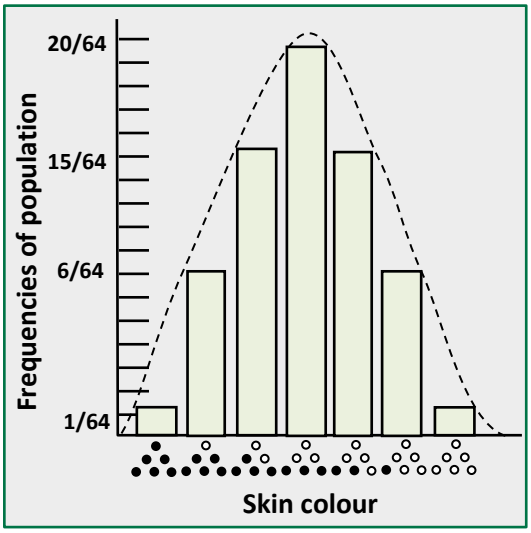
Phenotype F_2	1	:	4	:	6	:	4	:	1
	Full Red		Light Red		Intermediate Red		Very light Red		Full White
No. of Dominant allele	4		3		2		1		0
	AABB-1		AaBB-2 AABb-2		AaBb-4 aaBB-1 AAbb-1		aaBb-2 Aabb-2		aa bb-1

1	:	14	:	1
Full red		Intermediate		Full white

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**.
- 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.

- 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 AA BB CC X **White aa bb cc**

↓

Aa Bb Cc → F_1 - generation

[Intermediate brown/mulatto]

↓ **(Inbreeding)**

F_2-generation

↓

Phenotypic ratio will be :-

Negro Black	:	Very Dark Brown	:	Dark Brown	:	Mulatto	:	Light Brown	:	Very Light Brown	:	White
1	:	6	:	15	:	20	:	15	:	6	:	1

11. CYTOPLASMIC INHERITANCE

- Inheritance of characters which are controlled by **cytogene** or cytoplasmic gene is called cytoplasmic inheritance. Genes which are present in cytoplasm called **'cytogene'** or **'plasmagene'** or **extra nuclear gene**.
- A gene which is located in the nucleus is called **'karyogene'.**
- Inheritance of cytogene occurs only through the female. **(so it is also called as maternal inheritance)**.
- Cytoplasmic inheritance involving organelles like, Chloroplast and mitochondria is called as **organellar inheritance.**

★ 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-2

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

BPV509

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

BPV510

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

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

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

BPV511

4. 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

BPV512

5. When a pea plant with round seed (Bb) is crossed with other plant having small sized starch grain in seed, the total number of seeds obtained in progeny is 630. What is correct for this progeny?

(1) 330 (large size), 330 (small size)

(2) 330 (small size), 330 (medium size)

(3) 315 (intermediate size), 315 (small size)

(4) 158 (large size), 158 (small size), 314 (intermediate size)

BPV513

6. Absence of SBE in *Pisum* leads to the development of :-

(1) Round seeds with large sized starch grains

(2) Round seeds with small sized starch grains

(3) Wrinkled seed with intermediate sized starch grains

(4) Wrinkled seed with small sized starch grains

BPV514

7. Carrier of sickle cell anaemia (Hb^A Hb^S) is an example of :

(1) Co-dominance (2) Multiple allele

(3) Incomplete dominance (4) Both (1) and (2)

BPV515

8. Multiple alleles can be found only when :-

(1) Population studies are made (2) Individual study is made

(3) Mutation is absent (4) Dominance is present

BPV516

9. In *Antirrhinum majus* when a pink flowered plant is crossed with white flowered plant then total 120 plants are obtained. What will be percentage of phenotype in offspring ?

(1) 100% red flowered (2) 50% white flowered

(3) 75% pink flowered (4) 100 pink flowered

BPV517

10. If a mulatto man marries with a mulatto woman, then :-

(1) Seven types of genotypes will be obtained.

(2) Only mulatto will be produced.

(3) Offspring with three dominant alleles will be maximum.

(4) 1 : 4 : 6 : 4 : 1 phenotypic ratio will be obtained.

BPV518

12. CHROMOSOMAL THEORY OF INHERITANCE

NCERT Question :

Who had proposed the chromosomal theory of the 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 (coloured 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.**

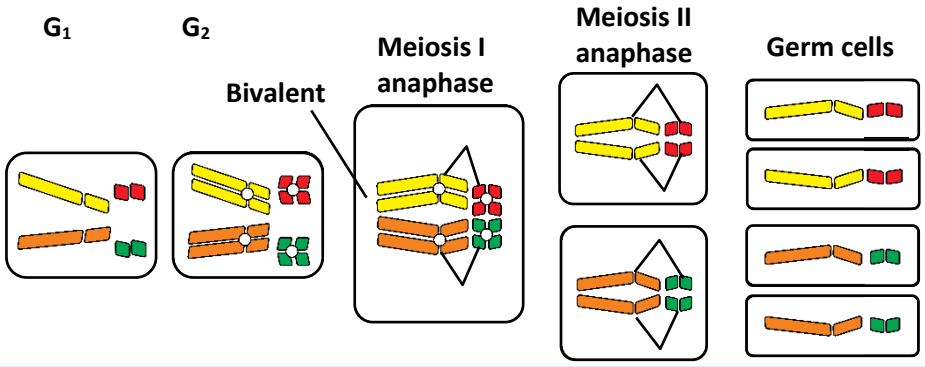


Fig. 4.8 Meiosis and germ cell formation in a cell with four chromosomes. Can you see how chromosomes segregate when germ cells are formed?

- 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.
- 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.

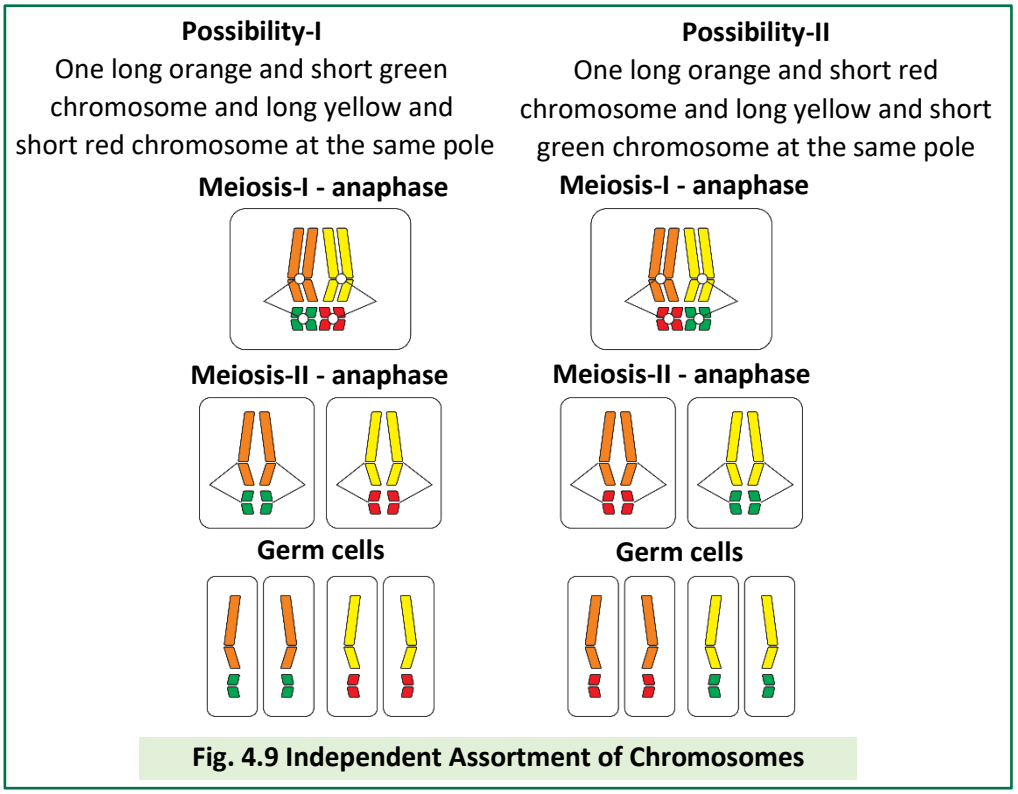


Fig. 4.9 Independent Assortment of Chromosomes

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

- 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**.

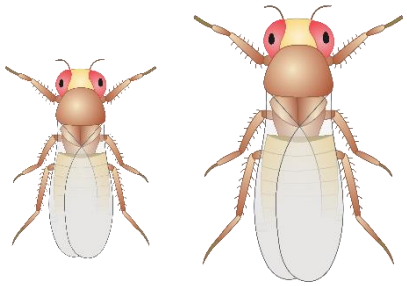


Fig. 4.10 *Drosophila melanogaster*
(a) Male (b) Female

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

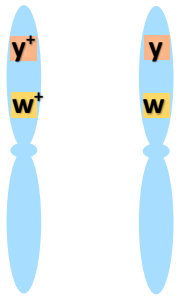
13. LINKAGE AND RECOMBINATION

- The physical association of genes on a chromosome is called linkage.
- Collective inheritance of characters is due to linkage.

NCERT Question :
Briefly mention the contribution of T.H. Morgan in genetics.

- Sex linkage was first discovered by Morgan in *Drosophila* and he coined the term linkage. He proposed the theory of linkage.
- Linkage and independent assortment can be represented in dihybrid plant, as :-

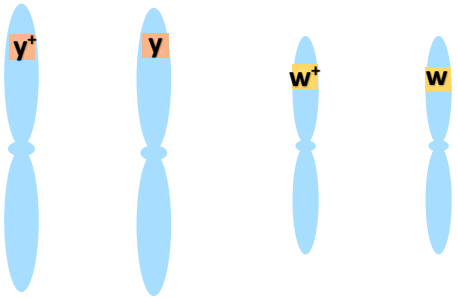
In case of linkage in dihybrid $\left(\frac{y^+w^+}{yw}\right)$



It produces two types of gametes

y^+w^+ : yw

In case of independent assortment in dihybrid (y^+yw^+w)



It produces four types of gametes

y^+w^+ : yw : yw^+ : y^+w

(1) THEORY OF LINKAGE :

(A) Linked genes are linearly located on same chromosome. They may get separated by crossing over.

(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 inherit together so we consider one linkage group.
- **Number of Linkage groups = 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
Human male	46	23	23	24
Human female	46	23	23	23

(C) **Strength of linkage** $\propto \frac{1}{\text{distance between the genes}}$

- 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.

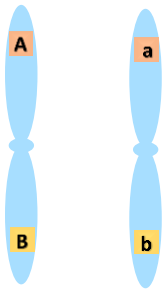
Factors affecting crossing over (C.O.) :-

(a) Distance ↑ = C.O. ↑ (b) Age ↑ = C.O. ↓ (c) Sex - Male = C.O. ↓

(2) ARRANGEMENT OF LINKED GENES ON CHROMOSOMES :

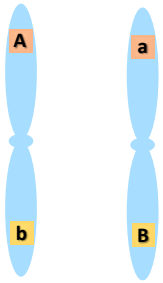
The arrangement of linked genes in any dihybrid is of two types.

(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. Two types of gametes 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 gametes are also formed in trans-arrangement but it is different from cis-arrangement - (Ab) and (aB).

(3) TYPES OF LINKAGE :

(A) 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.

(B) Incomplete Linkage :-

- When new combinations (<50%) also appear along with parental combination in offspring, this type of linkage is called incomplete linkage.
- The new combinations are formed due to crossing over.

(4) APPLICATIONS OF LINKAGE :

- Distance between linked genes can be determined by the incomplete linkage. Its unit is centi- Morgan (cM) or map units.

$$\text{Strength of linkage} \propto \frac{1}{\text{Distance between linked gene}} \propto \frac{1}{\text{Crossing Over}}$$

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.

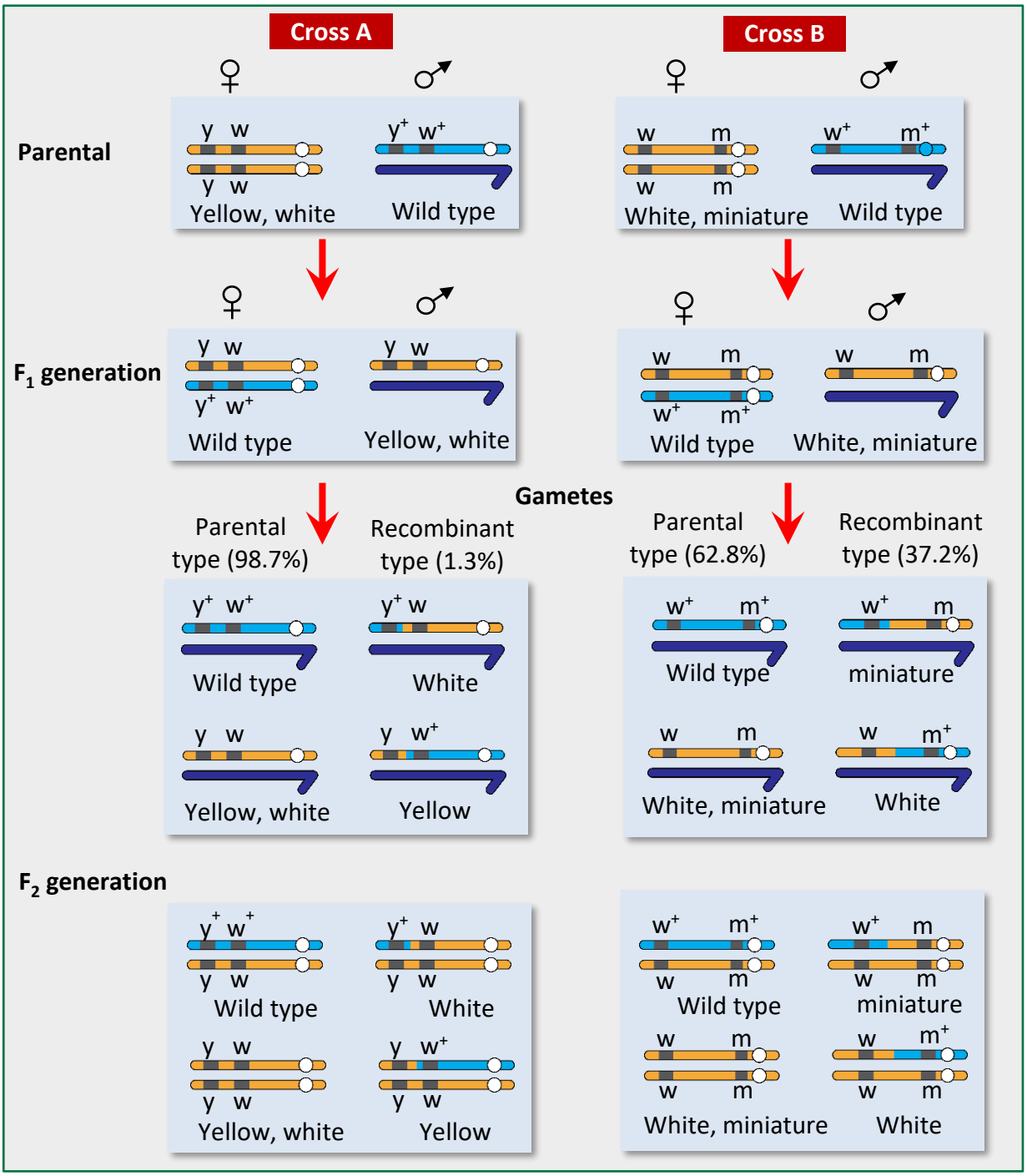


Fig. 4.11 Linkage : Results of two dihybrid crosses conducted by Morgan. Cross A shows crossing between gene y and w; Cross B shows crossing between genes w and m. Here dominant wild type alleles are represented with (+) sign in superscript.

Note : The strength of linkage between y and w is higher than w and m.

14. SEX LINKED INHERITANCE

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

Types of Sex Linkage :

(1) X-LINKAGE :

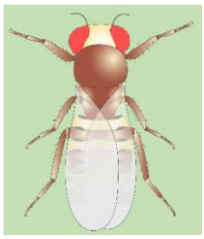
- Genes of many somatic characters are found on X-chromosome.
- The inheritance of X-linked character may be through the males and females.

Examples of X-linkage :-

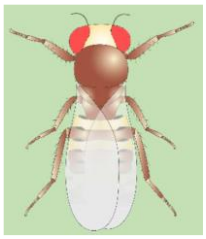
(A) Eye colour in *Drosophila* :-

- Eye colour in *Drosophila* is controlled by a X-linked gene.
- If a red eye colour gene is represented as '+' and white eyed colour represented as 'w' because gene for red eye is dominant (+) and white colour of eye is recessive (w), then on basis of this different type of genotypes are found in *Drosophila*.

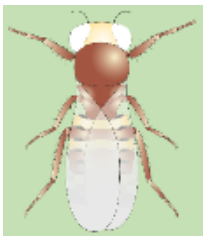
(a) Homozygous red eyed female = $X^{+} X^{+}$



(b) Heterozygous red eyed female = $X^{+} X^{w}$



(c) Homozygous white eyed female = $X^{w} X^{w}$



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

(e) Hemizygous white eyed male = $X^{w} Y$

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

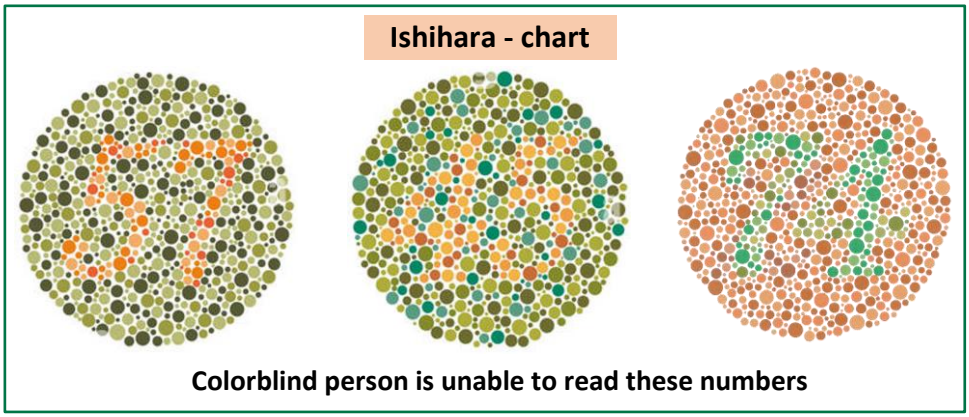
(B) Haemophilia :-

- 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-linkage, following types of genotype are found.
  X^hX = Carrier female
  X^hX^h = Affected female (dies during embryonic stage)
  X^hY = Affected male.

Haemophilia : This sex linked recessive disease, which shows its transmission from unaffected carrier female to some of the male progeny has been widely studied. In this disease, a single protein that is a part of the cascade of proteins involved in the clotting of blood is affected. Due to this, in an affected individual a simple cut will result in non-stop bleeding. The heterozygous female (carrier) for haemophilia may transmit the disease to sons. The possibility of a female becoming a haemophilic is extremely rare because mother of such a female has to be at least carrier and the father should be haemophilic (unviable in the later stage of life). The family pedigree of Queen Victoria shows a number of haemophilic descendants as she was a carrier of the disease.

(C) Colour Blindness :-

- The inheritance of colour-blindness is similar to 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:- Colour blindness 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 due to mutation in certain genes present in the X chromosome.
- It occurs in about 8 percent of males and only about 0.4 percent 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 percent chance of being colour 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

(D) Diabetes insipidus (X-linked recessive).

(E] Duchenne muscular dystrophy (X-linked recessive)

(F) Pseudorickets (X-linked Dominant)

(G) Defective enamel of teeth (X-linked 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.

Example :-

(i) Gene which forms TDF / sry-gene

(ii) 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:

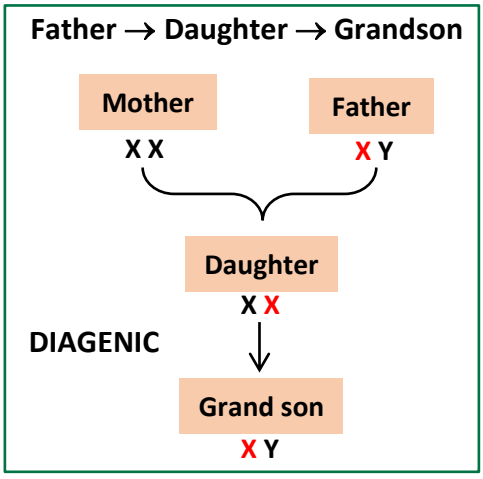
1. Criss Cross Inheritance (Morgan) :

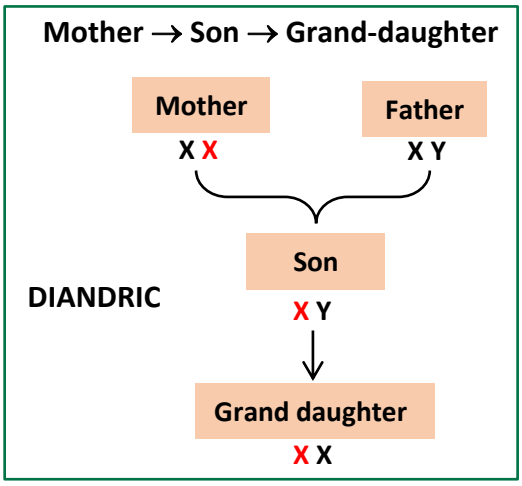
- In criss-cross inheritance male or female parent transfer a X-linked character to grandson or grand daughter through the offspring of opposite sex.

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

Father → Daughter → Grandson.

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



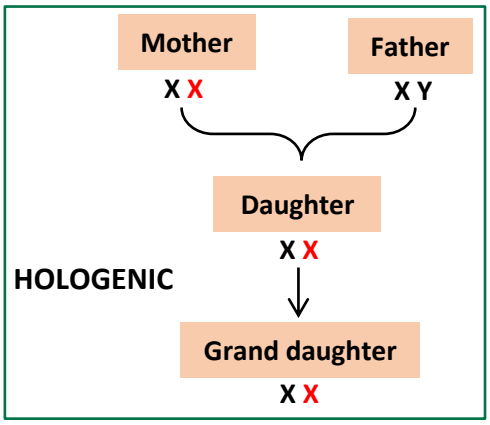


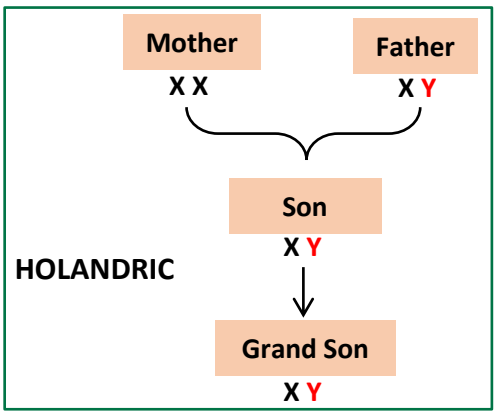
2. Non Criss-Cross Inheritance :

- In this inheritance male or female parent transfer sex linked character to grandson 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 dependent 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 is dependent upon presence or absence of sex-hormones.
- For example **: genes of beard-moustache** express their effects only in the presence of male sex 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 present	Baldness absent
bb	Baldness absent	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, haploid-diploid mechanism, genic balance, etc.

(1) SEX DETERMINATION ON THE BASIS OF FERTILIZATION :

(A) Progamic :- Sex is determined before fertilization. ***e.g.*** - drone in honey bee.

(B) Syngamic :- Sex is determined during fertilization. ***e.g.*** - most of plants & animals.

(C) Epigamic :- Sex is determined after fertilization. ***e.g.*** - crocodile and 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"** and called it 'X-body'.
- **Wilson & Stevens** proposed chromosomal theory for sex determination.

(i) XX - XY type or Lygaeus type :

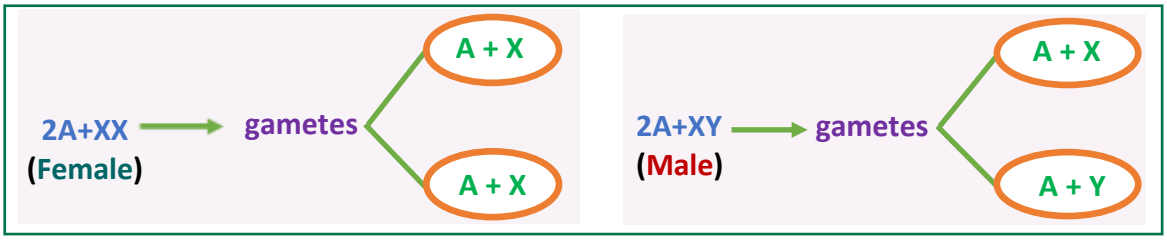
This type of sex determination first observed by **Wilson & Stevens** in Lygaeus insect. It is of two types :

NCERT Question :

How is sex determined in human beings?

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

Male is heterogametic (male produces two types of gametes)



e.g. Humans, many mammals and *Drosophila*.

(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.**

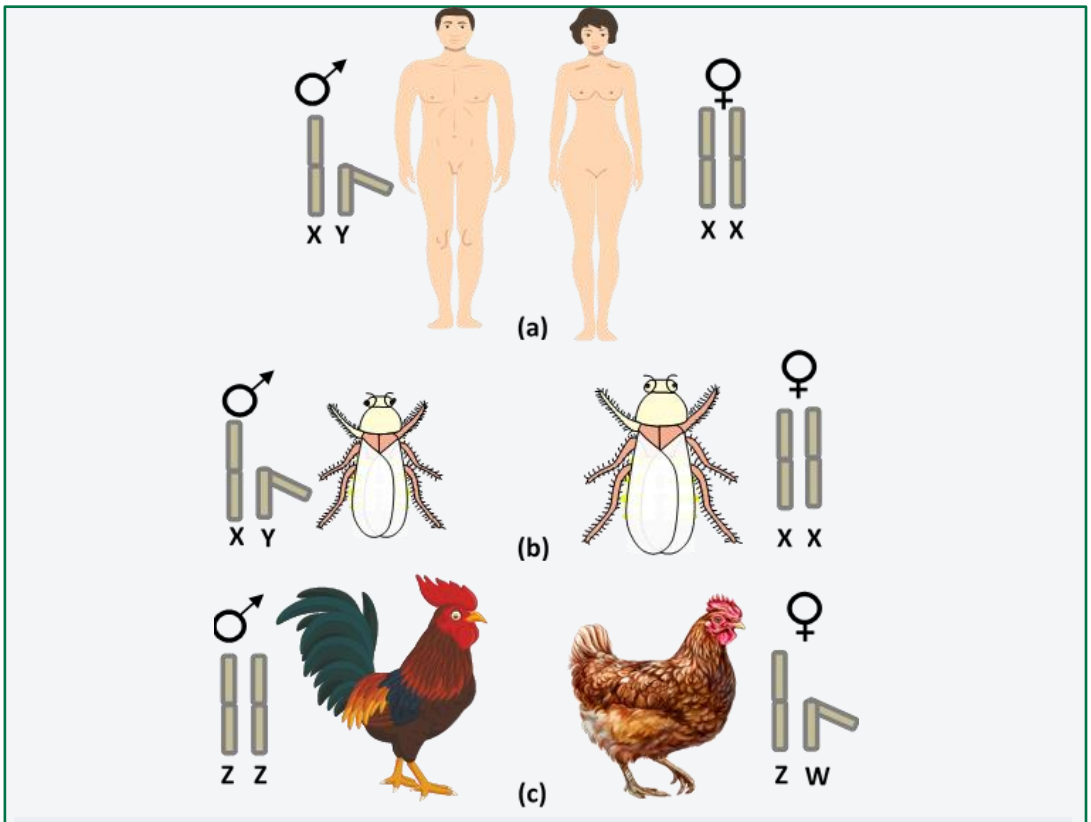
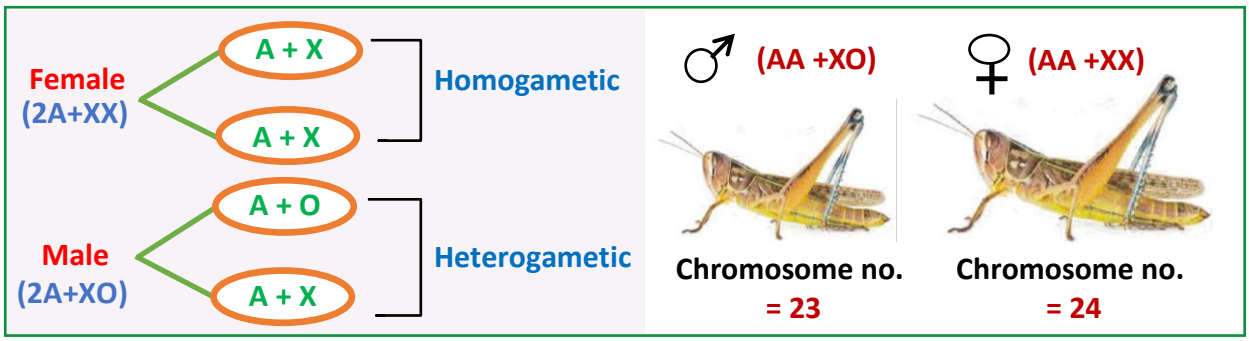


Fig. 4.12 Determination of sex by chromosomal differences:
(a, b) Both in humans and in *Drosophila*, the female has a pair of XX chromosomes (homogametic) and the male XY (heterogametic) composition; (c) In many birds, female has a pair of dissimilar chromosomes ZW and male two similar ZZ chromosomes.

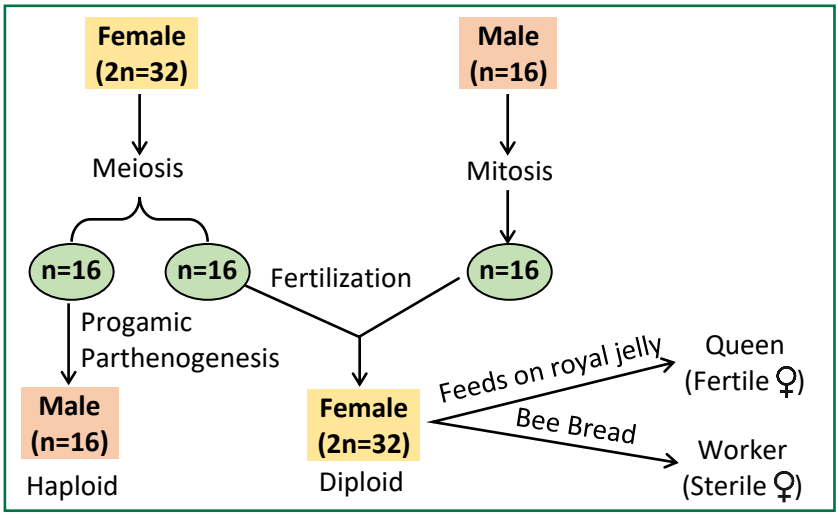
(ii) XX female and XO male or "Protenor type" :- In this type of sex determination there is deficiency of one chromosome in male. In this type, female is homogametic and male is heterogametic.



Examples : Grasshopper, Cockroach

(B) 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) by parthenogenesis. Queen and worker bees develop from diploid eggs *i.e.* fertilized egg.



- 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 chromosomes than that of a female.
- The females are diploid having 32 chromosomes as haplodiploid 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.

BEGINNER'S BOX-3
CHROMOSOMAL THEORY OF INTERITANCE TO SEX DETERMINATION

1. An exception to the law of independent assortment is :-
(1) Dominance (2) Incomplete dominance
(3) Segregation (4) Linkage
BPV519

2. Experimental proof for sex-linked gene was given by :-
(1) Morgan (2) Muller (3) Mendel (4) Johannsen
BPV520

3. X-linked recessive gene is easily expressed in :-
(1) male (2) female
(3) equal in male and female (4) not easily expressed
BPV521

4. Maize has 10 pairs of chromosomes. How many linkage groups are present.
(1) 05 (2) 10 (3) 20 (4) 40
BPV522

5. The mechanism of sex determination in birds shows :-
(1) Male heterogamety (2) Both heterogametic
(3) Female heterogamety (4) Both homogametic
BPV523

6. Crossing over is advantageous because it brings about :-
(1) Variation (2) Linkage (3) Inbreeding (4) Stability
BPV524

7. Mendel's law of independent assortment does not hold true for the genes that are located closely on:-
(1) Same chromosome (2) Non-homologous chromosomes
(3) Different chromosomes (4) Autosomes
BPV525

8. Position of genes on chromosomes was mapped for the first time by:
(1) Muller (2) Alfred Sturtevant
(3) C. Bridges (4) Bateson
BPV526

9. Colour-blindness is :-
(1) Sex-linked recessive (2) Sex-linked dominant
(3) Autosomal recessive (4) Autosomal dominant
BPV527

10. '*Drosophila* is very suitable for genetic studies' because :-
(1) Shorter life cycle
(2) No clear differentiation of sexes
(3) Single mating produces less number of offspring
(4) No hereditary variations are present
BPV528

18. 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 :-

- Cells of human body are relatively smaller and number of chromosomes present in them is more.
- It is not possible to perform various experiments on humans in the laboratory.
- Due to greater life span of humans, a lot of time is required to study their genetic characteristics.
- Rate of reproduction is slow in humans.
- Individuals of human species are generally heterozygous for various characters and it is very difficult to find homozygous individuals.
- 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:-

- Longer life span of humans, the abnormalities that require relatively long period to express themselves can be easily studied.
- By pedigree study and analysis of families, many genetic characters of human can be traced out.
- 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
Rh Factor	Rh^+	Rh^-

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 one, that is, pedigree analysis is being described here.

19. GENETIC DISORDERS

NCERT Question :

What is pedigree analysis? Suggest how such an analysis, can be useful.

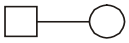
(1) PEDIGREE ANALYSIS :

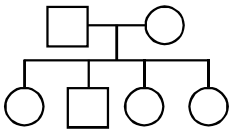
- Study of ancestral history of man of transmission of genetic characters from one generation to next, is pedigree analysis. Dwarfism, albinism, colour blindness, haemophilia etc. are genetically transmitted characters. To study and analyse them a pedigree of genetic facts/data and following symbols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

Symbol used in Pedigree analysis:

1.  Normal Male

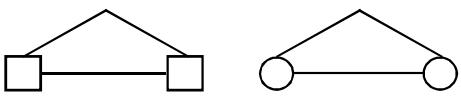
2.  Normal Female

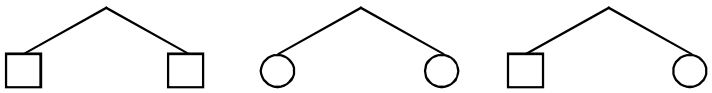
3.  Mating (marriage)

4.  The siblings are indicated in chronological order of birth

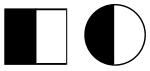
5.  Sex unspecified

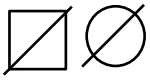
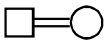
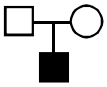
6. Twins

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 information regarding genetic makeup of human beings.
- If any genetic disease is occurring in a family, then pedigree analysis provides guidance to forthcoming parents about their future progenies for example- polydactyly in humans.

Examples :-

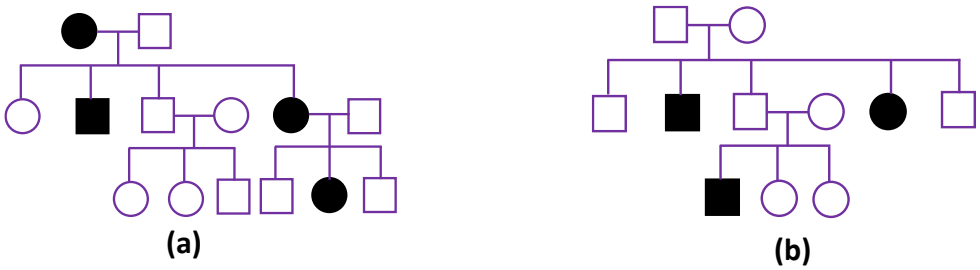


Fig. 4.14 Representative pedigree analysis of :

(a) Autosomal dominant trait (Myotonic Dystrophy)

(b) Autosomal recessive trait (Sickle Cell Anaemia)

(2) MENDELIAN DISORDERS :

- Broadly, genetic disorders may be grouped into two categories – **Mendelian disorders** and **Chromosomal disorders**. Mendelian disorders are mainly determined by alteration or mutation in the single gene. These disorders are transmitted to the offspring on the same lines as we have studied in the principle of inheritance. The pattern of inheritance of such Mendelian disorders can be traced in a family by the pedigree analysis.
- Most common and prevalent Mendelian disorders are Haemophilia, Cystic fibrosis, Sickle cell anaemia, Colour blindness, Phenylketonuria, Thalassemia, etc. It is important to mention

here that such Mendelian disorders may be dominant or recessive. By pedigree analysis one can easily understand whether the trait in question is dominant or recessive. Similarly, the trait may also be linked to the sex chromosome as in case of haemophilia. It is evident that this X-linked recessive trait shows transmission from carrier female to male progeny.

NCERT Question :

Mention any two autosomal genetic disorders with their symptoms.

Phenylketonuria :

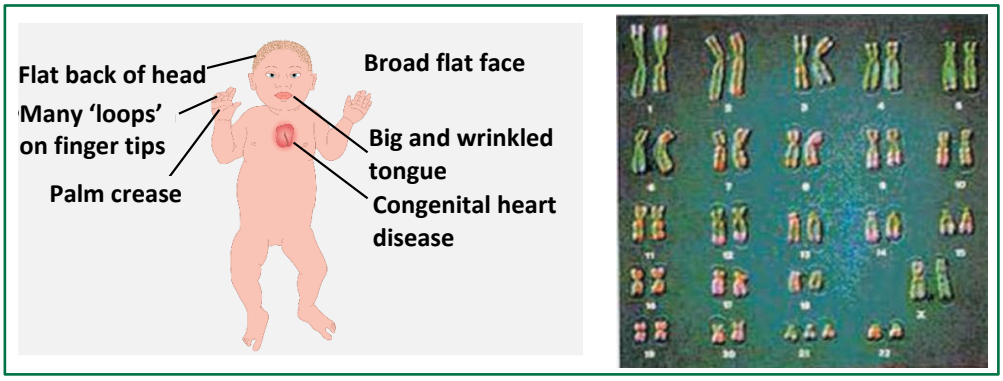
- This inborn error of metabolism is also inherited as the **autosomal recessive trait.**
- The affected individual lacks an enzyme (phenylalanine hydroxylase) that converts the amino acid **phenylalanine into tyrosine.**
- As a result of this phenylalanine is accumulated and converted into phenylpyruvic acid and other derivatives. Accumulation of these in brain results in **mental retardation.** These are also excreted through urine because of its poor absorption by kidney.

Thalassemia :

- This is also an **autosome-linked recessive blood disease** transmitted from parents to the offspring when both the partners are unaffected carrier for the gene (or heterozygous).
- The defect could be due to either mutation or deletion which ultimately results in reduced rate of synthesis of one of the globin chains (α and β chains) that make up haemoglobin.
- This causes the formation of abnormal haemoglobin molecules resulting into anaemia which is characteristic of the disease.
- Thalassemia can be classified according to which chain of the haemoglobin molecule is affected. In **α-Thalassemia,** production of α globin chain is affected while in **β-Thalassemia,** production of β-globin chain is affected.
- α-Thalassemia is controlled by two closely linked genes **HBA1 and HBA2 on chromosome 16** of each parent and it is observed due to mutation or deletion of one or more of the four genes. The more genes affected, the less alpha globin molecules produced.
- While **β-Thalassemia is controlled by a single gene HBB on chromosome 11** of each parent and occurs due to mutation of one or both the genes.
- Thalassemia differs from sickle-cell anaemia in that the former is a **quantitative** problem of synthesising too few globin molecules while the latter is a **qualitative problem** of synthesising an incorrectly functioning globin.

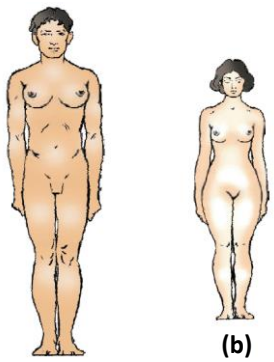
(3) CHROMOSOMAL DISORDERS :

- The chromosomal disorders on the other hand are caused due to absence or excess or abnormal arrangement of one or more chromosomes.
- Failure of segregation of chromatids during cell division cycle results in the gain or loss of a chromosome(s), **called aneuploidy.**
- For example, **Down's syndrome** results in the gain of extra copy of **chromosome 21.** Similarly, **Turner's syndrome** results due to **loss of an X chromosome** in human females.
- **Failure of cytokinesis after telophase** stage of cell division results in an increase in a whole set of chromosomes in an organism and, this phenomenon is known as **polyploidy.** This condition is often seen in plants. The total number of chromosomes in a normal human cell is 46 (23 pairs). Out of these 22 pairs are autosomes and one pair of chromosomes are sex chromosome. Sometimes, though rarely, either an additional copy of a chromosome may be included in an individual or an individual may lack one of any one pair of chromosomes. These situations are known as trisomy or monosomy of a chromosome, respectively. Such a situation leads to very serious consequences in the individual. Down's syndrome, Turner's syndrome, Klinefelter's syndrome are common examples of chromosomal disorders.
- **Down's Syndrome:** The cause of this genetic disorder is the presence of an additional copy of the chromosome number 21 (trisomy of 21). This disorder was first described by Langdon Down (1866). The affected individual is short statured with small round head, furrowed tongue and partially open mouth. Palm is broad with characteristic palm crease. Physical, psychomotor and mental development is retarded.



A representative figure showing an individual inflicted with Down's syndrome and the corresponding chromosomes of the individual.

- **Klinefelter's Syndrome:** This genetic disorder is also caused due to the presence of an additional copy of X- chromosome resulting into a karyotype of **47, XXY**. Such an individual has overall masculine development, however, the feminine development (development of breast, i.e., Gynaecomastia) is also expressed. Such individuals are sterile.
- **Turner's Syndrome:** Such a disorder is caused due to the absence of one of the X chromosomes, i.e., **45 with XO**. Such, females are sterile as ovaries are rudimentary besides other features including lack of other secondary sexual characters.



(a) Tall stature with feminised character

(b) Short stature and underdeveloped feminine character

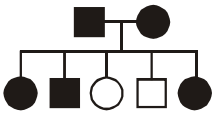
NCERT Fig. 4.17 Diagrammatic representation of genetic disorders due to sex chromosome composition in humans : (a) Klinefelter Syndrome; (b) Turner's Syndrome

BEGINNER'S BOX-4 HUMAN GENETICS TO CHROMOSOMAL DISORDER

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

(1) □ = unaffected female
(2) ◇ = affected male
(3) ■ = affected female
(4) ● = affected female

BPV529

2.  The pedigree shows:-

(1) Dominant inheritance
(2) Recessive inheritance
(3) Sex linked recessive inheritance
(4) Cytoplasmic inheritance

BPV530

3. A gene located on Y-chromosome and transmitted from father to son is known as :-

(1) Epistatic gene
(2) Hypostatic gene
(3) Complementary gene
(4) Holandric gene

BPV531

4. PKU in humans occurs due to absence of which of the following enzyme :-

(1) Homogentisic acid oxidase

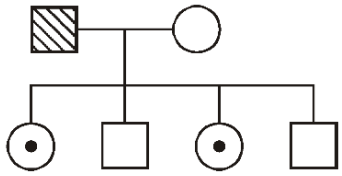
(2) Tyrosinase

(3) Phenylalanine hydroxylase

(4) More than one option is correct

BPV532

5. Given pedigree chart shows which type of inheritance :-



(1) Autosomal recessive (Albinism)

(2) X-linked recessive (color blindness)

(3) Y-linked

(4) Autosomal dominant

BPV533

6. Match the following columns :-

Column I		Column II	
A.		P.	Death
B.		Q.	Five unaffected offspring
C.		R.	Sex unspecifed
D.		S.	Female
E.		T.	Male

(1) A → P; B → Q; C → T; D → S; E → R

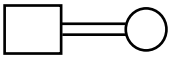
(2) A → P; B → Q; C → S; D → R; E → T

(3) A → P; B → Q; C → R; D → S; E → T

(4) A → T; B → S; C → R; D → Q; E → P

BPV534

7. Which one is the incorrect match ?

(1)		Consanguineous mating
(2)		Sex unspecified
(3)		Male
(4)		Affected individuals

BPV535

8. Klinefilter syndrome has the genetic make up :-

(1) 44 autosomes + XXY

(2) 44 autosomes + XO

(3) 45 autosomes + XX

(4) 45 autosomes + XY

BPV536

9. In a family, father is affected by Down syndrome and mother is normal then what will be percentage of affected offspring.

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

BPV537

BEGINNER'S BOX — ANSWERS KEY

BEGINNER'S BOX-1	Que.	1	2	3	4	5	6	7	8	9	10
	Ans.	3	3	1	4	2	1	1	2	4	3

BEGINNER'S BOX-2	Que.	1	2	3	4	5	6	7	8	9	10
	Ans.	2	3	3	4	3	4	1	1	2	3

BEGINNER'S BOX-3	Que.	1	2	3	4	5	6	7	8	9	10
	Ans.	4	1	1	2	3	1	1	2	1	1

BEGINNER'S BOX-4	Que.	1	2	3	4	5	6	7	8	9	
	Ans.	4	1	4	3	2	4	3	1	1	

Parents characters ⟶ Progeny

Inheritance of characters

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

Back cross

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

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.

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

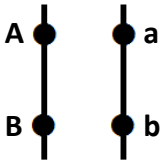
(2) Epistasis
- A gene prevents expression of another non-allelic gene
- Dominant epistasis
  Phenotype – 12 : 3 : 1

Linkage → Physical association of genes on a chromosome

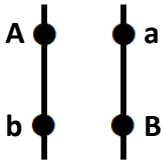
- Linkage group
  - All the genes which are located on one pair of homologous chromosomes form one linkage group.
  - Number of linkage groups = Haploid no. of chromosomes
  - Linked genes can separate by crossing over.
  - Linkage $\propto \frac{1}{\text{Distance between genes}} \propto \frac{1}{\text{Crossing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

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 → Diagenic, Diandric

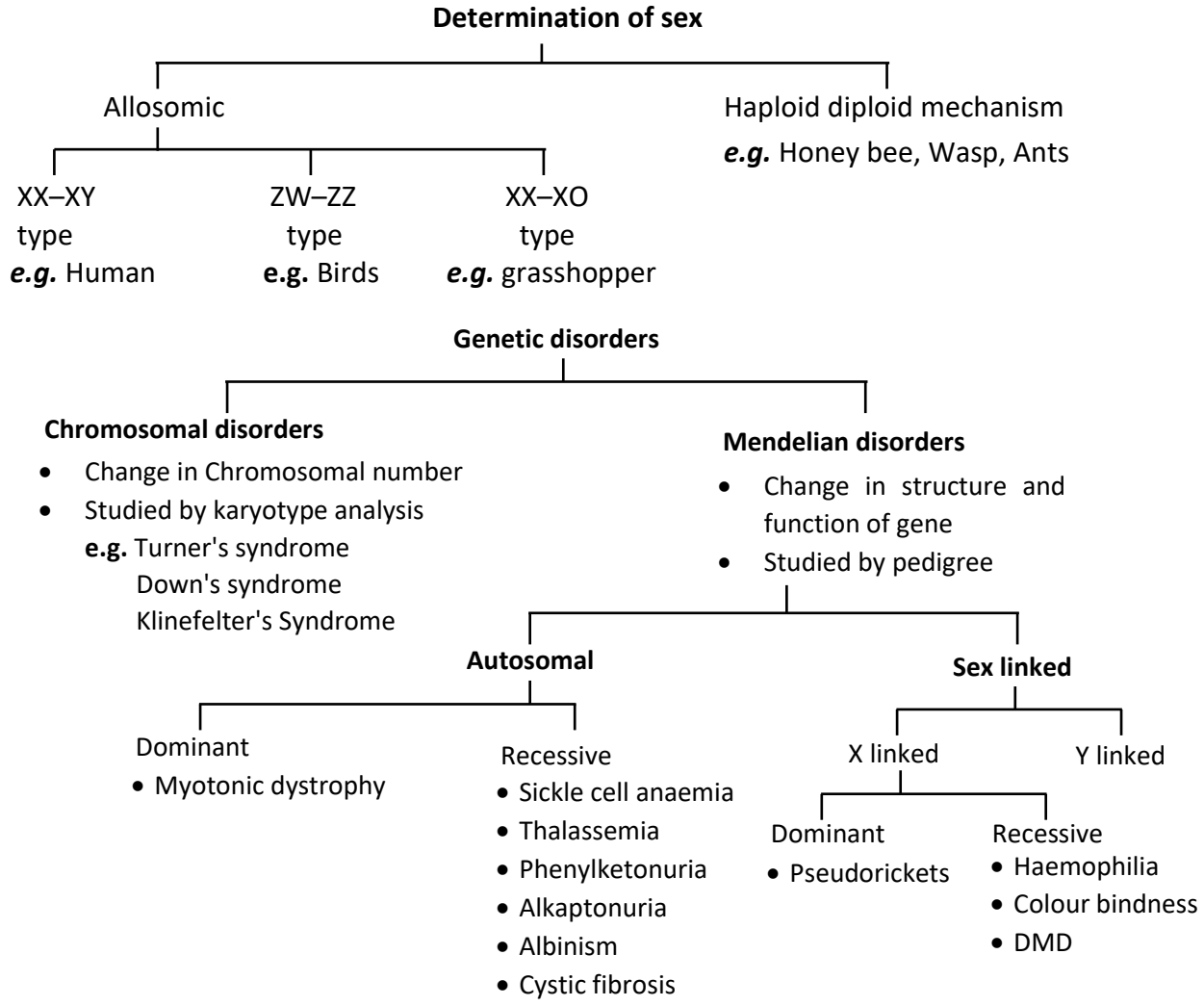
Non criss-cross → Hologenic, Holandric

- **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

- **Mechanism of sex determination** → On the basis of fertilization

Progamic	Syngamic	Epigamic
↓	↓	↓
Before fertilization	During fertilization	After fertilization



NCERT SUMMARY

Genetics is a branch of biology which deals with principles of inheritance and its practices. Progeny resembling the parents in morphological and physiological features has attracted the attention of many biologists. Mendel was the first to study this phenomenon systematically. While studying the pattern of inheritance in pea plants of contrasting characters, Mendel proposed the principles of inheritance, which are today referred to as 'Mendel's Laws of Inheritance'. He proposed that the 'factors' (later named as genes) regulating the characters are found in pairs known as alleles. He observed that the expression of the characters in the offspring follow a definite pattern in different-first generations (F_1), second (F_2) and so on. Some characters are dominant over others. The dominant characters are expressed when factors are in heterozygous condition (Law of Dominance). The recessive characters are only expressed in homozygous conditions. The characters never blend in

heterozygous condition. A recessive character that was not expressed in heterozygous condition may be expressed again when it becomes homozygous. Hence, characters segregate while formation of gametes (Law of Segregation).

Not all characters show true dominance. Some characters show incomplete, and some show co-dominance. When Mendel studied the inheritance of two characters together, it was found that the factors independently assort and combine in all permutations and combinations (Law of Independent Assortment). Different combinations of gametes are theoretically represented in a square tabular form known as 'Punnett Square'. The factors (now known as gene) on chromosomes regulating the characters are called the genotype and the physical expression of the characters is called phenotype.

After knowing that the genes are located on the chromosomes, a good correlation was drawn between Mendel's laws : segregation and assortment of chromosomes during meiosis. The Mendel's laws were extended in the form of 'Chromosomal Theory of Inheritance'. Later, it was found that Mendel's law of independent assortment does not hold true for the genes that were located on the same chromosomes. These genes were called as 'linked genes'. Closely located genes assorted together, and distantly located genes, due to recombination, assorted independently. Linkage maps, therefore, corresponded to arrangement of genes on a chromosome.

Many genes were linked to sexes also, and called as sex-linked genes. The two sexes (male and female) were found to have a set of chromosomes which were common, and another set which was different. The chromosomes which were different in two sexes were named as sex chromosomes. The remaining set was named as autosomes. In humans, a normal female has 22 pairs of autosomes and a pair of sex chromosomes (XX). A male has 22 pairs of autosomes and a pair of sex chromosome as XY. In chicken, sex chromosomes in male are ZZ, and in females are ZW.

Mutation is defined as change in the genetic material. A point mutation is a change of a single base pair in DNA. Sickle-cell anaemia is caused due to change of one base in the gene coding for beta-chain of haemoglobin. Inheritable mutations can be studied by generating a pedigree of a family. Some mutations involve changes in whole set of chromosomes (polyploidy) or change in a subset of chromosome number (aneuploidy). This helped in understanding the mutational basis of genetic disorders. Down's syndrome is due to trisomy of chromosome 21, where there is an extra copy of chromosome 21 and consequently the total number of chromosome becomes 47. In Turner's syndrome, one X chromosome is missing and the sex chromosome is as XO, and in Klinefelter's syndrome, the condition is XXY. These can be easily studied by analysis of Karyotypes.