- *In heredity, the genes are obtained from father and mother.*
- *Nucleus and chromosomes are stained by* ***hematoxylin****.*
- ***Johannsen*** *coined the term* ***genotype, phenotype, pure line****.*
- *The term* ***hybrid vigour (heterosis)*** *given by* ***Shull.***
- *In mitosis, the daughter cells resemble each other and also the parent cell, in meiosis they differ not only from parent cell in having half the number of chromosomes, but also differ among themselves qualitatively in genetic constitution due to crossing over, independent assortment and segregation.*
- *Mendelian genetics is also called as* ***forward genetics****.*
- *Mendel either avoided the result or could not conduct independent assortment between pod form and stem length.*
- *Mendel also observed that flower colour and colour of the seed coat may not assort independently.*
- *Mendel failed to produce same results of his experiments of pea in Hawkweed (Hieracium) and Beans.*
- *Mendel's typical monohybrid phenotypic ratio was 3 : 1 which was in reality a hidden 1 : 2 : 1 ratio of genotypes.*
- *Mendelism gave well-defined principles even in early stage in compare to* ***Darwinism****.*
- *Mendel did not recognize the linkage phenomenon in his experiments because characters, he studied were located on different chromosomes.*
- *Mendelian factor are separated during* ***Anaphase- I in Meiosis- I****.*
- *If Mendel had studied the seven traits using a plant with 12 chromosomes instead of 14, He would not have discovered the law of independent assortment.*
- ***Law of filial regression*** *was postulated by Galton.*
- *Mendel didn't imagine of linkage.*
- *Mendel in his experiments on pea considered quantity in relation to quality.*
- *In bound seeds (RR/Rr) starch branching enzyme (SBE -1) is found but it is absent in wrinkled seeds or in rr seeds.*
- *It was thought previously that seven traits in pea studied by Mendel were located on seven different chromosomes but recent studies proves that these are on four chromosomes.*
- *The genes for seed form in pea was present on chromosomes no. 7.*
- *Independent assortment is shown by the allels present on different loci.*

Interaction of genes.

Genes interaction is the influence of alleles and non-alleles on the normal phenotypic exprssion of genes. It is of two types.

(1) **Inter–allelic or intra–genic gene interaction :** In this case two alleles (located on the same gene locus on two homologous chromosomes) of gene interact in such a fashion to produces phenotypic expression *e.g.* co-dominance, multiple alleles.

(i) **Incomplete dominance (1:2:1 ratio) :** After Mendel, several cases were recorded where F_1 hybrids were not related to either of the parents but exhibited a blending of characters of two parents. This is called incomplete dominance or blending inheritance.

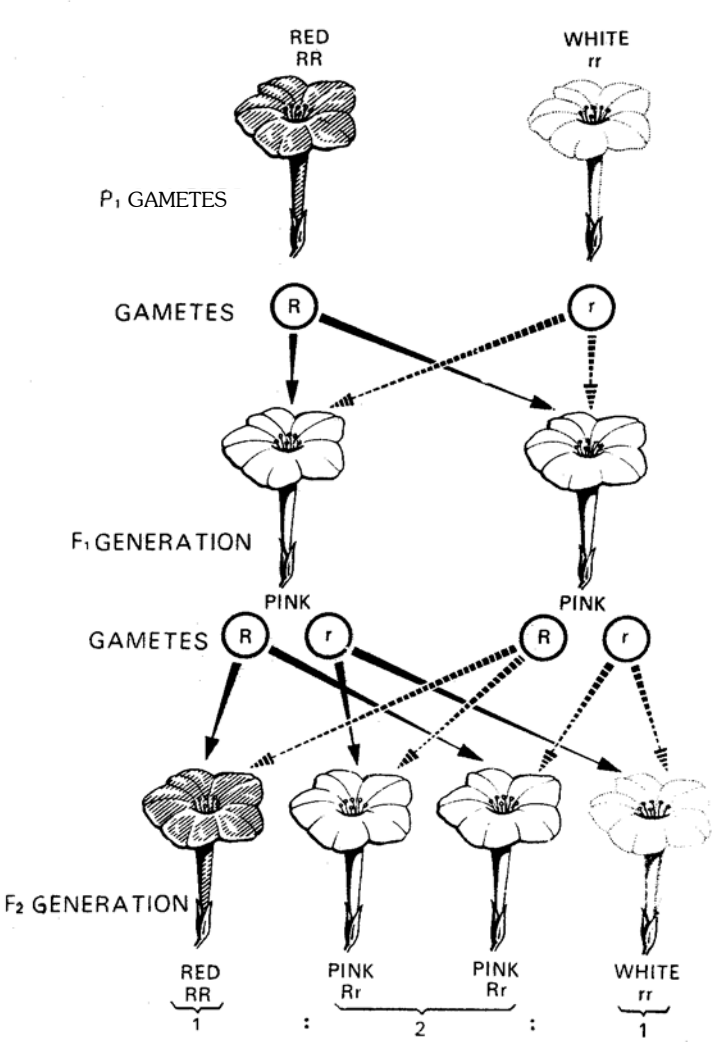

Fig : Incomplete dominance of flower colour in Mirabillis jalapa

Example : In 4-O'clock plant, (*Mirabilis jalapa*), when plants with red flowers **(RR)** are crossed with plants having white flowers **(rr)** the hybrid F_1 plants **(Rr)** bear pink flowers. When these F_1 plants with pink flowers are self pollinated they develop red **(RR),** pink **(Rr)** and white **(rr)** flowered plants in the ratio of 1:2:1 (F_2 generation).

Example : In Snapdragon or dog flower (*Antirrhinum majus*) the dominant character of leaf (Broadness) and flower (Red) shows incomplete dominance over recessive characters (Narrowness and white) in dihybrid cross.

(ii) **Codominance (1:2:1 ratio) :** In codominance, both the genes of an allelomorphic pair express themselves equally in **F_1** hybrids. 1:2:1 ratio both genotypically as well as phenotypically in **F_2** generation.

Example : Codominance of coat colour in cattle.

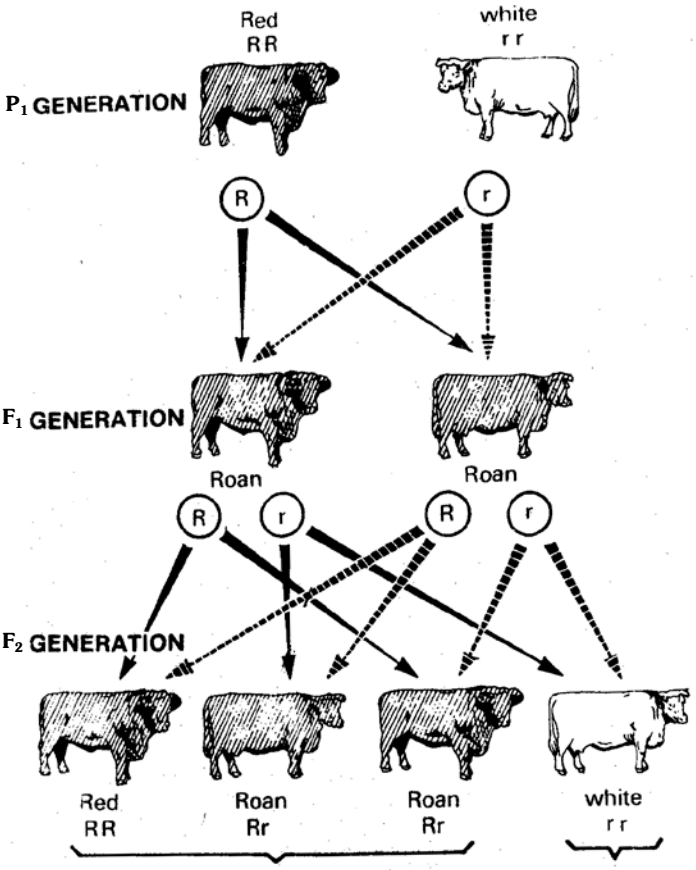

Fig : Inheritance of coat colour in cattle

In cattle gene **R** stands for red coat colour and gene **r** stands for white coat colour. When red cattle **(RR)** are crossed with white cattle **(rr),** the **F_1** hybrids have roan coloured skin (not the intermediate pink). The roan colour is actually expressed by a mixture of red and white hairs, which develop side by side in the heterozygous **F_1** hybrid. In **F_2** generation red, roan and white appear in the ratio of 1 : 2 : 1. The phenotypic ratio equal to genotypic ratio **RR, Rr, rr** (1 : 2 : 1).

Example : Codominance in andalusian fowl

In andalusian fowl a cross between pure black and pure white varieties results in blue hybrids.

Example : Codominance of blood alleles in man

(a) **MN** blood type in man is an example of codominance. The persons with **MN** genotype produce both antigen **M** and **N** and not some intermediate product indicating that both the genes are functional at the same time.

(b) In **ABO** blood group system gene **A** and **B** responsible for blood group **A** and **B** are codominant. The hybrid has **AB** blood group.

Differences between incomplete dominance and codominance

Incomplete dominance	Codominance
Effect of one of the two alleles is more conspicuous.	The effect of both the alleles is equally conspicuous.
It produces a fine mixture of the expression of two alleles.	There is no mixing of the effect of the two alleles.
The effect in hybrid is intermediate of the expression of the two alleles.	Both the alleles produce their effect independently, e.g., I^A and I^B, Hb^S and Hb^A.

(2) **Non–allelic or inter-genic gene interaction :** Here two or more independent genes present on same or different chromosomes, interact to produce a new expression *e.g.* epistasis, complementary genes, supplementary genes, duplicate genes, inhibitory genes, lethal genes etc.

(i) **Complementary genes (9 : 7 ratio) :** The complementary genes are two pairs of nonallelic dominant genes (*i.e.* present on separate gene loci), which interact to produce only one phenotypic trait, but neither of them if present alone produces the phenotypic trait in the absence of other.

Example : Complementary genes for flower colour in sweet pea. In sweet pea (*Lathyrus odoratus*) the purple colour of flowers is dependent on two nonallelic complementary genes ***C*** and ***P***. Gene ***C*** produces an enzyme that catalyzes the formation of colourless chromogen for the formation of anthocyanin pigment. Gene P controls the production of an enzyme, which catalyzes the transformation of this chromogen into anthocyanin. These genes are complementary to each other. It means the pigment anthocyanin is produced by two-biochemical reactions and the end product of first reaction forms the substrate for the other.

P_1 Generation White Flower CC pp × White Flower cc PP

Gametes Cp, cP

F_1 Generation Coloured Flower Cc Pp

Gametes ♂ : CP, Cp, cP, cp

Gametes ♀ : CP, Cp, cP, cp

♀ \ ♂	CP	Cp	cP	cp
CP	CC PP coloured	CC Pp coloured	Cc PP coloured	Cc Pp coloured
Cp	CC Pp coloured	CC pp white 1	Cc Pp coloured	Cc pp white 2
cP	Cc PP coloured	Cc Pp coloured	cc PP white 3	cc Pp white 4
cp	Cc PP coloured	Cc pp white 5	cc Pp white 6	cc pp white 7

F_2 Generation = Coloured Flower : 9, White Flower : 7

Fig : The results of an experiment to show the operation of complimentary genes in the production of flower colour in sweet pea (*Lathyrus*)

If a plant possesses dominant gene ***C*** and ***P*,** it produces purple flowers. But if a plant has a genotype **CCpp,** it produces the raw material but is unable to convert it into anthocyanin. Therefore, it produces white flowers. Similarly, if it possesses dominant gene **P,** but no dominant **C (ccPP),** it produces white flowers because gene **P** can convert colourless chromogen into anthocyanin but cannot form chromogen.

(ii) **Supplementary genes (9 : 3 : 4 ratio) :** Supplementary genes are two independent pairs of dominant genes. Which interact in such a way that one dominant gene will produce its effect whether the other is present or not. The second dominant when added changes the expression of the first one but only in the presence of first one. In rats and guinea pigs coat colour is governed by two dominant genes **A** and **C,** the agouti-coloured guinea pigs have genotype **CCAA**. The black mice possess factor for black colour **C** but not the gene **A** for agouti colour. If gene for black colour is absent agouti is unable to express itself and mice with a genotype **ccAA** are albino. Here presence of gene **C** produced black colour and addition of gene **A** change its expression to agouti colour.

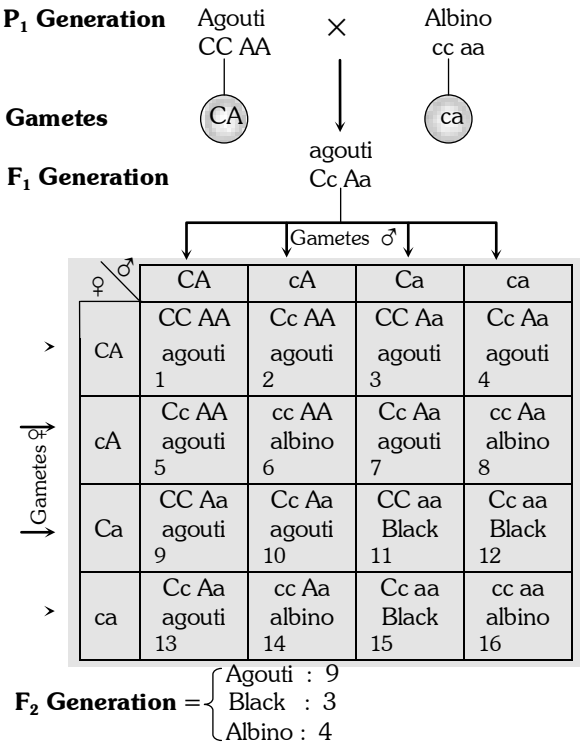

Fig : Interaction of supplementary genes in mice for coat colour

(iii) **Epistasis (Inhibiting genes) :** Epistasis is the interaction between nonallelic genes (Present on separate loci) in which one-gene masks, inhibits or suppresses the expression of other gene. The gene that suppresses the other gene is known as inhibiting or epistatic factor and the one, which is prevented from exhibiting itself, is known as hypostatic. Although, it is similar to dominance and recessiveness but the two factors occupy two different loci. Therefore, while dominance involved intragenic or interallelic gene suppression, the epistasis involves intergenic suppression. Epistasis can be of the following types – dominant epistasis, recessive epistasis.

(a) **Dominant epistasis (12:3:1 or 13:3 ratio) :** In dominant epistasis out of two pairs of genes the dominant allele, (*i.e.,* gene **A**) of one gene masks the activity of other allelic pair **(Bb)**. Since the dominant epistatic gene **A** exerts its epistatic influence by suppressing the expression of gene **B** or **b**, it is known as dominant epistasis.

Example – Dominant epistasis in dogs : In dogs white coat colour appears to be dominant. It develops due to the action of epistatic gene **I** which prevents the formation of pigment, controlled by hypostatic gene **B**.

The hypostatic gene **B** produces black coat while its hypostatic allele **b** produces brown coat colour only when gene **I** is recessive. The progeny of dominant gene **I** does not allow them to function and results in white colour.

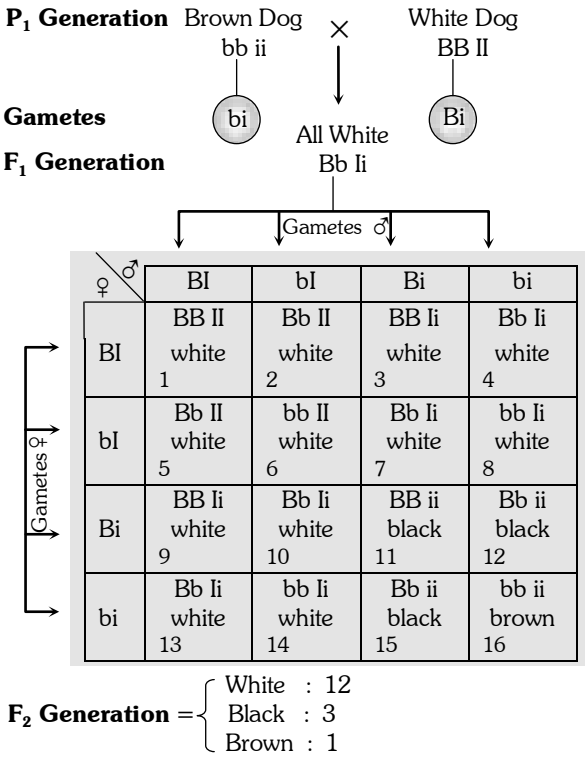

Fig : Interaction of inhibiting genes in dog for coat colour showing dominant epistasis

When two white coat dogs are crossed, they produce white, black and brown in the ratio of 12 : 3 : 1 The white dogs in this case possess gene for black or brown colour but does not produce the pigment because of the presence

of gene **I** in dominant state. Similar phenomena have been seen in *fruit colour in cucurbita as summer squash and coat colour in chickens*.

(b) **Recessive epistasis (9:3:4 ratio) :** Epistasis due to recessive gene is known as recessive epistasis, *i.e.*, out of the two pairs of genes, the recessive epistatic gene masks the activity of the dominant gene of the other gene locus. The dominant **A** expresses itself only when the epistatic locus **C** also has the dominant gene if the epistatic locus has recessive gene **c**, gene **A** fails to express.

Triangular AABB × Top Shaped aabb ------- **Parents**

AaBb × Triangular AaBb -------- F_1

	AB	**Ab**	**aB**	**ab**
A B	AABB Triangular	AABb Triangular	AaBB Triangular	AaBb Triangular
Ab	AABb Triangular	AAbb Triangular	AaBb Triangular	Aabb Triangular
aB	AaBB Triangular	AaBb Triangular	aaBB Triangular	aaBb Triangular
ab	AaBb Triangular	Aabb Triangular	aaBb Triangular	aabb Top shaped

F_2

Example : In mice agouti colour, characterised by banding of hairs is controlled by gene **A**, which is hypostatic to recessive allele **c**. The dominant epistatic gene **C** in absence of **A** gives black coloured mice and in presence of dominant gene **A** gives agouti, but dominant gene **A** is unable to produce agouti colour in absence of gene **c**. Therefore, recessive c gene acts as epistatic gene **A**.

(iv) **Duplicate genes (15 : 1 ratio) :** Sometimes two pairs of genes located on different chromosomes determine the same phenotype. These genes are said to be *duplicate* of each other. The dominant triangular fruit shape of *Capsella bursa pastoris* (shepherd's purse) is determined by two pairs of genes, say **A** and **B**. If any of these genes is present in dominant form, the fruit shape is triangular. In double recessive forms the fruits are top shaped and thus we get a 15 (triangular) : 1(top shaped) ratio in F_2 generation.

(v) **Collaborator genes :** In collaboration two gene pairs, which are present on separate loci but influence the same trait, interact to produce some totally new trait or phenotype that neither of the genes by itself could produce.

Example : Inheritance of combs in poultry, where two genes control the development of comb.

P_1 Generation Rose Comb RR pp × pea Comb rr PP

Gametes Rp, rP

Walnut comb Rr Pp

F_1 Generation

Gametes ♂

♀ \ ♂	RP	Rp	rP	rp
RP	RR PP walnut 1	RR Pp walnut 2	Rr PP walnut 3	Rr Pp walnut 4
Rp	RR Pp walnut 5	RR pp rose 6	Rr Pp walnut 7	Rr pp rose 8
rP	Rr PP walnut 9	Rr Pp walnut 10	rr PP pea 11	rr Pp pea 12
rp	Rr Pp walnut 13	Rr pp rose 14	rr Pp pea 15	rr pp single 16

Gametes ♀

F_2 Generation = { Walnut : 9, Rose : 3, Pea : 3, Single : 1 }

Fig : Inheritance of rose and pea comb in poultry

(a) Gene **R** gives rise to **rose comb**.

(b) Gene **P** produces **pea comb**. Both rose and pea combs are dominant over single comb.

(c) Gene **R** and **P** for rose and pea comb together produce a new phenotype the *walnut comb.*

Wyandotte variety of domestic chicken possesses rose comb, whereas Brahmas have pea comb. Bateson crossed rose-combed Wyandottes and pea–combed Brahmas. The F_1 chickens developed walnut comb, a phenotype not expressed in either parent. When F_1 chickens mated among themselves, the resultant F_2 chickens exhibited the familiar dihybrid ratio 9 : 3 : 3 : 1. These four phenotypes were – walnut comb, rose comb, pea comb and single comb in the ratio of 9 : 3 : 3 : 1. Out of these four phenotypes two phenotypes were different from those expressed in the parents.

The analysis of F_2 results indicates that the presence of two dominant genes **R** and **P** results in the walnut comb. The double recessive **(rrpp)** genotype produces single comb. The rose comb develops when dominant gene for rose comb is present and dominant gene for pea comb is absent **(RRpp)** whereas pea comb develops when gene for rose comb is recessive and gene for pea comb **(P)** is dominant **(rrPP)**.

(vi) Pleiotropic effect of genes

(a) **Lethal genes :** Certain genes are known to control the manifestation of some phenotypic trait as well as affect the viability of the organism. Some other genes have no effect on the appearance of the organism but affect the viability alone. These genes are known as lethals or semilethals depending upon their influence. Complete lethal genes in homozygous condition kill all or nearly all homozygous individuals, while in case of semilethal genes some homozygous individuals are able to survive. The lethal genes are always recessive for their lethality and express the lethal effect only in homozygous condition.

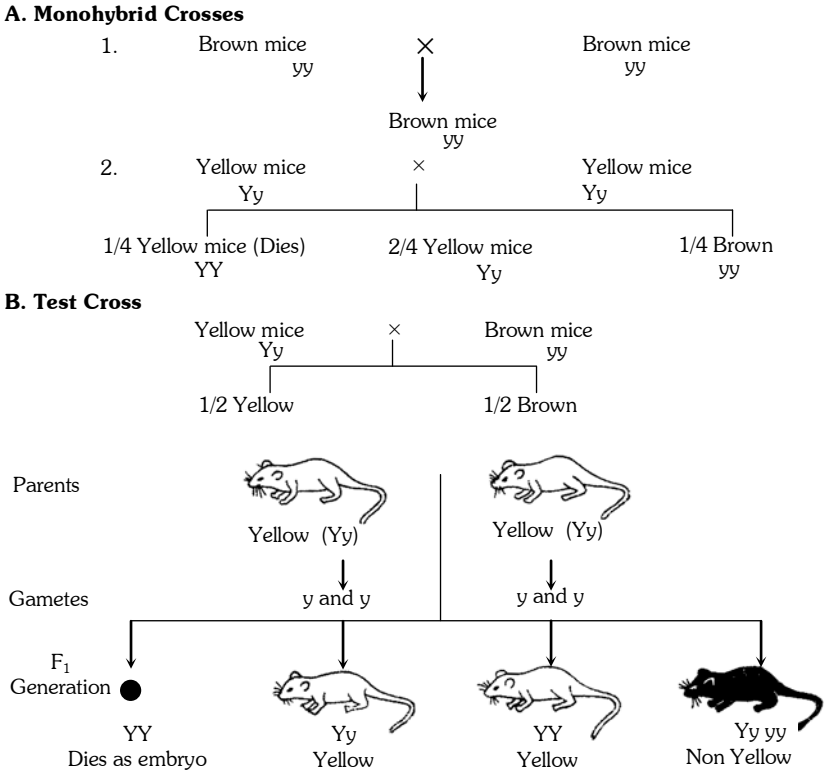

Fig : Inheritance of lethal gene-Y in mice

Dominant lethals : The dominant lethal genes are lethal in homozygous condition and produce some defective or abnormal phenotypes in heterozygous condition. Their most serious effect in heterozygous may also cause death. Following are the examples of dominant lethal genes.

Example – Yellow lethal in mice : A well known example of such lethals is from mice, given by *Cuenot.* He found that the yellow mice never breed true. Whenever the yellow mice were crossed with yellow mice, always yellow and brown were obtained in the ratio of 2:1. A cross between brown and brown mice always produced brown offsprings and a cross between brown and yellow produced yellow and brown in equal proportions.

In 1917, *Stiegleder* concluded that yellow mice are heterozygous. The homozygous yellow ($1/4^{th}$ of the total offsprings) dies in the embryonic condition. When there unborn ones are added to the 2:1 ratio of yellow and brown, these form typical 3:1 ratio. *Cuenot* suggested that gene Y has a multiple effect.

Note : • It controls yellow body colour and has a dominant effect.

• It affects viability and acts as a recessive lethal.

Example – Inheritance of sickle cell anaemia in man : The disease sickle cell anaemia is caused by a gene (Hb^S), which is lethal in homozygous condition but has a slight detectable effect in the heterozygous condition. The homozygous for this gene (Hb^s/ Hb^s) generally die of fatal anaemia. The heterozygotes or carriers for Hb^s, (*i.e.*, Hb^A / Hb^s) show signs of mild anaemia as their **R.B.C.** become sickle shaped in oxygen deficiency.

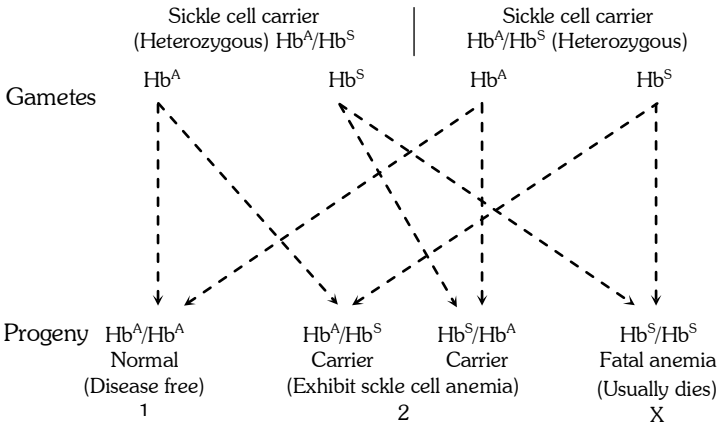

Fig : Diagram depicting inheritance of lethal gene for sickle cell anaemia

A marriage between two carriers, therefore, results in carrier and normal offsprings in the ratio of 2:1. The variation in 3:1 ratio is due to the death of homozygous and incomplete dominance of normal gene over recessive gene for sickle cell anaemia in which glutamic acid is substituted by valine in β chain. Which is disorder in Africans that reduce oxygen uptake.

Example – Brachyphalangy : Persons exhibiting brachyphalangy have short fingers apparently having two joints in their fingers, the middle bone being greatly shortened and often fused with one of the other two bones of the finger. Mohr described one case where one child was born without any fingers or toes and did not survive. Two other children showed short fingers and one was normal. This is exact 1 : 2 : 1 ratio.

Example – Huntington's chorea in man : The gene causing Huntington's chorea in man can express itself even when a single dominant allele is present. In both homo and heterozygous condition, the gene expresses itself only at middle–age, usually after forty years. The person suffers from muscular failure, mental retardation and finally death. Thus a dominant gene in heterozygous condition may also produce lethal effect. The gene is transmitted to next generation only because it expresses itself only after the start of reproductive period.

Recessive lethals : The recessive lethals produce lethal effect only in homozygous condition. Their heterozygotes are normal. Therefore, recessive lethals remain unnoticed in the population but are established in the population because female are carrier for lethal gene. These are detected only when two heterozygous persons get married.

Example – Tay Sach's lethal : The recessive lethal gene for Tay Sach's disease causes death of young children only in homozygotes which are unable to produce enzymes needed for normal fat metabolism. The accumulation of fat in nerve sheaths hampers transmissions of nerve impulse leading to poor muscular control and mental deficiency.

(vii) **Qualitative inheritance :** Qualitative inheritance or monogenic inheritance is that type of inheritance in which one dominant allele influences the complete trait, so that two such allele do not change the phenotype. Here dominant allele is monogene.

(viii) **Quantitative inheritance :** Quantitative inheritance or polygenic inheritance can be defined as, '*two or more different pairs of alleles which have cumulative effect and govern quantitative characters*. The quantitative

inheritance is due to incomplete dominance. It has been suggested that the multiple gene inheritance may have following characteristics:

(a) The effects of each contributing gene are cumulative or additive.

(b) Each contributing allele in a series produces an equal effect.

(c) There is no dominance involved.

(d) Epistasis does not exist among genes at different loci.

(e) No linkage is involved in the process.

(f) Effects of environment are absent or may be ignored.

Example – Human skin colour : This character was studied by *Davenport*, 1913 in the marriages between negroes and whites. The F_1 offsprings arising as a result of these marriages are called as mulattoes. The human skin colour is determined by two pairs of genes, P_1 and P_2. A negro having very dark skin with four colour genes *i.e.*, $P_1P_1\ P_2P_2$, when married to a white with no colour gene ($p_1p_1\ p_2p_2$) produce mulattoes with only two colour genes. These mulattoes show intermediate type of skin colour. If this mulatto is married to a similar genotype, the inheritance of pigment forming gene in F_2 offspring shall be as under:

Very dark	–	4 Colour genes	–	One
Intermediates	–	3 Colour genes	–	Four
	–	2 Colour genes	–	Six
	–	1 Colour genes	–	Four
White	–	No colour gene	–	One

If the mulatto is married to a pure white (test cross), the distribution of skin colour shall be as follows:

25% offsprings with two colour genes (P_1 and P_2),

50% offsprings with one colour gene (P_1 or P_2)

25% offsprings with no colour gene

If the mulatto is married to a negro (back cross) the distributions of skin colour shall be as under:

25% offspring with four colour genes

50% offspring with three colour genes

25% offsprings with two colour genes

Example – Ear size in maize : Emerson and East (1913) studied the for the ear length in maize**.** The size difference between two strains of maize is generally due to two or more pairs of genes. If it is due to two pairs of genes, the ratio of different sizes shall be 1 : 4 : 6 : 4 : 1. But, if it is due to three pairs of genes, the size ratio shall be 1 : 6 : 15 : 20 : 15 : 6 : 1.

Example – White spotting in mice : This trait is also polygenic for it is governed by two or more pairs of gene. Depending upon the involvement of the number of gene pairs for determine the trait, the ratios also vary in the manner stated above.

Example – Grain colour in wheat : This character was examined by *Nilsson-Ehile* 1908, which is similar as polygenic gene of skin colour in human. When a red grain was crossed with a white, the F_1 offspring produced light red grains due to incomplete dominance of red over white. The ratio is come out as 1 : 6 : 15 : 20 : 15 : 6 : 1.

(ix) **Multiple alleles :** The multiple alleles can be defined as a set of three, four or more allelomorphic genes or alleles, which have arisen as a result of mutation of the normal gene and which occupy the same locus in the homologous chromosomes. Characters of multiple alleles are following –

(a) Multiple alleles occupy the *same locus* with in the homologous chromosomes. It means only one member of the series is present in a given chromosome.

(b) Since only two chromosomes of each type are present in each diploid cell, only two genes of the multiple series are found in a cell and also in a given individual.

(c) The gametes contain only one chromosome of each types, therefore, only one allele of the multiple series in each gamete.

(d) Crossing over does not occur in the multiple alleles.

(e) Multiple alleles control the same character, but each of them is characterised by different manifestation. *Sturtevent* has summarised it that *they carry the same function but with varying degree of efficiency*.

(f) The multiple alleles of a series are more often related as dominant and recessive. More commonly, the normal gene is dominant to all other mutant alleles. Even the intermediate members of the series may be related as dominant and recessive, or they may exhibit codominance. Therefore, multiple alleles act in some way to control the various steps in a chemical reaction.

Examples : Coat colour in rabbit, blood group in human beings.

(x) **Pedigree analysis :** As man is not a suitable material for genetic research, the human genetics is studied from different point of view. Pedigree analysis is one such method based on Mendelism. It was started by *Galton*.

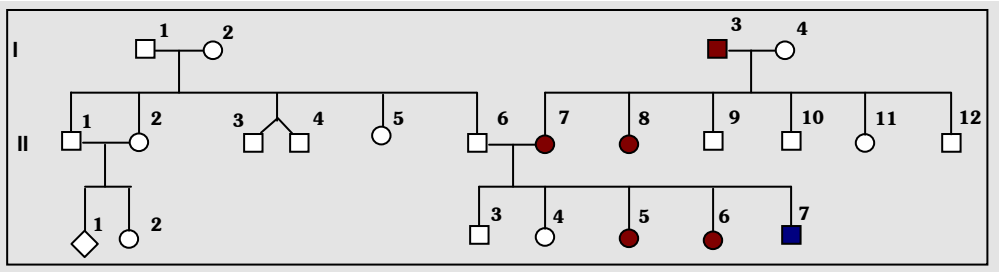

Fig : Pedigree analysis of three generations

A *pedigree* is a record of inheritance of certain genetic traits for two or more generations presented in the form of a diagram or family tree or case history or genealogy. Pedigree analysis is a system of analysis of pedigree to find out the possibility of absence/presence of a particular trait in the progeny. It is mainly employed in domesticated *animals* and *men* also. The person from whom the case history of a pedigree starts is called *Proband* (called *Propositus* or Prosipitus if it is male and *Proposita* if it is female). The children are called **Sibs**. Empty/open circles and squares represent normal female ( ) and normal male ( ) solid/shaded (●/■) symbols stand for those which bear the trait under study. ◑ or ⊙ represent carrier normal female having recessive allele of the trait under study