27

Principles of Inheritance and Variation

Through the process of reproduction, all organisms produce offspring like themselves. The transfer of characters from one generation to the next generation is the central idea of this chapter.

Heredity

It is the study of transmission of characters from parents to offspring or from one generation to the next. Thus, the transmission of structural, functional and behavioural characteristics from one generation to another is called heredity.

Basis of Heredity

Mendel (1866) proposed that inheritance is controlled by paired germinal units or **factors**, now called **genes**. These represent small segments of **chromosome.**

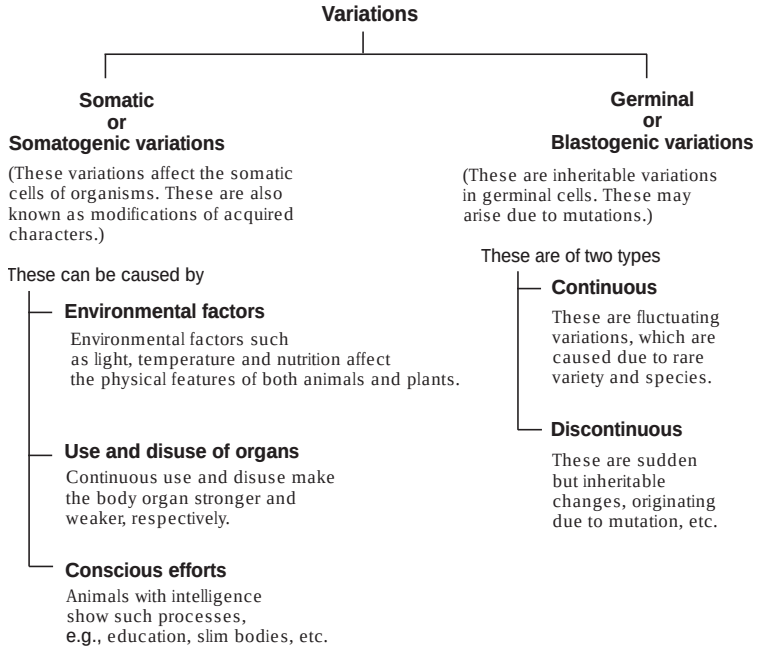
The genetic material present in chromosomes is DNA. Genes are segments of DNA, called **cistrons**. Therefore, DNA is regarded as the chemical basis of heredity.

Inheritance

It is the process by which characters or traits pass from one generation to the next. Inheritance is the basis of heredity.

Variations

It is the difference in characteristics shown by the individuals of a species and also by the offspring or siblings of the same parents.



Terms Related to Genetics

1. **Characters** It is a well-defined morphological or physiological feature of an organism.
2. **Trait** It is the distinguishing feature of a character.
3. **Gene** Inherited factor that determines the biological character of an organism.
4. **Allele** A pair of contrasting characters is called alleles or alternate forms of genes are called alleles.
5. **Dominant allele** The factor or an allele which can express itself in both homozygous and heterozygous state.
6. **Recessive allele** The factor or allele which can express itself only in homozygous state.
7. **Wild allele** The allele which was originally present in the population and is dominant and widespread.

8. **Homozygous condition** The state in which organism has two similar genes or alleles of a particular character, **e.g.,** TT or tt.
9. **Heterozygous condition** In this, the organism contains two different alleles for a particular character, **e.g.,** Tt.
10. **Monohybrid cross** When only one allelic pair is considered in cross breeding.
11. **Dihybrid cross** When two allelic pairs are used in crossing, it is called dihybrid cross.
12. **Genotype** Genetic constitution of an individual is called genotype.
13. **Phenotype** External features of an organism.
14. **Punnet square** It is a checker board which was invented by RC Punnett and used to show the result of a cross between two organisms.
15. **Polyhybrid cross** Involvement of more than two allelic pairs in a cross is called polyhybrid cross.
16. **F_1 or First Filial generation** The second stage of Mendel's experiment is called F_1-generation.
17. **Hybrid vigour or heterosis** The superiority of hybrid over either of its parents in one or more traits.
18. **Gene pool** All the genotypes of all organisms in a population are combinely called gene pool.
19. **Genome** It is the complete set of chromosomes where every gene is present singly as in gamete.
20. **Pureline or pure breeding line** It is a strain of individuals homozygous for all genes considered. The term was coined by Johannsen.
21. **Haploid, diploid and polyploid cell** A single genome is present in haploid, two in diploid and many genomes are present in polyploid cells.
22. **Test cross** The cross of F_1 offsprings with their recessive parents is called test cross.
23. **Back cross** The cross of an organism with the organism of its previous generation is known as back cross.
24. **Reciprocal cross** A cross in which same two parents are used in such a way that, if in one experiment 'A' is used as female parent and 'B' is used as the male parent, in other experiment 'A' will be used as male parent and 'B' is used as female parent.

Gregor Johann Mendel

He was born on July 22, 1822 in Austria. He graduated from Gymnasium in 1840. In 1843, Mendel was admitted to the Augustinian Monastery at Brunn, where he took the name **Gregor**. From 1851-53 he studied mathematics and natural science.

In spring of 1856, he began experimenting with pea plants. In 1866, his paper 'Experiment on Plant Hybridisation' published in volume IV of the proceedings of the natural society. He died on January 6, 1884 and was buried in Brunn central cemetery.

Mendel's experiments involved four steps

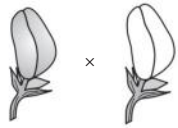
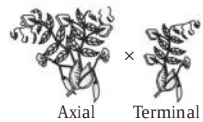
1. **Selection** The selection of characters for hybridisation is the first and an important step.
2. **Hybridisation** The pollination and hybridisation between the individuals of two different /contrasting characteristics.
3. **Selfing** It is the specific hybridisation between the organisms of same origin (siblings).
4. **Calculation** The counting and categorising the products on the basis of character identified takes place in calculation.

Mendel performed his experiments on pea plant and chose seven contrasting characters in it for observation.

These are

(i) Colour of seed
(ii) Shape of seed
(iii) Flower colour
(iv) Colour of pod
(v) Shape of pod
(vi) Position of flower
(vii) Height of plant

These characters and their inheritance patterns are given in the following table

Character or Trait Studied	Parent forms Crossed (F_1 Cross)	F_1 Phenotype	F_2 Products Dominant form, Recessive form	Total	Actual Ratio	Chromosome Location
Colour of seed	 Yellow (cotyledon) × Green	All yellow	6022 yellow, 2001 green	8023	3.01 : 1	1
Shape of seed	 Round × Wrinkled	All round	5474 violet, 1850 wrinkled	7324	2.96 : 1	7
Flower colour	 Violet × White	All violet	705 violet, 224 white	929	3.15 : 1	1
Colour of pod	 Green ×  Yellow	All green	428 green, 152 yellow	580	2.82 : 1	5
Shape of pod	 Inflated ×  Constricted	All inflated	882 inflated, 299 constricted	1181	2.95 : 1	4
Position of flower	 Axial × Terminal	All axial	651 axial, 207 terminal	858	3.14 : 1	4
Height of plant	 Tall Dwarf	All tall	787 tall, 277 dwarf	1064	2.84 : 1	4

Emasculation and Bagging

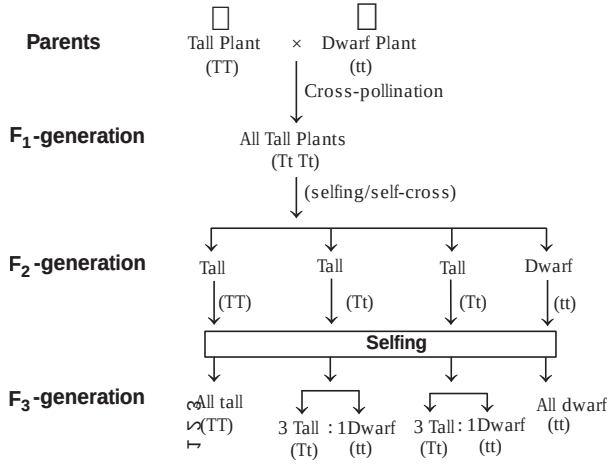
Mendel required both self and cross-fertilisation within the plants for his experiments. Due to its self-fertilising nature, the anthers of pea plants require removal before maturity (emasculation) and the stigma is protected against any foreign pollen (bagging). Through the process of emasculation and bagging, the pollen of only selected parent can be used for cross-fertilisation.

Inheritance of One Gene/Monohybrid Cross

Mendel performed several experiments on pea by considering one character at a time.

It is a cross made to study simultaneous inheritance of a single pair of Mendelian factors.

The schematic presentation of the monohybrid cross is as follows



Monohybrid cross in pea plant

Mendel's Laws of Inheritance

From the three laws of inheritance (**i.e.,** Law of dominance, Law of segregation and Law of independent assortment), the first two laws are based on the monohybrid cross.

These are explained in detail below

1. Law of Dominance

According to this law, 'when a cross is made between two homozygous (pure line) individuals considering contrasting trait of simple character then the trait that appears in F_1 hybrids is called dominant and the other one that remains masked is called recessive trait'.

In pea plant, out of the 7 characters, Mendel studied the dominant and recessive traits. These characters are discussed earlier.

The dominant and recessive traits are also found in other animals, **e.g.,**

Cat	(a) Skin colour	Tabby colour is dominant over black or blue.
	(b) Length of hair	Short hair are dominant over long hair (Angora).
Cattle	(a) Colour of face	White face colour is dominant over coloured face.
	(b) Horn	Polled or hornless are dominant over horned cattle.
Dog	(a) Skin colour	Grey colour is dominant over black colour.
	(b) Tail	Stumpy tail is dominant over normal tail.
Drosophila	(a) Eye colour	Red colour is dominant over white.
	(b) Wings	Flat and yellow wings are dominant over curled and white.
	(c) Body colour	Grey body colour is dominant over white.
Salamander	Body colour	Dark body colour is dominant over light.

The law of dominance explains why individuals of F_1-generation express the trait of only one parent and the reason for occurrence of 3:1 ratio in F_2 individuals.

Exceptions to Law of Dominance

These are as follows

(i) Incomplete Dominance/Blending Inheritance (CORRENS, 1903)

It is also known as **Intermediate** or **Partial** or **Mosaic** inheritance.

When F_1 hybrids exhibit a mixture or blending of characters of two parents, it is termed as blending inheritance.

It simply means that the two genes of allelomorphic pair are not related as dominant or recessive, but each of them expresses themselves partially, **e.g., 4 O'clock plant** (**Mirabilis jalapa**), **snapdragon** (**Antirrhinum**) and **homozygous fowl**. In 4 O'clock plant when a cross is made between dominant (red) and recessive

(white) variety, the result of F_2-generation shows deviation from Mendel's predictions.

Here, both phenotypic and genotypic ratios came as 1 : 2 : 1 for Red : Pink : White.

(II) Codominance

The phenomenon of expression of both the alleles in a heterozygote is called **codominance**.

The alleles which do not show dominant-recessive relationship and are able to express themselves independently when present together are called **codominant allele,** e.g., **coat colour in short horned cattles and MN blood group in humans.**

In short horned cattle, when a cross is made between white (dominant) and red (recessive) variety, appearence of all Roan offsprings in F_1-generation and then white, roan and red in 1:2:1 ratio in F_2-generation show codominance of both the colours in roan.

The roan coloured F_2 individuals in above cross have both red and white hairs in the form of patches but no hair is having the intermediate colour.

(III) Pleiotropic Gene

The ability of a gene to have multiple phenotypic effects, because it influences a number of characters simultaneously, is known as pleiotropy and such genes are called pleiotropic genes.

It is not essential that all traits are equally influenced, sometimes it is more evident in case of one trait (major effect) and less evident in other (minor effect), **e.g.,** in garden pea, the gene controlling flower colour, also controls the colour of seed coat and the presence of red spot on leaf axil.

2. Law of Segregation/Law of Purity of Gametes

According to this law, 'In F_1 hybrid, the dominant and recessive characters though remain together for a long time, but do not contaminate or mix with each other and separate or segregate at the time of gamete formation. Thus, the gamete formed receives either dominant or recessive character out of them.'

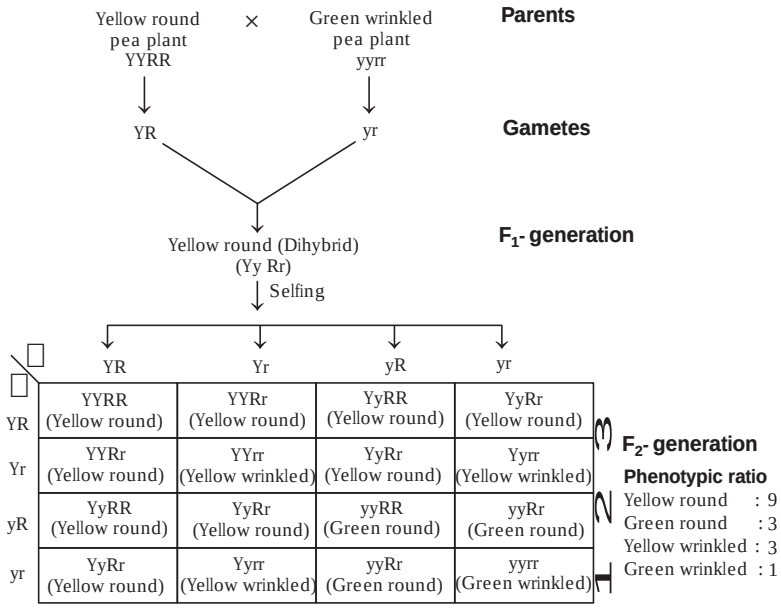
For proper understanding of Mendel's law of segregation, the formation of hybrid is considered from pureline homozygous parents through monohybird cross given before first law.

As the purity of gametes again established in F_2-generation, it is called law of purity of gametes.

Inheritance of Two Genes/Dihybrid Cross

These crosses are made to study the inheritance of two pairs of Mendelian factors or genes.

The schematic representation of the dihybrid cross is as follows



Dihybrid cross in pea plant

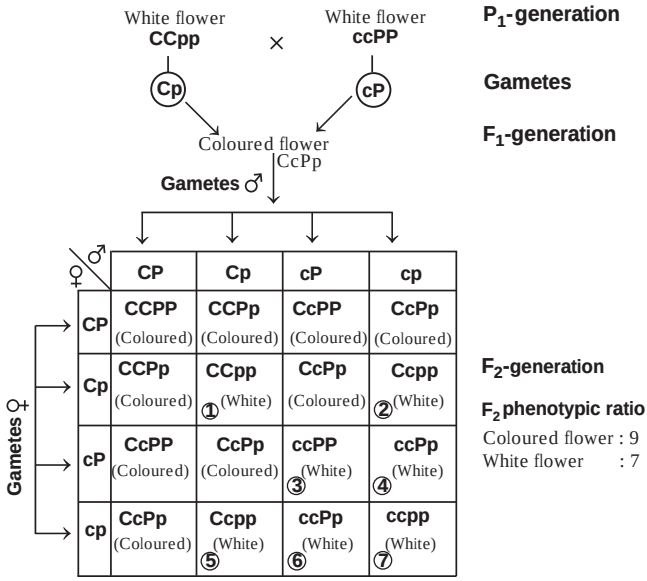
Exceptions to Law of Segregation

These are as follows

(I) Complementary Genes

The two pairs of non-allelic dominant genes, which **interact to produce only one phenotypic trait**, but neither of them (if present alone) produces the trait in the absence of other. It shows the phenotypic ratio of 9 : 7.

This cross is shown as



The results of an experiment to show the operation of complementry genes in the production of flower colour in sweet pea (Lathyrus)

(II) Epistatic Gene or Inhibitory Gene

It is the interaction between two non-allelic genes, in which one gene masks or suppresses the expression of other. The gene which got suppressed is called hypostatic factor and the suppressor gene is called epistatic factor. Such an interaction is called epistasis.

The epistasis may be

(A) Dominant Epistasis

In this, out of two pairs of genes, the dominant one masks the expression of other gene pair.

The ratio obtained in this may be 12 : 3 : 1 or 13 : 3, **e.g.,** coat colour gene in dog.

(B) Recessive Epistasis

In this, out of the two pairs of genes, the recessive epistatic gene masks the activity of dominant gene of the other gene locus. The ratio obtained in this may be 9 : 3 : 4, **e.g.,** coat colour gene in mice.

3. Law of Independent Assortment

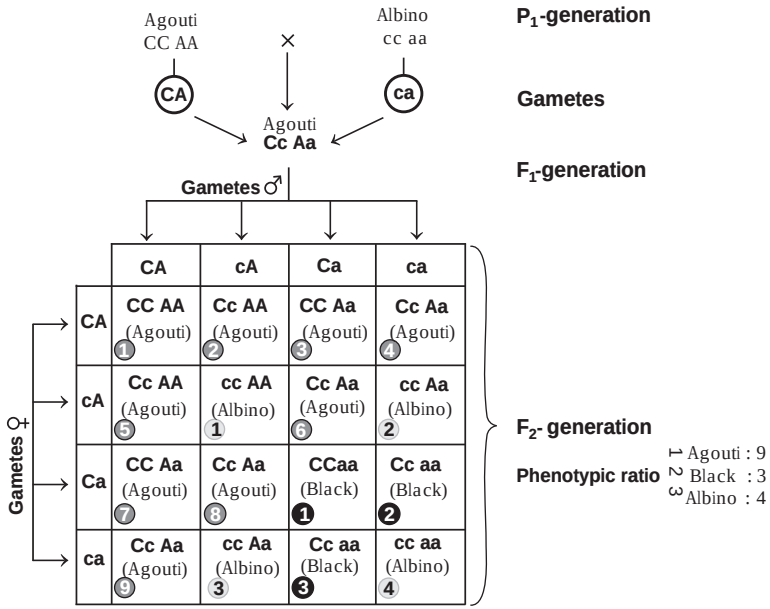
This law states that, 'the inheritance of one character is always independent to the inheritance of other character within the same individual'. The dihybrid cross of Mendel can be a very good example of independent assortment.

Exceptions to Law of Independent Assortment

These are as follows

(I) Supplementary Genes

Two independent dominant gene pairs, which interact in such a way that one dominant gene produces its effect irrespective of the presence or absence of other, **e.g.,** the coat colour in mice. **The cross is represented as**



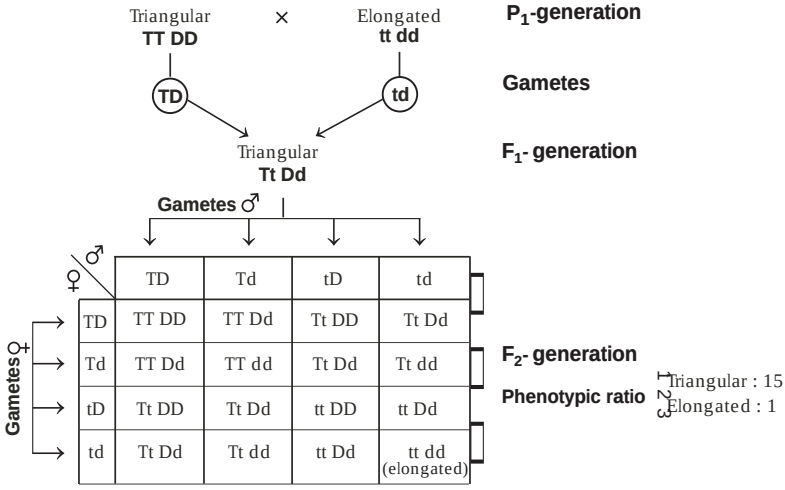
Interaction of supplementary genes in mice for coat colour

Here, the presence of gene C produces black colour which along with gene A changes its expression in agouti colour. Thus in all, combinations with at least one C and one A produce agouti colour.

(II) Duplicate Gene

The two pairs of genes which determine same or nearly same phenotype, hence either of them is able to produce the character. The duplicate genes are also called **pseudoalleles**, **e.g.,** fruit shape in Shepherd's purse.

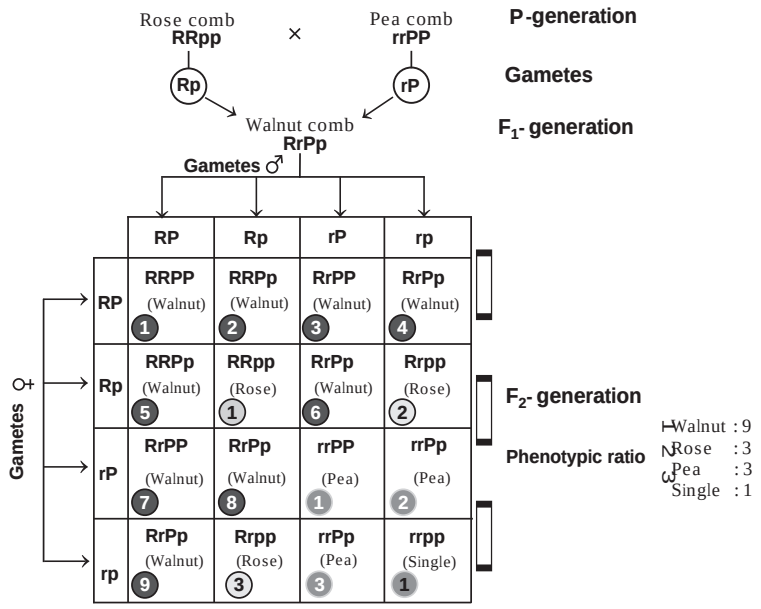
The inheritance can be seen as



Interaction of duplicating genes in Shepherd's purse for seed pod's shape

(III) Collaborator Gene

In this, the two gene pairs which are present on separate locus, interact to produce totally new trait or phenotype, **e.g.,** inheritance of comb in poultry.



Inheritance of rose and pea comb in poultry

Multiple Allelism

It is the presence of more than two alleles for a gene, **e.g.,** ABO blood group in human beings is controlled by three alleles, but only two of these are present in an individual.

Polygenic Inheritance

Genes when acting individually have a small effect but that collectively produce a significant phenotypic expression are called **polygenes**, **e.g.**, genes for height or weight. The polygenes show polygenic inheritance.

Chromosomal Theory of Inheritance

Walter Sutton and **Theodore Boveri** in 1902 united the knowledge of chromosomal segregation with Mendelian principles and called it chromosomal theory of inheritance.

According to this theory,

(i) All hereditary characters are carried with sperms and egg cells, as they provide bridge from one generation to the other.

(ii) The hereditary factors are carried in the nucleus.

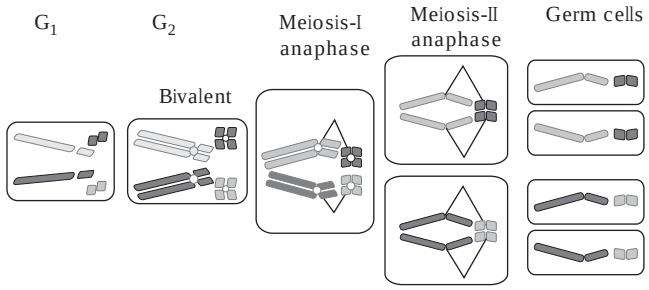
(iii) Chromosomes are also found in pairs like the Mendelian alleles.

(iv) The two alleles of a gene pair are located on homologous sites on the homologous chromosomes.

(v) The sperm and egg have haploid sets of chromosomes, which fuse to re-establish the diploid state.

(vi) The genes are carried on the chromosomes.

(vii) Homologous chromosomes synapse during meiosis and get separated to pass into different cells. This is the basis for segregation and independent assortment.



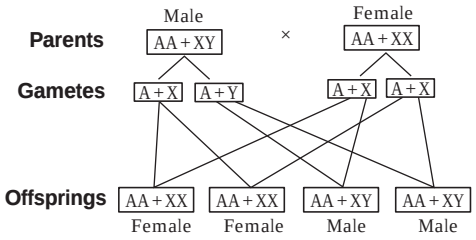
Meiosis and germ cell formation in a cell with four chromosomes

Sex-Determination

It is the method by which the distinction between male and female is established in a species. It is usually under genetic control of specific chromosomes called **sex chromosomes** or **allosomes**.

There are five main genetic mechanisms of sex-determination

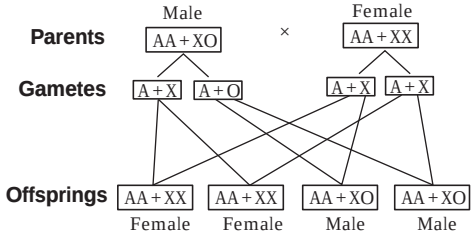
(i) XX-XY Method



Examples are mammals (as in humans).

(ii) XX-XO Method

In this, female has XX chromosomes and produces homogametic eggs, while male has only one chromosome and produces two types of sperms, **e.g., gynosperms** (with X) and **androsperms** (without X), **e.g.,** insects and roundworms.



(iii) ZW-ZZ Method

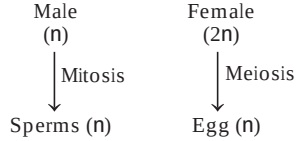
In this, the male is homogametic and female is heterogametic, **e.g.,** certain insects, fishes, reptiles and birds.

(iv) ZO-ZZ Method

In this, female is heterogametic while the male is homogametic, **e.g.,** moths and certain butterflies.

(v) Haploid-Diploid Method

In this method, the unfertilised egg develops into male (Arrhenotoky) while fertilised egg develops into female. This type of sex-determination is the characteristic feature of insects like honeybees, ants, etc.



Sex-Determination in Humans

The human shows XY type of sex-determination. Out of total (23 pairs) chromosomes, 22 pairs are exactly similar in both males and females, known as **autosomes**.

The female contains a pair of X-chromosome and male contains both X and Y-chromosomes. The sex is determined by the genetic make up of sperm.

During spermatogenesis among males, two types of gametes are produced, 50% of the total sperms carry X-chromosomes and the rest 50% carry Y-chromosomes.

Linkage (Exception to Independent Assortment)

It is the phenomenon of certain genes staying together during inheritance through generations without any change or separation. In other words, 'It is the tendency of genes staying together during inheritance.'

Morgan (1910) clearly proved and defined linkage on the basis of his breeding experiments on fruitfly, **Drosophila melanogaster**.

Linked genes are inherited together with the other genes as they are located on the same chromosome.

Linkage group are equal to the number of chromosomes pair present in cells, **e.g.,** humans have 23 linkage groups.

According to Morgan et. al., the linkage can be

(i) **Complete or Perfect** In this, genes remain together for at least two generations.

(ii) **Incomplete or Imperfect** In this, genes remain together within the same chromosome for less than two generations.

Sex-Linked Inheritance

Sex-linked characters are governed by the genes located on sex chromosomes. The phenomenon of the inheritance of such characters is known as sex-linked inheritance, **e.g.,** haemophilia, colour blindness, etc.

The sex-linked genes located on X-chromosomes are called X-linked genes, while these present on Y-chromosomes are called holandric genes.

Few examples of sex-linked inheritance in human beings are given below

(i) **Haemophilia** It is a sex-linked recessive disease. It is transmitted from an unaffected carrier female to some of the male progeny. In this disease, a protein involved in the clotting of blood is affected due to which a small cut results in profuse bleeding and sometimes may lead to death.

A heterozygous female (carrier) for haemophilia may transmit the disease to sons (50% chances), if she marries a normal male. The possibility of female becoming haemophilic is extremely rare because mother of such a female has to be at least carrier and the father should be haemophilic.

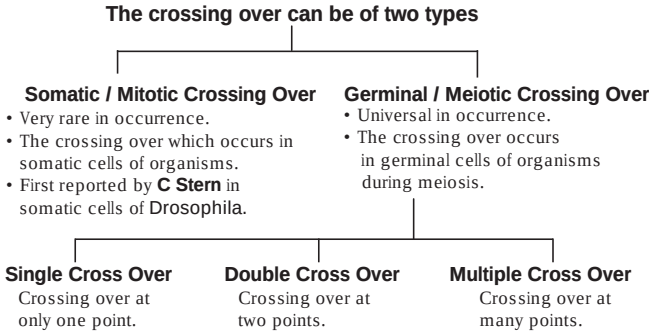
(ii) **Colour blindness** It is also a sex-linked recessive disorder. It is due to defect in either red or green cone of eye resulting in failure to discriminate between red and green colour. The defect occurs due to mutation in certain genes present in the X-chromosomes. The son of a woman who is carrier for the disease has 50 per cent chance of being colourblind.

The carrier mother is not colourblind herself because the gene is recessive. The daughter will be colourblind only if the mother is at least carrier and father is colourblind.

(iii) Duchenne Muscular Dystrophy (DMD) is also a sex-linked.

Crossing Over/Recombination

Those genes which show non-linkage, result into non-parental combinations in F_1-generation. Presence of such combinations indicates that in these genes, the process of interchange of alleles within non-sister chromatids of homologous chromosomes takes place, this is known as crossing over.



The mechanism of crossing over is explained by various theories, some of them with their propounders are listed below

1. **Copy choice theory** — J Lederberg (1955)
2. **Precocity theory** — C D Darlington (1931)
3. **Belling hypothesis** — Belling
4. **Break and exchange theory** — Stern and Hotta (1969)
5. **Hybrid DNA Model** — R Holliday (1964)

Linkage Maps/Genetic Maps/Chromosomal Maps

'It is the graphic representation of the relative distance between the genes in a linkage group'.

The first linkage map was given by **Sturtevant** and **Morgan** in 1920s. In linkage maps, the intergenic distances can be explained through arbitory unit of measurement called, **map unit** to describe the distance between linked genes.

1 map unit =1% of crossing over

One map unit is now referred as cM (centiMorgan) in the honour of Morgan's contribution.

Steps to Construct Genetic Map

Step 1 Determination of linkage group and total number of genes

By hybridising wild and mutant strains, we can determine the total number of genes and link groups in an organism.

Step 2 Determination of map distance

For determining map distances, the test crosses are performed. The relative distance can be calculated according to the percentage of crossing over, as cross over frequency is directly proportional to the distance between the genes.

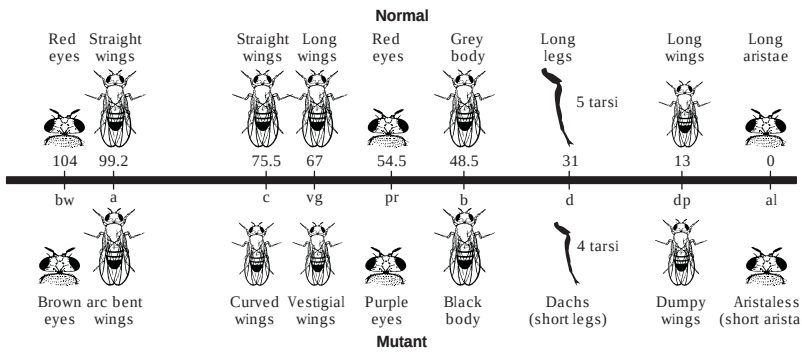
Step 3 Determination of gene order

After determining the relative distance, the genes can be placed in proper linear order.

Step 4 Combining map segments

Finally different segments forming linkage group of a chromosome, are combined to form genetic map.

Thus, **chromosomal map of chromosome number** 2 **of Drosophila melanogaster can be seen as**



The genetic map of chromosome number-2 of Drosophila melanogaster

Cytoplasmic Inheritance/Extranuclear Inheritance

The total self-replicating hereditary material of cytoplasm is called **plasmon** and cytoplasmic units of inheritance are described as plasma genes.

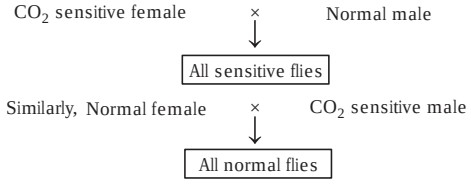
Cytoplasmic inheritance have two distinct features

(i) It is maternal inheritance, **i.e.,** only maternal parent contributes for inheritance.

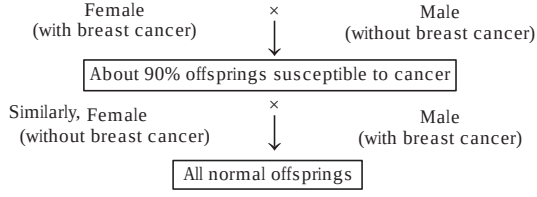
(ii) The reciprocal crosses are not same due to the participation of female parents only, **e.g.,** sigma particle inheritance in **Drosophila**, Kappa particle inheritance in **Paramecium** and breast tumor in mice, etc.

In **Drosophila,** one strain shows more sensitivity towards CO_2 (these are comparatively easily immobilised by exposing them to CO_2). This more sensitivity was discovered by L Heritier and Teissier. The sensitive trait is regulated by a heat labile substance present in cytoplasm called sigma.

The inheritance of sensitive fly can be seen as



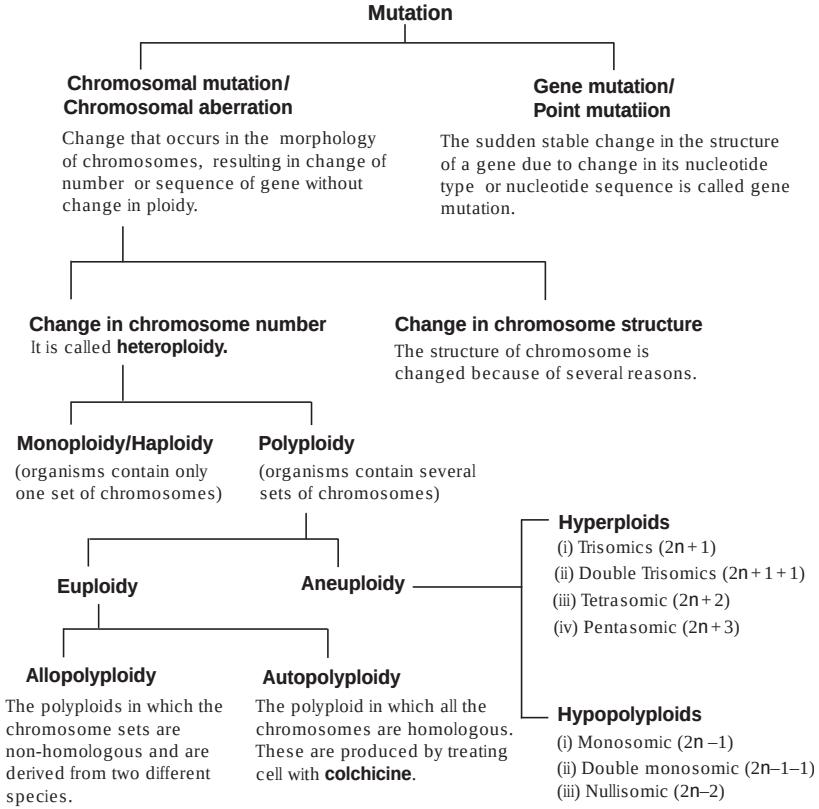
Results of reciprocal crosses clearly indicate the inheritance of more CO_2 sensitivity through females. The mammary cancer or breast tumour in mice has been found to be maternally transmitted. It was noted by **JJ Bitiner**. **He performed following crosses regarding cancer in mice**



Such a difference in reciprocal crosses suggests the presence of maternal inheritance.

Mutation (Hugo de Vries; 1901)

A sudden inheritable discontinuous variation which appears in an organism due to permanent change in their genotypes.



Change in Chromosomal Structure

The variations occur due to following four processes

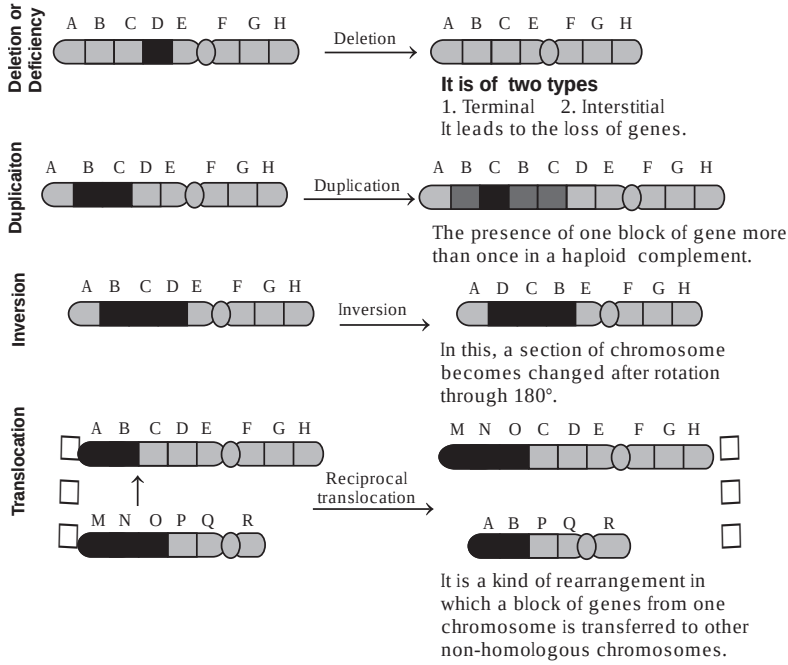
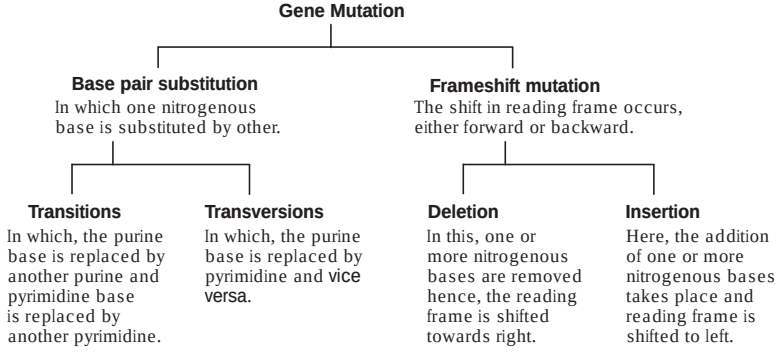


Diagram showing the forms of chromosomal mutations

Gene Mutation

The intragenic or point mutations involve alterations in the structure of gene by altering the structure of DNA. **It is of two types**



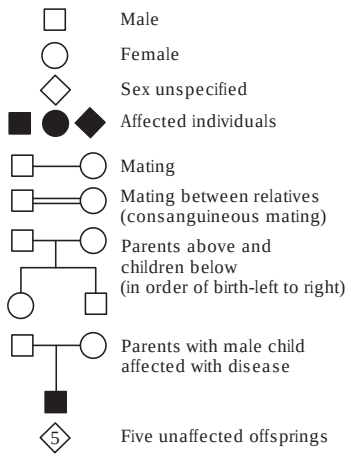
All these mutations cause various genetic disorders. **A list of some important genetic disorders is given below.**

Disorder	Dominant/ Recessive	Autosomal/ Sex linked	Symptom	Effect
Sickle-cell anaemia	Recessive	Autosomal, gene on chromosome 11	Aggregation of erythrocytes, more rapid destruction of erythrocytes leading to anaemia.	Abnormal haemoglobin in RBCs.
Phenylketonuria	Recessive	Autosomal, gene on chromosome 12	Failure of brain to develop in infancy, mental retardation, idiots	Defective form of enzyme phenylalanine hydroxylase.
Cystic Fibrosis (CF)	Recessive	Autosomal, gene on chromosome 7	Excessive thick mucus, clogging in lungs, liver and pancreas anomalies.	Failure of chloride ion transport mechanism through cell membrane.
Huntington's Disease (HD)	Dominant	Autosomal, gene on chromosome 4	Gradual degeneration of brain tissues in middle age, loss of motor control.	Production of an inhibitor of brain cell metabolism.
Haemophilia A/B	Recessive	Sex-linked, gene on X-chromosome	Failure of blood to clot.	Defective form of blood clotting factor VIII/IX
Colour blindness	Recessive	Sex-linked, gene on X-chromosome	Failure to discriminate between red and green colour.	Defect in either red or/ and green cone cells of retina.
Down's syndrome		Autosomal, aneuploidy (trisomy + 21)	Mongolian eyefold (epicanthus), open mouth, protruded tongue, projected lower lip, many loops on finger tips, palm crease	Retarded mental development, IQ below 40.

Disorder	Dominant/ Recessive	Autosomal/ Sex linked	Symptom	Effect
Turner's syndrome		Sex chromosome monosomy 44+X0	Short stature females (<5'), webbed neck, body hair absent, menstrual cycle absent, sparse pubic hair, under developed breasts, narrow lips, puffy fingers.	Sterile, hearing problem
Klinefelter's syndrome		Sex chromosomal aneuploidy (Tri/tetrasomy of X chromosome), **i.e.**, 44+ XXY, 44+XXXY	These males are tall with long legs, testes small, sparse body hair, Barr body present, breast enlargement.	Gynaecomastia, azoospermia, sterile

Pedigree Analysis

Scientists have devised another approach, called pedigree analysis, to study the inheritance of genes in humans. This is also useful while studying the population when progeny data from several generations is limited. It is also useful in studying the species with long generation time. A series of symbols is used to represent different aspects of a pedigree. **These are as follows**



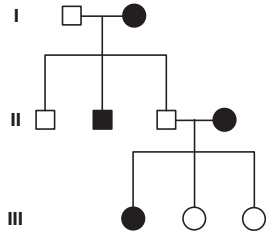
Symbols used in the human pedigree analysis

Once phenotypic data is collected from several generations and the pedigree is drawn, careful analysis will allow you to determine whether the trait is dominant or recessive.

For those traits exhibiting dominant gene action

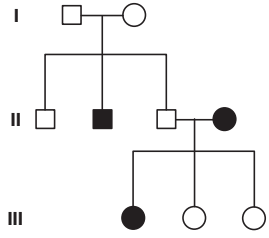
- Affected individuals have at least one affected parent.
- The phenotype generally appears in every generation.
- Two unaffected parents only have unaffected offspring.

It is called dominant pedigree and shown as



Those traits which exhibit recessive gene action

- Unaffected parents can have affected offspring.
- Affected progeny are both male and female and it is called **recessive pedigree** and shown as



In due course of time, the genetics and its principles will help in the solution of several heredity problems.