

GENETICS BASIS OF INHERITANCE

INTRODUCTION

The biological basis of the Continuity of life and perpetuation of species is due to the remarkable characteristic of living organisms to reproduce i.e. transmission of hereditary characters from the parents to the offsprings.. Therefore offsprings resemble their parents and they resemble each other also. But are they exactly identical to their parents? Or do they show certain -differences also? Have you noticed differences in kittens produced by cats and pups produced by dogs? If you observe minutely, you will definitely notice the differences also. But cat will always produce kittens and dogs will produce pups. Pea seeds will germinate into pea plant and mango seed will grow into a big mango tree. Therefore **'Like begets like' is the law of the living world.**

Every living organism possesses characteristics of a particular species but can be identified from others of the same species due to certain differences. Similarities and differences are the two aspects of the same mechanism of inheritance.

Heredity: is defined as transmission of characters from one generation to the next or from parents to offsprings. The different hereditary characters include colour of skin, eye, hair, shape of fruit, shape of wings etc.

Variations: are the visible differences between the parents and offsprings or, among the offsprings of the same parents and among the individuals of the same species. (Variations play important role in the evolution of new species.)

Genetics : is the branch of biology that deals with heredity and variations. The term genetics was coined by William Bateson in 1906. It is derived from Greek word genesis is to grow or **“ organization of an organism”**

The first scientific explanation regarding inheritance was given by Gregor Johann Mendel in 1866 by performing experiments on garden pea in a very scientific manner and proposed principles which are now popularly known Mendel's Laws of Inheritance and his epoch making work is known as **“Mendelism”**. On the basis of experiments which he carried out on pea plant ,showed that genetics is the experimental branch of Biology. He laid down the foundations of genetics therefore he is called **“Father of Genetics”**. After 19th century many scientists worked on different organisms and observed deviations from Mendel's ratios. Here we have studied both Mendelian inheritance as well as deviations to Mendelian principles.

MENDELIAN INHERITANCE

Organisms produced by asexual reproduction or plants produced by vegetative propagation can be called clones and are identical to their parents. Organisms produced by sexual reproduction are called offsprings, and they are not identical to either of their parents but inherit some of the characteristics from both the parents. They may show intermediate characters. Therefore, till 19th century, it was believed that there is a blending inheritance. Blood or heredity units of both the parents get mixed in the offsprings.

This theory however, cannot explain why in some cases offsprings resemble only one of the parents? For example offsprings of a black and a white horse were black and not grey. Sometimes offsprings resemble their grandparents or uncle or aunt and not their parents. Theory of blending inheritance cannot explain this.

Johann Mendel was the first one to say, that the characters are transmitted from one generation to the next through particles, which he called 'factors', that are now known as genes. Mendel was genius, much ahead of times as he has given this concept of factor before the discovery of mitosis, meiosis, chromosomes and without use of microscope. Let us know his biography in brief before learning about his experiments and laws.

Gregor Johann Mendel (1822-1884):

Gregor Johann Mendel was born on 22nd July 1822 to family of a poor farmer in a village, in Austria. After graduation he joined the Augustinian Monastery of Brunn as a priest. For higher studies in physics, mathematics and natural science, he was sent to University of Vienna. On his return he continued to work as a priest and teacher.

In 1856 he began his famous hybridization experiments on garden pea. In 1865 he presented his work in a paper entitled **"Experiments in plant hybridization"** at the meeting of the Natural History Society of Brunn. It was published in the journal "Annual Proceedings of Natural History Society" in 1866.



Unfortunately, his work remained unnoticed. He died in 1884, as a highly respected priest and teacher but unrecognized as a scientist.

In 1900, his findings were rediscovered by three scientists; Hugo De Vries from Holland, Karl Correns from Germany and Erich Tschermak from Austria, independently. DeVries found Mendel's research paper and he got it published in

Flora, (1901). During these years mitosis meiosis were discovered and the scientific world realized the greatness of Mendel and his work. Based on Mendel's principles/fndings, Correns postulated three laws that are known Mendel's Laws of Inheritance and a new branch 'genetics' in the field of biological science was opened.

Mendel's Experiments: How are the characteristics transmitted from parents to the offspring? Why there are similarities as well as differences? How certain, traits reappear in grand children? Answers to all these questions were to be found out through experimentation. Can you think about suitable experimental material for hybridization experiments? What characteristics the experimental material should have?

Selection of material.

Mendel selected garden pea (*Pisum sativum*) for his experiments because of following characteristics.

- 1) Garden pea is an annual plant and completes the life cycle within three to four months. Due to the short life span result can be obtained in very short time.
- 2) It is easy to cultivate and it does not required much more care.
- 3) The pea plant has various contrasting characters among its different varieties such as stem may be tall and dwarf , cotyledon may be green or yellow, violet and white flowers etc. There were no intermediate characters.
- 4) Flowers are bisexual and naturally self pollinating , thus pure line variety can be easily available / obtained from them.
- 5) The flowers are sufficiently large and are large enough for easy emasculation required for artificial cross.
- 6) Pea plant neither to large nor to small ,thus they can be easily handle.

Selection of characters (contrasting traits)

Mendel selected in all fourteen different varieties of pea plant and grouped them into seven characters/ pairs. Each pair was considered for a specific trait such as flower colour or seed shape or stem length etc. Two members of each pair consist of one contrasting character of the chosen trait. E.g. In a pair selected for stem length one variety had tall stem while the other had dwarf stem. These seven pairs of contrasting traits are shown in table .Before learning about Mendel's Experiments let us get acquainted with genetic terms and symbols.

Character	Dominant trait	Recessive trait
1 Stem height	 Tall (T)	 Dwarf (t)
2 Seed colour (Cotyledon)	 Yellow (Y)	 Green (y)
3 Seed shape	 Round (R)	 Wrinkled (r)
4 Pod colour	 Green (G)	 Yellow (g)
5 Pod shape	 Inflated (I)	 Constricted (i)
6 Flower position	 Axial (A)	 Terminal (a)
7 Flower colour	 Coloured (C)	 White (c)
7(a) Seed coat colour	 Coloured (C)	 White (c)

Mendel's seven pairs of contrasting

Genetic Terminology:

- **Character**- It is a feature of the organism e.g. Height of stem.
- **Trait** - An inherited character and its detectable variant e.g. Tall or dwarf. (Some traits can be environmentally determined or modified)
- **Factor**- It is a unit of heredity, a particle present in the organism which is responsible for the inheritance and expression of a character.
- **Gene** - It is a particular segment of DNA which is responsible for the inheritance and expression of that character which carries information from one generation to next, Gene is equivalent to mendelian factor..
- **Homologous chromosomes** - The morphologically and structurally similar chromosomes present in a diploid cell where one set of chromosome comes from female parent and other comes from male parent are called homologous chromosomes.
- **Alleles or Allelomorph**- The two (or more) alternative forms of a gene (factor) are called alleles of each other. They occupy identical loci (positions) on homologous chromosomes and control the expression of same character. **Ex.** In a genotype (Tt) T is present on one chromosome and t is present on other chromosome but at same locus such a pair of factor or gene called alleles.
- **Dominant** - It is an allele that expresses its trait even in the presence of an alternative allele, i.e. in heterozygous condition. (Only dominants express in hybrids.) **OR** Out of the two alternating forms of a trait, the one which appears in F₁ generation is called dominant trait or dominant allele. **Ex.** In a cross between homozygous tall pea plant with dwarf plant all the F₁ hybrid produced were found to be heterozygous tall plant, thus tallness is the character which is found to be dominant.
- **Recessive** - It is an allele which is not expressed in the presence of an alternative allele, i.e. in heterozygous condition. It expresses only in the presence of another identical allele. **e.g.** In the same example of above cross dwarfness is recessive character because it is totally suppressed.
- **Phenotype** - The external appearance of an individual for any trait is called phenotype for that trait. **E.g.** Height of plant is tall and dwarf are the phenotype.
- **Genotype** - It is representation of the genetic constitution of an individual with respect to a single character or a set of characters, **e.g.** in pea, tall plants can

have genotype TT or Tt and dwarf will have tt. (Entire genetic constitution of an organism is called 'genome'.)

- **Homozygous** –The organism or an individual possessing similar alleles for a particular trait is called homozygous or pure for that trait. Homozygous breeds true to the trait and produces only one type of gamete, **e.g.** A plant with genotype TT and dwarf plant with tt is homozygous for tallness.
- **Heterozygous** - The organism or an individual possessing dissimilar alleles for a particular trait is called heterozygous or hybrid for that trait. Heterozygous does not breed true for that trait and produce two types of gametes, **e.g.** A plant with genotype (Tt.) is said to be homozygous for tallness.
- **Pure line** - An individual or a group of individuals (population) that is homozygous or true breeding for one or more traits.
- **Hybrid**- A heterozygous individual form by the parents having contrasting character is said to be hybrid. is said to be hybrid. **e.g.** Individual with genotype (Tt) is said to be hybrid.
- **Monohybrid cross** - It is cross between two parents differing in a single pair of contrasting character is known as monohybrid cross. **e.g.** Hybrid tall produced in a cross between pure tall and dwarf parent.
- **Monohybrid ratio** : It is the phenotypic ratio which is obtained in F₂ generation from monohybrid cross . Mendel's monohybrid ratio is always 3:1
- **F₁ generation OR First filial generation** - (F stands for filial meaning offsprings produced in sexual reproduction. Latin word filin means a son). The progeny (offsprings) produced from a cross between two parents (P₁) is called first filial or F₁generation.
- **F₂ generation OR second filial generation** – The progeny resulting from selfing or interbreeding of F₁ individuals offsprings is called second filial or F₂ generation. It shows two or more types of individuals in a particular ratio.
- **Dihybrid cross** : It is the cross between two parents differing in two pair of contrasting character. **e.g.** Hybrid tall and red plant are produced in a cross between homozygous tall with red colour and homozygous dwarf and white colour of flower.
- **Dihybrid ratio** : The phenotypic ratio obtained in F₂ generation from dihybrid cross is called dihybrid ratio. Mendel's dihybrid ratio is always 9:3:3:1

- **Punnett Square** – (Punnet 1927) : It is a square divided into smaller squares which shows the mathematical or probable result of a cross , both phenotypic and genotypic.

Parental generation	P or P ₁
First filial generation	F ₁
Second filial generation	F ₂
Third filial generation	F ₃
Dominant -	Capital 'T' for Tall, Capital 'Y' for Yellow
Recessive -	Small 't' for dwarf, Small 'y' for yellow
Female -	♀
Male -	♂

Procedure of the Experiments:

Mendel was very methodical. He first considered (studied) inheritance of just one trait at a time unlike others who had considered the organism as a whole. Then he studied two traits together and then three, i.e. he performed monohybrid, dihybrid and trihybrid crosses.

He started with pure lines i.e. true breeding plants. He meticulously maintained complete record of actual number of each type of offsprings. The quantitative as well as qualitative record was kept for three generations. He conducted ample crosses and reciprocal crosses to eliminate chance factor.

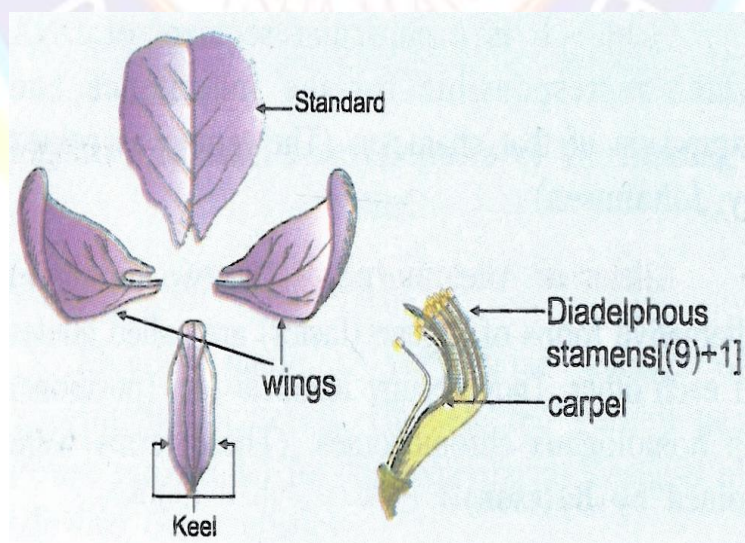
He conducted experiments in three steps as follows:

Step 1- Selection of parents and obtaining pure lines - : In the first step he selected a pair of parents with contrasting form of the chosen trait . Then he obtained each parent (plant) in pure form by repeated self fertilization. Ex. Mendel ensured that the plant selected for tallness produce only tall plant and for that he self fertilize a tall pea plant for many generations till the resulted offsprings always produced only tall plants likewise he got genetically pure variety for dwarf pea plant.

Step 2- Artificial cross of the selected parents to raise F₁ generation : This step involved crossing of the selected parents of the pair , one plant was used as the male parent and the other as the female parent. Pollen from the male were dusted on the stigma of the female parents for cross pollination Mendel conducted reciprocal crosses also.e.g. In a cross between tall and dwarf variety in one cross , tall variety was considered as a male parent and dwarf variety as female parent while in reciprocal crosses of the same parents, dwarf variety was used as male parent and the tall variety as a female parent. This may be represented as follows.

	Male parent	Female parent
Cross I	Tall variety	Dwarf variety
Reciprocal crosses	Dwarf variety	Tall variety

He was found that all the offspring develop are similar to one the parent (i.e. only tall plants are produced) while these generation is called F₁ generation and offspring are called F₁ hybrid. (Emasculation is removal of stamens well before anthesis i.e. opening of flower buds after formation of pollen grains. Therefore it is done in bud condition. Pea flowers are with papilionaceous -butterfly like- corolla with five petals. Stamens and carpels are enclosed in. the innermost two petals that are appressed together forming somewhat boat shaped structure called carina. Stamens and carpels mature simultaneously and naturally there is always self pollination. There are ten stamens but the filaments of nine are fused together and one is free. Therefore emasculatation is easy, just open the carina and remove (9) +1 by forceps.)



Pea flower

Step 3- Selfing of F₁ generation :

Mendel allowed the natural self pollination in the offsprings of F₁ hybrid and made further observation on the offsprings of second generation. He collected seeds separately and raised F₂ generation i.e. second filial generation, He was found to note both tall and dwarf offsprings in the second filial generation. This showed him that the character of dwarfness disappeared in first filial generation but again reappeared in second filial generation. Further, the tall and dwarf plants of second filial generation were always in the ratio 3:1 (3 Tall : 1 Dwarf) This ratio is called monohybrid ratio.

Monohybrid Cross Experiments and Monohybrid Ratio:

It is a cross between two pure (homozygous) parents in which inheritance pattern of only one pair of contrasting characters is studied is called monohybrid cross. In other words, it is a cross between two pure (true breeding) parents differing in a single pair of contrasting characters.

Step I : Mendel first selected pure tall pea plant as a female parent and pure dwarf pea plant as a male parent. He confirmed that they are true breeding by selfing them for three generations.

Step II : He then performed artificial cross, collected seeds, sowed the seeds and waited for the results. He was expecting that the offsprings (F₁ hybrids) will either be of intermediate height or 50% will be tall and 50% will be dwarf, But to his surprise, he found that all were Tall. He started thinking, where the dwarfness has gone? Why and how this character got eliminated? Is it eliminated for ever? Are the tall offsprings similar to the parental tall? To find out answers of all these questions, he continued further his experiments.

S.No.	Cross	F ₁	F ₂	Ratio
1.	Tall x dwarf	Tall	787 Tall, 277 dwarf	2.84:1
2.	Yellow x green seeds	Yellow seed	6022 Yellow, 2001 green	3.01:1
3.	Round x wrinkled seeds	Round seed	5474 Round, 1850 wrinkled	2.96:1
4.	Green x yellow pods	Green pods	428 Green, 152 yellow	2.82:1
5.	Inflated x constricted pods	Inflated pods	882 Inflated, 299 constricted	2.95:1
6.	Axial x terminal flower	Axial flower	651 Axial, 207 terminal	3.14:1
7.	Violet x white flower	Violet flower	705 Violet, 224 white	3.15:1
7a.	Grey x white seed coat	Grey seed coat	705 Grey, 224 white	3.15:1

Step III : He allowed-natural selfing of all the F_1 hybrids, collected the seeds, sowed the seeds and anxiously waited for the results. He found that dwarfness reappeared in some plants. In all, he had collected 1064 seeds of F_2 generation, from this 787 grew tall and 277 remained dwarf.

He thought that it may be due to female dominance, tallness was with ovules and dwarfness with tiny pollen grains. So he performed reciprocal cross i.e. now he selected tall plant as a male parent and dwarf as a female parent and repeated the procedure. He got similar results i.e. all tall in F_1 and dwarf in approximately 3:1 ratio in F_2 generation.

In this manner he did monohybrid crosses and reciprocal crosses with all the seven pairs of contrasting characters separately and obtained similar results, i.e. only one of the two characters was expressed in F_1 generation. In F_2 , the character which was expressed in F_1 , was in large number and the other in small number and the ratio was 3:1. This is called monohybrid ratio. In all Mendelian crosses it is 3:1.

Mendel's Interpretation and conclusion :

On the basis of consistent pattern of results obtained in all monohybrid crosses Mendel concluded that-

- 1) Characters such as height of stem, colour of seed etc. are inherited separately from parent to the offspring in the form of small particle called factor or determinant which are now called gene
- 2) Particular hereditary character is controlled by the pair of factor. One of the factors is contributed by male parent and the other by female parent. (Now we know that sexually reproducing higher organisms are diploid ($2n$).)
- 3) Each factor exists in two contrasting or alternative forms.- For example for the factor for height of the plant there are two forms, one for tallness and other for dwarfness. Now we know that there can be two or more forms of a gene which are called alleles or allelomorphs.
- 4) Out of the two contrasting character in the pair, one is of dominant and the another is recessive. Only dominant expresses in F_1 generation.5)
- 5) The factor controlling the particular character are separated from each other during gamete formation hence gamete are always pure.

He designated appropriate alphabets, capital letter for dominant allele and small letter for recessive allele. e.g. Tall- (T) dwarf (t), Round (R) wrinkled (r) and so on. The

organism that receives, recessive factor from both the parents expresses the recessive trait.

Explanations for the result of monohybrid cross:

Mendel started with pure lines and confirmed that the parents are true breeding. Therefore parental tall plant is with factors (TT) and dwarf with (tt). During gamete formation factors separate and only one enters in each gamete. Thus pure parents will produce only one type of gametes. Tall parent plant will produce all gametes with (T) and dwarf parent plant will produce all gametes with (t). Therefore all F₁ generation offsprings (hybrids) are with (Tt). Due to dominance they all grow tall. Thus all plants are tall in F₁ generation.

Mendel allowed selfing of F₁ hybrids. As factors separate during gamete formation F₁ hybrids will produce two types of gametes in equal proportion i.e. 50% of all male gametes will be with capital (T) and 50% with small (t). Similarly 50% of female gametes will be with (T) and 50% with (t). (Pea flowers are bisexual). There will be random fusion and each type of male gamete has equal chance to fuse female gamete with (T) or (t) and vice versa. This chance fusion will produce (2 x 2 = 4) four combinations, which fall into three categories i.e. there are three different genotypes. One out of every four is (TT) pure tall, two are (Tt) hybrid tall and again only one is (tt) dwarf. Thus only one out of four i.e. 25% of the F₂ generation offsprings will receive (t) from both the parents and therefore will be dwarf. Thus recessive trait reappears in F₂ generation and the ratio of dominants and recessives is 3:1. However all tall plants of F₂ generation are not same, only 1/3rd are pure tall and 2/3rd are hybrid tall. (The dwarfs are always pure as recessive trait cannot be expressed in the presence of dominant allele.) The ratio of **pure tall: hybrid tall : dwarf** is called genotypic ratio but the phenotypic ratio, 3:1 is called monohybrid ratio.

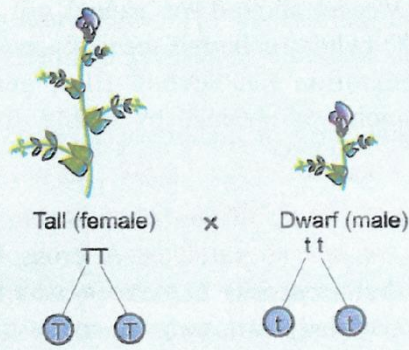
Thus Monohybrid ratio is defined as the phenotypic ratio of different types of offsprings (dominants and recessives) obtained in F₂ generation from monohybrid cross. It is 3:1 in all Mendelian crosses.

P generation

phenotype

genotype

gametes
(One type)



F₁ generation

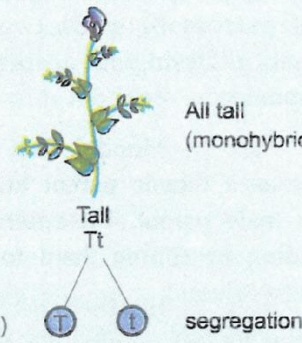
phenotype

genotype

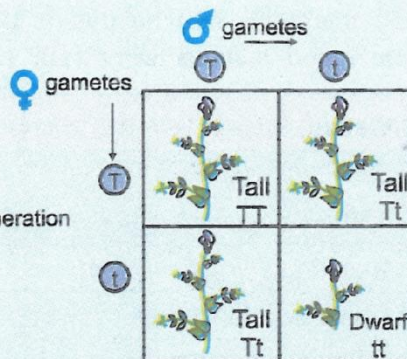
gametes
(Two types)

selfing of F₁ monohybrids

All tall
(monohybrids)



F₂ generation



Phenotypic ratio:- tall : dwarf
3 : 1

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

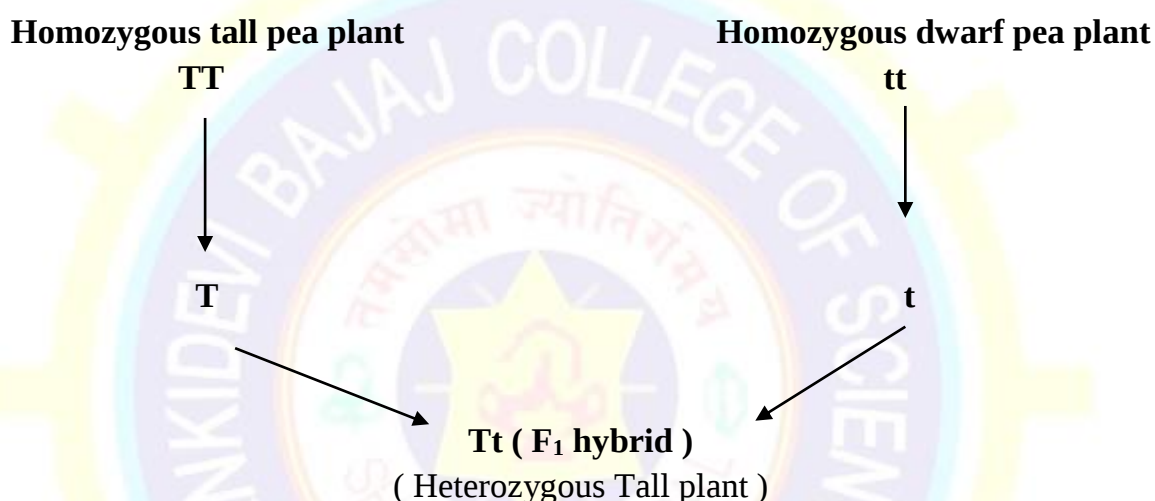
Mendel's laws of inheritance

❖ Law of Dominance or First Law of Inheritance

The law states that “ When a pair of allele or allelomorphs are brought together in F_1 hybrid then only one of them is expressed in F_1 generation masking the expression of other totally.” **OR**

The law states that “ When two homozygous parent differing in one or more sets of contrasting character are crossed then only one character is appear in F_1 generation , the character which is expressed in F_1 generation is called Dominant character and the character which is suppressed is called Recessive character.”

e.g. When a homozygous tall pea plant is crossed with dwarf pea plant then all F_1 hybrid produced were found to be heterozygous tall. Thus tallness is the character which is found or said to be dominant.



Each character is controlled by a pair of factors and when it is a pair of dissimilar factors only one factor expresses. Thus one is dominant and the other is recessive.

Law of dominance can be explained with the help of a monohybrid cross experiment

Some other examples of organisms and traits which show dominance are in guinea pigs, black coat colour is dominant over white. In human beings, curly hair is dominant trait over straight hair and brown eye colour over green or blue. In jowar, pearly grain is dominant over chalky grain and so on. However, there are many cases where the dominance is not complete or absent. We will learn about it later in this chapter under deviations from Mendelian inheritance. Phenomenon of dominance is significant as the harmful recessive traits are masked, not expressed in the presence of its normal dominant allele. For example, In humans a form of idiocy, and diabetes and hemophilia are recessive characters. Thus Law of dominance is significant and true but it is not universally applicable.

2. Law of Segregation or Second law of Inheritance:

The law states that when a pair of allele or allelomorphs are brought together in a F_1 hybrid, they remain together for a long time but do not contaminate or mix with each other and separate or segregate at the time of gametogenesis (gamete formation). Thus each gamete receives only one allele of a character either dominant or recessive and is pure for a given trait. Therefore, this is also known as Law of Purity of Gametes.

Law of segregation also can be explained with the help of monohybrid cross experiment.

Reappearance of recessive trait in F_2 generation proves the law.

All sexually reproducing higher organisms are diploid ($2n$) i.e. with two sets of chromosomes and gametes are haploid (n) with one set of chromosomes. Therefore, Law of segregation is universally applicable.

e.g. When a homozygous tall pea plant is crossed with a dwarf pea plant, then all F_1 hybrids produced were found to be heterozygous tall. This F_1 hybrid contains two factors, namely T and t , but these are separate at the time of gamete formation. Thus each gamete contains one gene or factor for a particular trait, i.e. T contains the gene for tallness and t contains the gene for dwarfness. Then gametes are always pure; hence the law of segregation is also said to be the law of purity of gametes.

Homozygous tall pea plant

TT



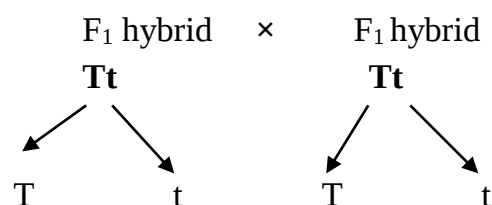
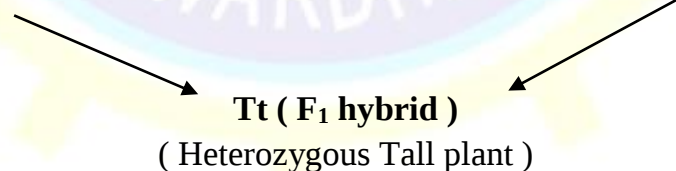
T

Homozygous dwarf pea plant

tt



t



Dihybrid Cross Experiments and Dihybrid Ratio:

Mendel established the principle of dominance and segregation on the basis of monohybrid crosses. However, in an organism, many characters are present and each is controlled by a pair of alleles. Does one pair of alleles affect / influence the inheritance pattern of other pairs of alleles? Or each pair is inherited independently as if in a monohybrid cross? To find out the answer, Mendel performed dihybrid crosses i.e. considered two characters / allelic pairs simultaneously and then trihybrid crosses.

Dihybrid cross : A cross between two pure (homozygous) parents in which the inheritance pattern of two pairs of contrasting characters is considered simultaneously is called a dihybrid cross. OR it is a cross between two pure (true breeding) parents differing in two pairs of contrasting characters.

Dihybrid : It is heterozygous for two traits and produced in a cross between two pure parents differing in two pairs of contrasting characters,

Dihybrid ratio : It is the phenotypic ratio of different types of offsprings (with different combinations) obtained in the F_2 generation of a dihybrid cross is called a dihybrid ratio. It is 9:3:3:1 in all Mendelian crosses.

Mendel performed many dihybrid crosses, with different combinations and every time he got a consistent pattern of results and the same ratio in F_2 generation. From this he established the principle of independent assortment.

He followed the same three steps which is a classical experimental design that was followed by many breeders after him and discovered different gene interactions. Let us see his very first dihybrid cross experiment.

Step 1 - Mendel selected a variety of pea plant having yellow and round seeds as a female parent and another variety having green and wrinkled seeds as a male parent. He obtained pure lines by selfing. He confirmed that yellow round is producing yellow round and green wrinkled is producing green wrinkled for three generations.

Step 2 - He performed an artificial cross i.e. dusted pollen grain from flowers of pure male parent on stigma of emasculated flower of female parent. In due course fruits (pods) were formed, he opened all the pods and collected seeds. All seeds of F_1 generation were yellow and round as expected. (Yellow is dominant over green and Round over wrinkled.)

Step 3 - He sowed the seeds and raised the plants of F_1 generation and allowed selfing. When pods were formed, he opened and collected seeds of F_2 generation. He was expecting 75% yellow round and 25% green wrinkled seeds. But he was surprised

to find that seeds were of four types i.e. F₂ generation seeds showed four different phenotypes as follows -

Yellow round 315; Yellow wrinkled 108; Green round 101; green wrinkled 32, (Total = 556). That is in 9:3:3:1 ratio.

Out of four different types, two were parental combinations (Yellow round, green wrinkled) and two were new / recombinations. (Yellow wrinkled, green round.)

Mendel performed ample dihybrid crosses and reciprocal crosses with different combinations. Every time he got same pattern of results i.e. uniform expression- (both dominant in F₁ generation. In F₂ generation always he got both dominant in large number, one dominant and the other recessive in less number, the other dominant and the first one recessive also equally less in number and both recessive were least in number. Thus in all Mendelian dihybrid crosses the ratio in which four different phenotypes occurred was 9:3:3:1 which is called dihybrid ratio.

Mendel applied the principle of probability in statistics, i.e. 'Probability of two independent events occurring simultaneously is the product of their individual probabilities.'

Dihybrid ratio is a product of two monohybrid ratios. (3:1) X (3:1) = 9:3:3:1.

From this Mendel concluded that inheritance of seed colour and inheritance of seed shape are the two independent events. Therefore, both dominants occurred in large number and both recessive in the least. The different phenotypes / combinations occur in the ratio 9:3:3:1 as follows;

Yellow round 3x3 = 9

Yellow wrinkled 3x1 = 3

Green round 1x3 = 3

Green wrinkled 1x1 = 1

If we consider ratio of (proportion of) yellow and green in F₂, it is yellow 9+3 = 12; green 3+1 = 4. Therefore the ratio is 12:4 i.e. 3:1. Similarly ratio of round (9+3) and wrinkled (3+1) is 12:4 i.e. 3:1.

From the appearance of new combinations in the F₂ generation, uniform pattern of results of dihybrid cross and dihybrid ratio, Mendel established principle of independent assortment. He performed trihybrid crosses also and then concluded that, when a dihybrid (or a polyhybrid) forms gametes, each gamete receives only one allele from each pair (due to segregation) and the assortment (distribution) of alleles

of different traits is totally independent of their parental combinations i.e. each allele of any one pair of alleles is free to enter the gamete with any allele from each of the remaining pairs of alleles. Thus a dihybrid forms four types of gametes due to segregation and independent assortment.

Explanation of results of dihybrid cross:

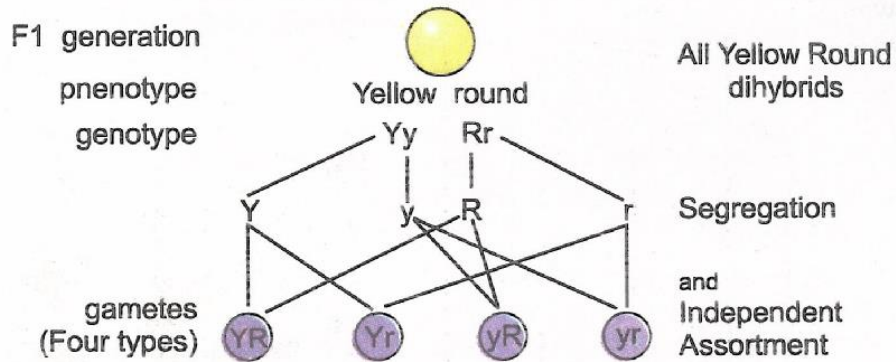
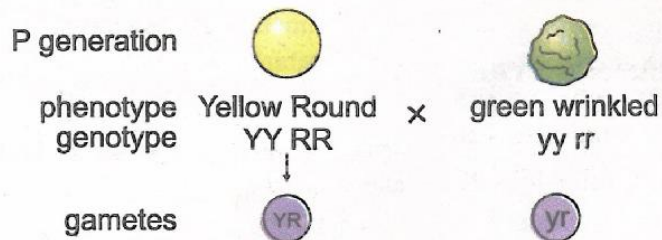
Mendel started with pure lines. Therefore, genotype of parent with yellow round seeds is YYRR and that of parent with green wrinkled seeds is yyrr. Both the parents are homozygous. Therefore, they would produce only one type of gamete i.e. YR and yr respectively. All F₁ dihybrid seeds resulting from the cross will be heterozygous for both the traits and with genotype YyRr. Due to dominance, all the seeds of F₁ generation will be yellow round.

Mendel allowed selfing of F₁ dihybrids. During gamete formation by the dihybrids, the alleles in both the pairs separate (law of segregation). Each gamete will receive only one allele from each pair. A gamete that receives 'Y' for colour may receive 'R' for shape or 'r' for shape. This would result in the formation of YR and Yr types of gametes. Similarly, a gamete that receives 'y' for colour may receive 'R' or 'r' for shape. This would result in formation of yR and yr types of gametes. Thus F₁ dihybrid would produce four different types of gametes in equal proportion. (Each type will be 25%).

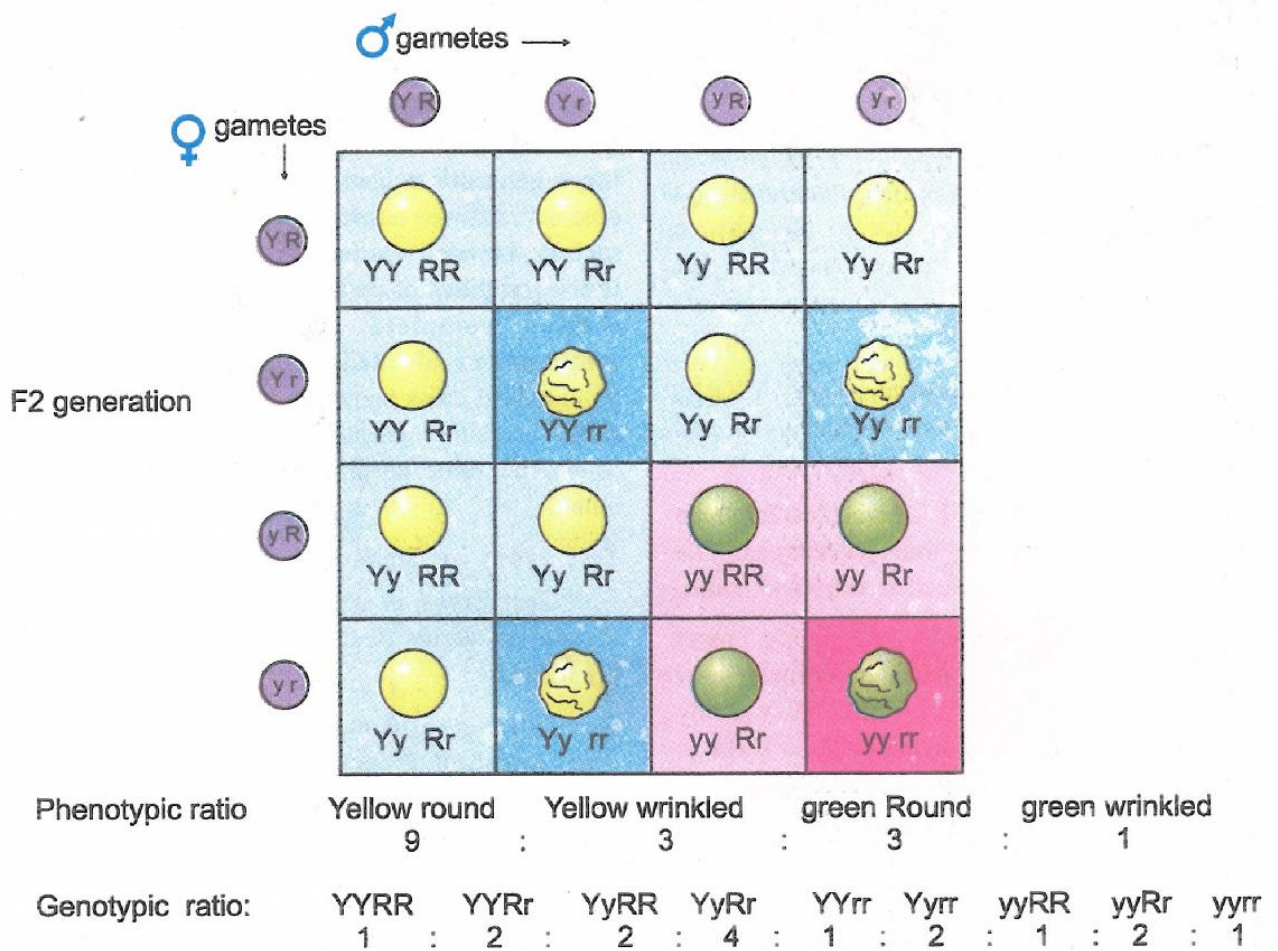
There would be four types of male gametes and four types of female gametes and, random fusion will take place during selfing. Due to this chance fusion (4 x 4 = 16) sixteen combinations which fall into nine categories as shown in Punnett square are possible.

The nine different genotypes are; YYRR, YYRr, YyRR, YyRr, YYrr, Yyrr, yyRR, yyRr and yyrr.

The genotypic ratio is 1:2:2:4:1:2:1:2:1. Due to dominance there are only four phenotypes. The phenotypic ratio 9:3:3:1, is called dihybrid ratio.



Selfing of dihybrids



Law of Independent Assortment or Third law of Inheritance :

Statement of law- “ When the parents differ from each other in two or more pairs of contrasting traits or characters are crossed, the inheritance of one pair of factors is independent to that of the other pair of factors.” In other words, when a dihybrid (or polyhybrid) forms gametes, assortment (distribution) of alleles of different traits is independent of their original combinations in the parents.

The mechanism law of independent assortment can be understood by considering the examples of dihybrid cross between round and yellow seeded pea plant with wrinkled and green seeded pea plant. The homozygous pea plant with Round Yellow seeds has the alleles RR and YY for the roundness and yellow cotyledons. Similarly the homozygous pea plant with wrinkled and green seeds contains the alleles rr and yy for wrinkleness and green cotyledons. The gametes which are produced by RRYy and rryy plants are RY and ry respectively. When both parents are crossed the union of both types of gametes takes place to give the F₁ hybrid (RrYy). The F₁ hybrid were found to be Round and Yellow plant showing the dominance of allele R for roundness over the r for wrinkleness and the dominance of allele Y for yellow colour over the recessive allele y for green colour. Now the F₁ hybrid have four types of alleles i.e. Y for Yellow , R for Round shape , r for wrinkleness and y for green colour. During gametogenesis these four alleles may combine in the following four combination.

- 1) The allele **R** may be associated with the allele **Y** to give rise **RY** combination
- 2) The allele **R** may be associated with the allele **y** to give rise **Ry** combination
- 3) The allele **r** may be associated with the allele **Y** to give rise **rY** combination
- 4) The allele **r** may be associated with the allele **y** to give rise **ry** combination

Thus four types of alleles are assorted independently to produce four types of gametes i.e. RY , Ry , rY , ry. From these four types of gamete four types of offsprings are produced in F₂ generation namely Round and Yellow , Round and green , Wrinkled and Yellow , and Wrinkled and green . These results have proved the law of independent assortment and showed that each pair of contrasting characters behave independently and bears no permanent association or relation with a particular character. The allele R was associated with allele Y in parent but it does not always remain associated with it and it is also associated with the allele y. Appearance of new combinations in F₂ generation proves the law.

Now we know that, many genes are located on one chromosome i.e. are linked. Therefore, they pass through gametes in the form of a linkage group. However recombinations are, due to the crossing over that takes place during

meiosis. Therefore, the law of independent assortment is applicable only for the traits which are located on different chromosomes. Thus law of independent assortment is not universally applicable.

Reasons for Mendel's success

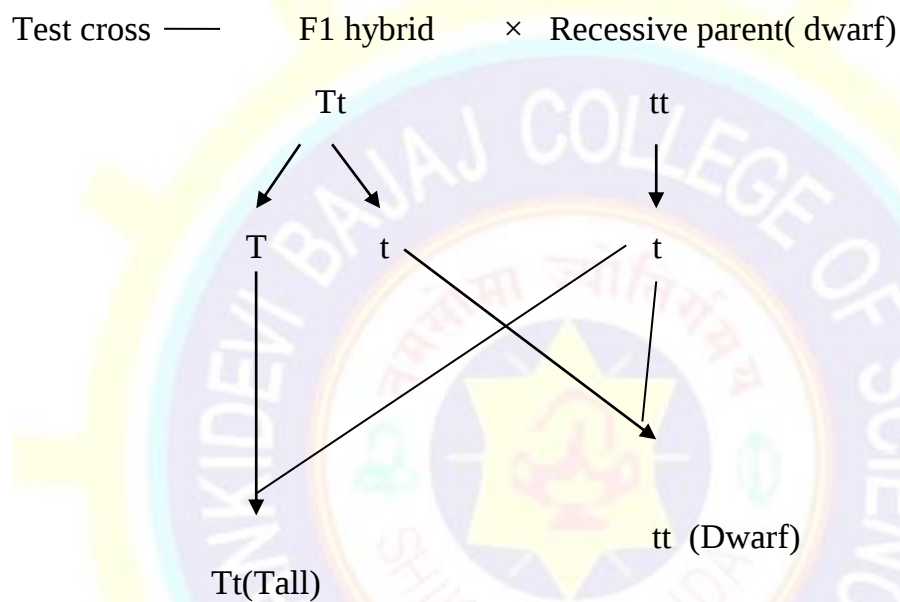
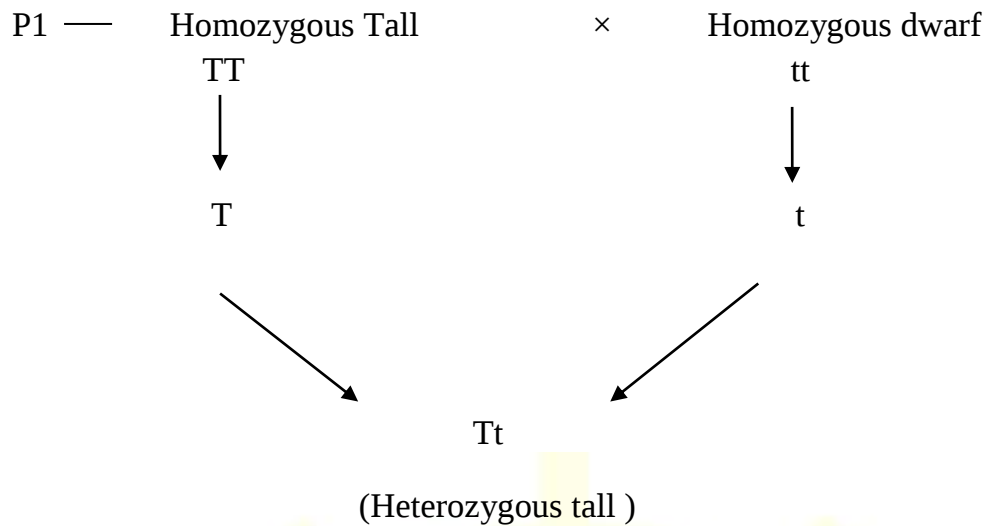
- ❖ Mendel used pure lines which he verified personally.
- ❖ His approach towards the experiments is logical and scientific.
- ❖ He analyzed the results using statistical method.
- ❖ He kept accurate records of his hybridization experiment(of every generation)
- ❖ His choice of pea plants which normally self fertilize.
- ❖ Luck factor also play very important role in his experiment.

Test cross or Back cross:

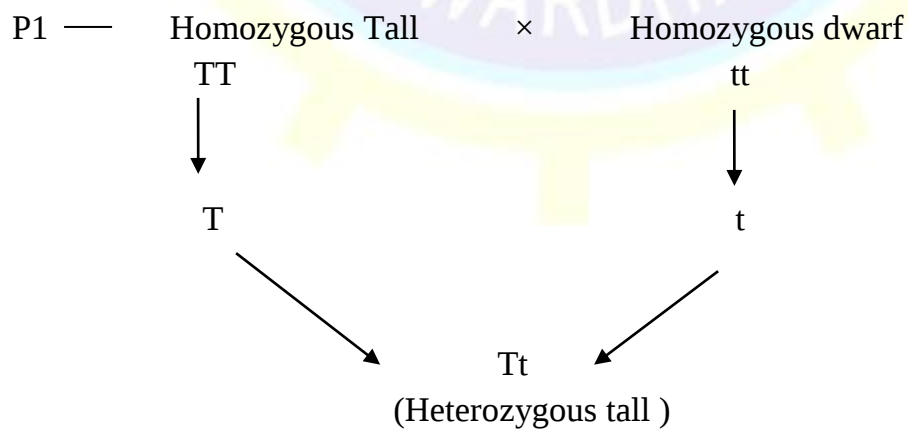
Test cross is a simple method devised by Mendel to verify (test) the genotype of F_1 or (F_2/F_3) hybrids. When F_1 individual are crossed with one of the parent from which they were derived, then such a cross is called back cross. In such back crosses, when F_1 is back crossed to the parent with dominant phenotype, no recessive individuals are obtained in the progeny. But when F_1 progeny is back crossed with its recessive parent, both phenotypes (i.e. dominant and recessive) appear in the progeny. While both of these crosses are back crossed, only the cross with the recessive parent is called test cross.

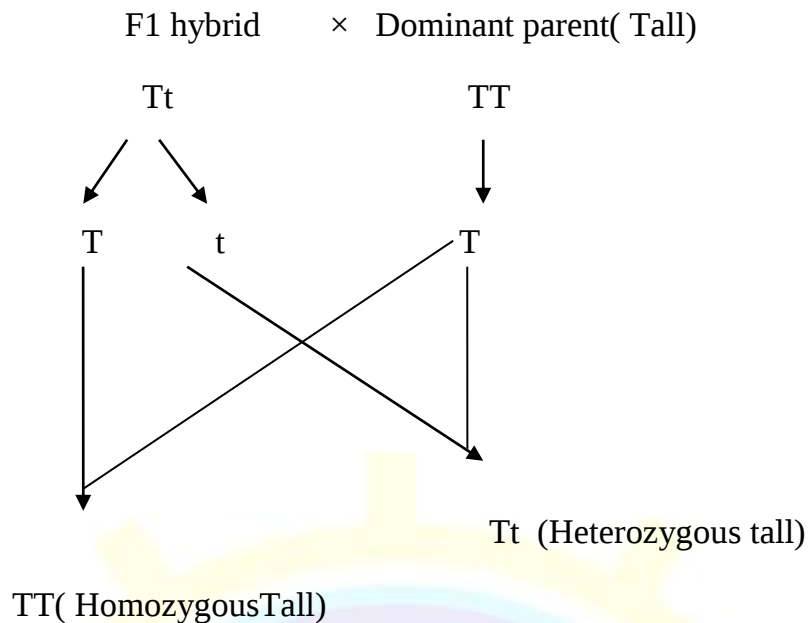
When hybrid expresses recessive trait, e.g. dwarf pea plant, its genotype is definitely 'tt', but if hybrid is tall, it is not possible to know its genotype. F_1 hybrids are definitely heterozygous but in F_2 generation all tall plants are not heterozygous, 25% are pure tall. Therefore to find out whether the tall plant (or any dominant expression) is homozygous or heterozygous test cross can be performed instead of selfing.

Monohybrid test cross – In monohybrid cross of pure tall and pure dwarf plant, all F_1 hybrids are tall and with genotype Tt. For the verification of their genotype they are crossed with recessive parent the progeny will be 50% tall and 50% dwarfs. This is because recessive parent will produce only one type of gamete t and hybrid will produce two types of gametes T and t. Therefore half the progeny will have genotype 'Tt' and half tt. Thus Tall and dwarf offsprings produced in the ratio 1:1. This is called monohybrid test cross ratio.



The cross between F1 hybrid and homozygous tall parent is also a back cross but this will produce all tall; 50% pure tall and 50% hybrid tall. Therefore back cross with dominant parent cannot be used to test the genotype.





Thus a test cross is a back cross but back cross is not Necessarily test cross.

Test cross can be used to find out genotype of any plant with dominant expression but it is not known whether it is pure or hybrid for that trait. In this case it is crossed with recessive plant with that particular trait. For example, there is a pea plant with violet flowers, a dominant trait, to find out its genotype it should be crossed with a pea plant with white flowers, a recessive trait. If all flowers produced are violet we can conclude that it is pure or homozygous and if we get violet and white in 1:1 ratio we can conclude that it is heterozygous. Therefore test cross can be defined as a cross between individual with unknown genotype for particular trait with a recessive plant for that trait.

Significance of test cross

- ❖ It is used to determine the genotype of plants whether they are homozygous.
- ❖ Test cross is also used to introduce useful recessive trait in the hybrids in rapid crop improvement -programmes. It is more popular with plant breeders for its simple 1:l ratio.
- ❖ It is easier and quicker method of obtaining the character in pure form.

Dihybrid test cross :

When homozygous yellow and round seeded pea plant is crossed with green and wrinkled seeded pea plant, all the F_1 hybrid produced were found to be heterozygous yellow and rounded pea plant. When this F_1 hybrid is test cross with its recessive parent i.e. with green and wrinkled seeded pea plant, all the offsprings produced in F_2 generation show the phenotypic ratio 1:1:1:1:

Homozygous Yellow & Round $\times$ Homozygous Green & Wrinkled

YYRR

yyrr



YR

yr



YyRr
(F_1 hybrid)

F_1 hybrid

$\times$ Recessive parent(Green & Wrinkled)

YyRr

yyrr

YR Yr yR yr

yr

♂ ♀ : YR Yr yR yr	YR	Yr	yR	yr
yr	YyRr(yellow round)	Yyrr (yellow wrinkled)	yyRr (green round)	Yyrr(green wrinkled)

DEVIATIONS FROM MENDELIAN RATIOS:

After the rediscovery of Mendel's laws, many scientists performed experiments on different plants and animals and many exceptions (deviations) to Mendelian ratios were observed. On the basis of these observations, different patterns of inheritance or gene interactions were discovered. These can be explained by suitably modifying Mendel's laws therefore this is called post-Mendelian genetics or Neo- Mendelian genetics.

It was observed that the phenotypic expression of a gene can be modified or influenced by the other gene.- These gene interactions are of two types; intragenic and intergenic.

- **Intragenic OR Interallelic Gene Interaction** : It occurs in between two allelomorphs of a single type of gene e.g. incomplete dominance, co-dominance and multiple alleles.
- **Intergenic OR Nonallelic Gene Interaction** : In this type of genetic interaction, the independent (non – homologous) genes located on the same or on different chromosomes interact with one another for the expression of single phenotypic trait of an organism.e.g. pleiotropy, polygenes, epistasis. supplementary complementary genes, etc.

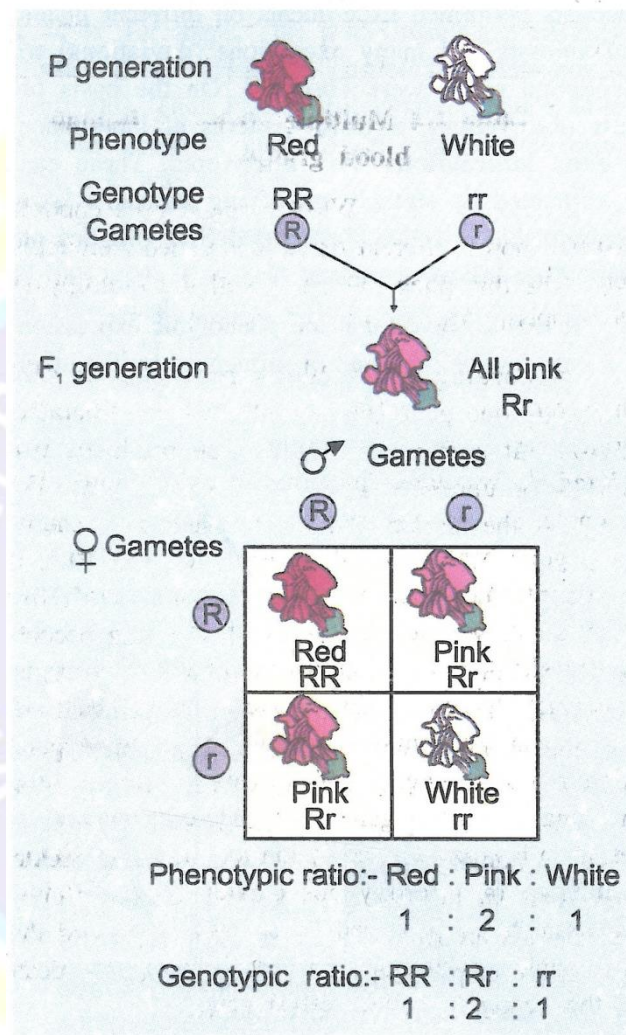
1. Incomplete dominance:

In incomplete dominance both the genes of an allelomorphic pair express themselves partially. One gene cannot suppress the expression of the other completely. Thus the pair is not as one dominant and other recessive. In such cases there is intermediate expression in F_1 hybrids.

e.g. In four o' clock plant, (*Mirabilis jalapa*), also just as in pea plants , the flower colour is controlled by a pair of alleles. Red flower colour is (RR) and white flower colour is (rr) genotypically. When these two are crossed (both are pure breeding) the F_1 heterozygote progeny will be pink (Rr) in colour. When F_2 is raised by selfing the F_1 , not only red and pink flower colours appear but the white colour also appears in the ratio of 1:2:1 red (RR.), pink (Rr) and white (rr) respectively.

Explanation of Results : while the inheritance pattern in four o clock plants is basically Mendelian , there is slight deviation, in that R is incompletely dominant over

r. Thus F_1 produces an entirely new phenotype not seen in the parents. Hence in four o'clock plants the heterozygous condition is always pink, while homozygous individuals are either red or white. Another significant feature in incomplete dominance is that the phenotype and genotype are always the same as there is no dominance. All characters (red, white) except pink can express themselves morphologically only in the homozygous condition. Hence the ratio of two progeny is always 1:2:1



2. Co-dominance — In Co-dominance, both the genes of an allelomorphic pair express themselves equally in F_1 hybrids. Such alleles which are able to express themselves independently even if present together in hybrids are called codominant alleles.

Classic example of co-dominance is coat colour in cattle. There are two types, one with red coat (skin with red colour hair) and the other with white coat (with white

hair). When red cattle (RR) are crossed with white cattle (WW), F₁ hybrids (RW) have roan colour. Roans have the mixture of red and white colour hair. Thus both the traits are expressed equally. In F₂ generation, red (RR), roans (RW) and white (WW) are produced in the ratio 1:2:1. Thus in co-dominance also genotype and phenotype ratios are identical.

In both, incomplete dominance and co dominance genotypic and phenotypic ratios are identical but in case of incomplete dominance the phenotype of hybrids is intermediate between the phenotypes of parents while in co-dominance both the genes are expressed equally.

Red coat colour of cattle

White coat colour of cattle

RR

WW

R

W

RW (Roan)

With Red and White colour skin

F₁ hybrid

×

F₁ hybrid

RW

RW

R

W

R

W

RR

RW

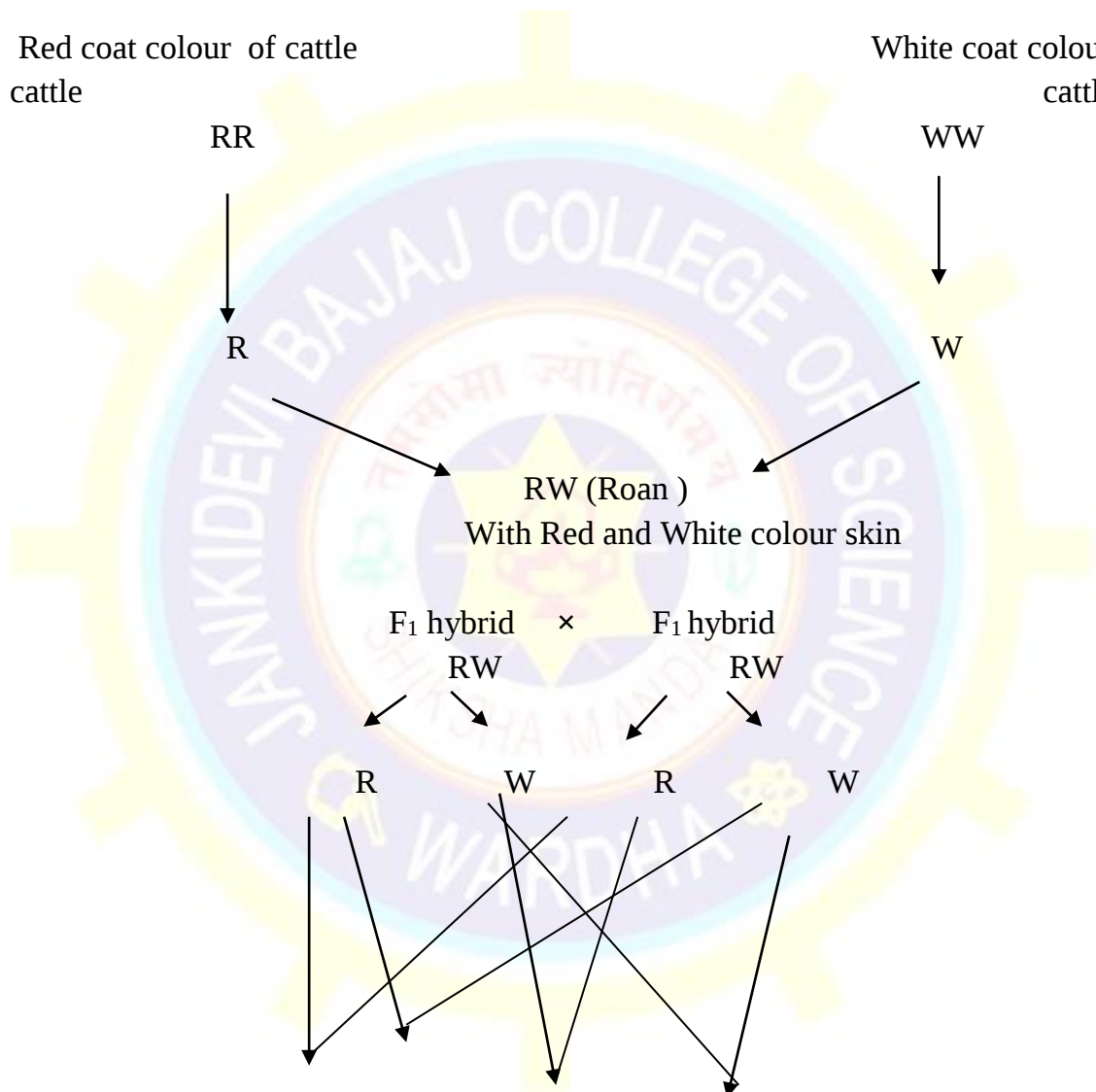
RW

WW

1 (Red)

: 2 (Roan)

: 1 (White)



Multiple alleles

More than two alternative forms (alleles) of a gene in a population occupying the same locus on a chromosome or its homologue are known as multiple alleles.

Characters of multiple allele :

- Multiple alleles of a series always occupy same locus in the chromosome.
- No crossing- over occurs within the alleles of a same multiple allele series.
- Multiple alleles always influence the same character.
- Multiple alleles arise by mutations of the wild type of gene.
- Wild type allele is nearly always dominant, while the other mutant alleles in the series may show dominance or there may be an intermediate phenotypic effect.
- When any two of the mutant multiple alleles are crossed, the phenotype is mutant type and not the wild type.
- A gene can mutate several times producing a series of alternative expressions.

The best examples of multiple allelic system have been observed in coat colour of rabbits, wings of Drosophila and blood groups in human beings.

Example 1 : In drosophila, a large number of multiple alleles are known. One of them is the series of wing abnormality ranging in size from normal wings to no wings. The normal wings is wild type and is denoted by Vg +. The extreme expression with no wings i.e. just stumps, is due to one allele vg, in homozygous condition. Following table shows the complete series.

PHENOTYPE	GENOTYPE
Normal wings	Vg +
Nicked wings	vgni
Notched wings	vgno
Strap wings	vgst
Vestigial wings	vg

ABO blood groups in human beings.

The gene I controls the ABO blood groups. It has three alleles; I^A , I^B and i .

The allele I^A and I^B produce a slightly different form of the antigen and allele i , does not produce any antigen. Since humans are diploid organisms, each person possesses any two of the three I gene alleles. I^A and I^B are co-dominants and are completely dominant over i . There are six different genotypes and only four different phenotypes, i.e. blood groups as follows; for example, alleles $I^A I^A$ or $I^A i$ develop A antigen (blood type A). The presence of alleles $I^B I^B$ or $I^B i$ produce B antigens (blood type B) and the codominant alleles $I^A I^B$ produce both A and B antigens which are distributed on different erythrocytes (blood type AB). ii genotype tends to produce blood type O (these alleles do not produce any antigen). The gene I for ABO antigenic characters of erythrocytes is found located on chromosome 9.

Genotype	Phenotype- Blood group
$I^A I^A$ or $I^A i$	A
$I^B I^B$ or $I^B i$	B
$I^A I^B$	AB
ii	O

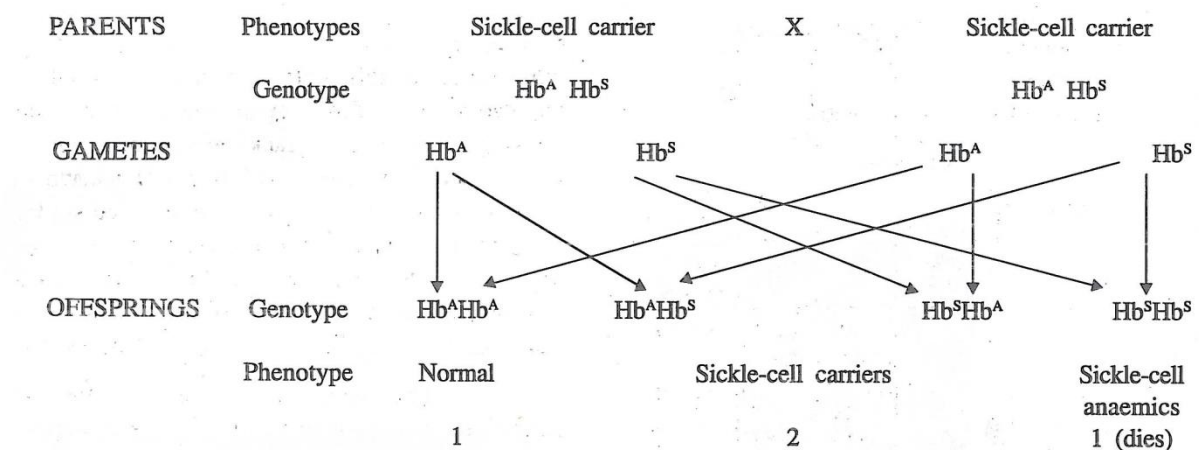
Pleiotropy

When a single gene controls two (or more) different traits, it is called a pleiotropic gene and this phenomenon is called pleiotropy or pleiotropism. The ratio is 2:1 instead of 3:1.

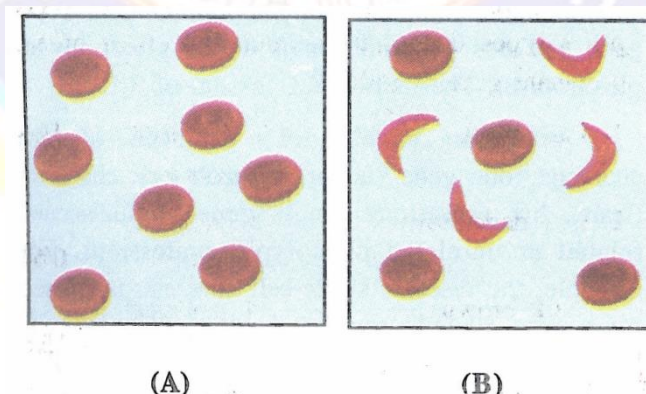
According to Mendel's principle of unit character, one gene (factor) controls one character (trait), but sometimes a single gene produces two related or unrelated phenotypic expressions. For example, the disease, sickle-cell anaemia, is caused by a codominant gene Hb^S in homozygous condition ($Hb^S Hb^S$) which leads to death, generally at least by late adolescence. In the blood of such persons, the erythrocytes become distorted, many being essentially sickle shaped. Such cells not only impede circulation by blocking capillaries, but also cannot properly perform their function of carrying oxygen. Normal or healthy gene Hb^A , is dominant. The carriers (heterozygotes- Hb^A/Hb^S) show signs of mild anaemia as their RBCs become sickle-

shaped (half-moon-shaped) in oxygen deficiency. They are said to have sickle-cell trait and are normal in normal conditions. Thus the gene for sickle-cell anaemia is lethal in homozygous condition and produces sickle-cell trait in heterozygous carrier. Two different expressions are produced by a single gene and this is example of pleiotropy. (A gene which causes death of the bearer is called lethal gene.)

A marriage between two carriers will produce normal, carriers and sickle-cell anaemic children in 1:2:1 ratio.



Sickle-cell anaemics die leaving carriers and normals in the ratio 2:1. The heterozygotes (carriers) can be identified by microscopic examination of blood. Arranged marriage's between two heterozygotes can be discouraged to avoid birth of children with fatal sickle-cell anaemia.



(A) RBCs of a normal person (B) RBCs of a carrier person in oxygen

Polygenic inheritance (Quantitative Inheritance)

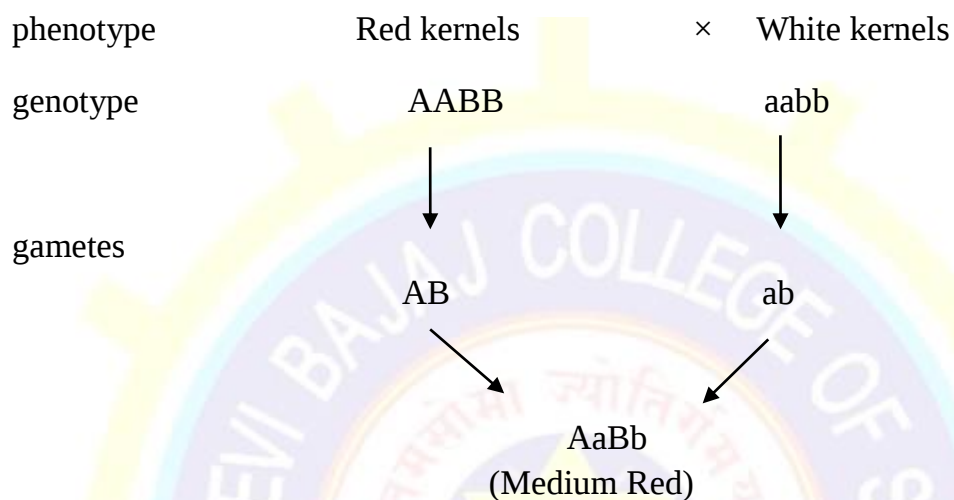
In human population, characters such as height, skin colour intelligence, susceptibility to pathological diseases and amount of flowers, fruits, seeds, milk, meat or egg produced by plants or animals show gradations (continuous variations) in expression and not only two contrasting expressions. The quantitative traits are also called **metric traits**. They do not show clear cut difference between individuals and form a spectrum of phenotypes which blend imperceptively from one type to another to cause continuous variations. These characters are determined by two or more gene pairs, and they have additive or cumulative effect. These genes are called cumulative genes or polygenes or multiple factors. The ratio is 1:4:6:4:1 if there are two genes having additive effect in wheat kernel colour and in case of three such genes (polygenes), the ratio is 1:6:15:20:15:6:1 in human skin colour.

Example 1: Wheat kernel colour: Swedish geneticist H. Nilsson-Ehle (1908) discovered the polygenic inheritance. He crossed red kernelled variety of wheat with white kernelled variety. In F_1 generation all plants had grains with intermediate colour between red and white, when F_1 are self fertilized, in F_2 generation five colour classes of kernels emerge in ratio 1:4:6:4:1. (Instead of red, intermediate and white in the ratio, 1:2:1.) Continuous variations in the expression were as follows

Sr No.	Phenotypes	Ratio
1	Darkest Red (like parent plant)	1/16
2	Medium Red (less than parent)	4/16
3	Intermediate red (as F_1 hybrids)	6/16
4	Light red (less than F_1 hybrids)	4/16
5	White (like parent)	1/16

Nilsson Ehle suggested that the kernel colour in wheat is controlled by two pairs of genes, Aa and Bb. Gene A and B determine the red colour of kernel (responsible for producing red pigment) and . are dominant over their recessive allele a and b which do not produce red colour pigment and the expression is white, if dominant gene is not present. Thus genotype of a parent with red kernels is AABB and that of parent with white kernels is aabb. Genotype of F_1 hybrids is AaBb. During gamete formation each

gene pair shows Mendelian segregation and four types of gametes are produced. Continuous variations (gradations) in expressions is observed as each dominant gene produces specific amount of pigment and the shade of red colour, (dark or light) depends on number of dominant genes present as shown in the following Punnett square. Only one, out of sixteen is with four dominant genes, and is the darkest red. Four, out of sixteen have three dominant genes and are medium red, six have two dominant genes and are intermediate red, four have only one dominant gene and are light red and again only one is without any dominant gene and is therefore with white kernel colour.



F2 generation :-

	AB	Ab	aB	ab
AB	AABB(dark red) 4dominant genes present	AABb (medium dark red) 3	AaBB (medium dark red) 3	AaBb (medium red) 2
Ab	AABb (medium dark red) 3	AAbb (medium red) 2	AaBb (medium red) 2	Aabb (light red) 1
aB	AaBB (medium dark red) 3	AaBb (medium red) 2	aaBB (medium red) 2	aaBb (light red) 1

ab	AaBb (medium red) 2	Aabb (light red) 1	aaBb (light red) 1	aabb (white) No dominant gene present
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Phenotypic ratio : 1:4:6:4:1

Human skin Colour

Davenport and Davenport (1910) studied the inheritance of skin colour in black and white populations in U.S.A. Populations derived from marriages between black and white show intermediate skin colour and are called mulattoes. When such individuals marry each other, all shades of colour (continuous variation) are observed in the population in the ratio, **1:6:15:20:15:6:1**. From this it can be concluded that skin colour in humans is controlled by three pairs of genes, **Aa, Bb, and Cc**. The presence of melanin pigment in the skin determines the skin colour. Each dominant gene is responsible for the synthesis of fixed amount of melanin. These were found to affect the character in additive fashion. and the amount of melanin synthesized is always proportional to the number of dominant genes. Thus, a true negro has six dominant genes, AABBCc, and a white has six recessive genes is aabbcc. Genotype of offspring (mulatto) is AaBbCc. A mating of two such mulattoes produces a wide variety of skin colour in the offsprings, ranging from skins as dark as the original negro parent to the white as the original white parent .There are seven different phenotypes due to the cumulative effect of each dominant gene as follows,

Sr. NO.	Phenotype	No. of dominant gene present.	Ratio
1	Pure black(negro)	6 dominant genes	1/64
2	Black (less dark than negro)	5 dominant genes	6/64
3	Lesser black or brown	4 dominant genes	15/64
4	Mulatto (intermediate- 'sanwla')	3 dominant genes	20/64
5	Fair	2 dominant genes	15/64
6	Very fair	1 dominant gene	6/64
7	Pure white (albino)	Nodominant gene	1/64

