ST. LAWRENCE HIGH SCHOOL

A JESUIT CHRISTIAN MINORITY INSTITUTION

STUDY MATERIAL -5

Class: XII **Sub: BIOLOGICAL SCIENCE** **Date: 26.06.2020**

Topic - Chapter 5 PRINCIPLES OF INHERITANCE AND VARIATIONS (PART 1)

TERMINOLOGIES IN GENETICS

1) GENE OR FACTOR: An inheritated factor determines a biological character of an organism is called gene .This is the functional unit of hereditary material. Chemically gene is a segment of DNA.
2) ALLELOMORPHS AND ALLELES: Alleles are pairs of contrasting character or alternative forms of the same gene .Each character has two determiners called factors.
3) GENE LOCUS : It is the portion or region on chromosome representing a single gene.The alles of a gene are present on the same gene locus on homologus chromosomes.
4) HOMOZYGOUS: The organism in which both the genes of a character are identical is said to be homozygous or genetical pure for that character .It gives rise to offsprings having the same character on self breeding .e.g. TT (Homozygous dominant) or tt (Homozygous recessive)
5) HETEROZYGOUS: The organism in which both the genes of a character are unlike is said to be heterozygous or hybrid .Such organisms do not breed true on self fertilization . e.g. Tt
6) GENOTYPE: The genotype is the genetic constitution of an organism . TT, Tt and tt are the genotypes of the organism with reference to these particular pairs of alleles. **Phenotype** expresses the character of individuals like form,sex ,colour and behavior etc.Phenotype of an organism is the expression or observable structural and functional traits produced due to interaction of genes and environment.
7) PURE LINE: Generation of homozygous individuals which produce offsprings of only type i.e they breed true for their phenotype and genotype .For example, tall pea plants when produced only tall plant generation on being self pollinated or cross pollinated among themselves.These form a pure line of tall pea plants.
8) MONHYBRIDS,DIHYBRID AND POLYHYBRID: When only one allelic pair is considered in cross breeding ,it is called monohybrid cross.Similarly when two allelic pairs are used for crossing Involvement of more than two allelic pairs in a cross is called polyhybrid cross.
9) RECIPROCAL CROSS: The reciprocal crosses involve two crosses concerning the same characteristics, but with reversed sexes.

10) GENOME: Total set of genes in the haploid dose of chromosomes and inherited as unit from parents to offspring is called genome.
11) GENE POOL: All the genotypes of all organisms in a population form the gene pool.

MENDEL'S LAWS OF INHERITANCE

The credit for our present understanding of the mechanism of heredity goes to an Austrian monk, Gregor Johann Mendel. He performed a large number of experiments on pea plants and deduced his observations to mathematical laws. These laws are called Mendel's laws of inheritance or Mendelism.

MENDEL'S LIFE

Gregor Johann Mendel (1822-1884) the **Father of Genetics**, was born on 22nd July, 1822 in Heizendorf, a village in Sudetan region of Silosia in Austria. His father's love for nature has influenced Mendel and he showed great interest in living beings since his childhood. He received his school education in a monastery in Bruno, and took a two-year university course in Philosophy at Olmitz Philosophical Institute. In 1843, Mendel was admitted to the Augustinian monastery at Brunn in Moravia. In 1848, he completed his theological studies and a year later took up the job of a teacher in a High School at Znaim. After a year, he joined University of Vienna and did courses in science and mathematics. In 1854, he joined Brunn Modern School as a teacher of Physics and Natural History and continued there for 14 years. During this period, he performed his famous experiments on garden pea. He presented first report of his work in 1865 before the Brunn Society for the Study of Natural Science. His original paper, *Versuche uber pflanzenhybriden* (**Experiments on plant hybridization**) was published in the **Proceedings of the Society** in 1866, Mendel's work was, however, ignored at that time. This was perhaps because of the following reasons.

- He published his work in an obscure journal.
- Scientists failed to notice his work because at that time the scientific world was busy in the controversy risen by Dawin's theory of origin of species.
- His ideas were ahead of his time as ignorance prevailed in that period about cytological basis of heredity.

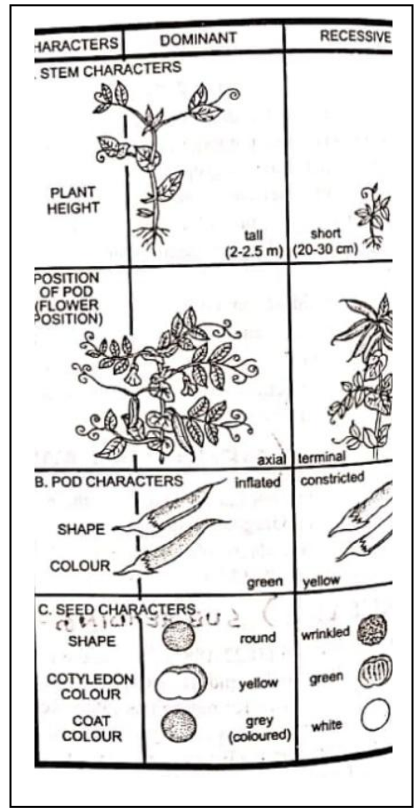
After a lapse of 34 years, in 1990, Mendel's work came to light when three other eminent biologists**, Karl Correns of Germany, Hugo de Vries of Netherlands and Erich Von Tshermak** of **Austria**, Working independently, rediscovered the Mendelian principles and they gave him the recognition he deserved. In 1902, it was suggested that Mendel's units of inheritance might be found on the chromosomes, and in the early 20th century, Thomas Morgan and his team in United States worked *Drosophila* to gain evidence for this idea. From this point onwards the study of genetics snowballed.

MENDEL'S EXPERIMENTS

Mendel carried out his experiments on the common garden pea, *Pisum sativum*, in his monastery garden. He procured seeds of 34 different varieties of peas from the local seedsman and grew them in the garden. He chose garden pea as plant material for his experiments due to the following reasons.

- Peas were available in many pure breeding varieties with observable alternative forms for a trait or characteristics.
- Pea flowers are bisexual.
- Peas are self pollinating as such the possibility of the introduction of outside genetical influences was eliminated.
- Hybrids resulting from crossing two varieties were perfectly fertile.
- Pea plants produce new generation in a reasonable short time and a single plant produces numerous seeds.

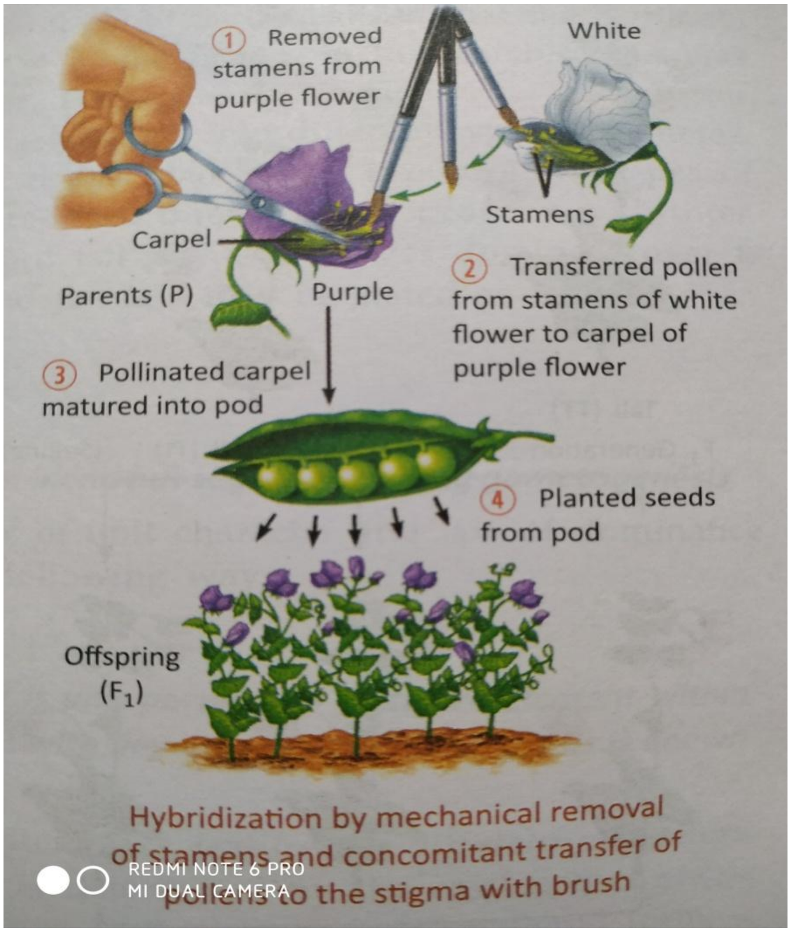
The varieties of garden pea have a large number of contrasting characters. But, for the purpose of his breeding experiments Mendel took into consideration seven characters, each with two alternatives. The seven characters (traints) and their contrasting alternatives considered by Mendel are shown.



CROSSING TECHNIQUE

Since, garden pea (*Pisum sativum*) is self-pollinating, anthers from the flower were removed before maturity. This operation of removal of anthers is called emasculation. The stigma was protected against any foreign pollen with the help of a muslin bag. Pollens from dehisced anthers of the plant to be used as a male parent were brought and dusted on the feathery stigma of the emasculated flower. At the time of pollination, it was assured that the pollen is mature and the stigma receptive.

For each of the seven pairs of characters, plants with one trait were used as female and the other as male. The plants of unlike characters with which hybridization is first made constitute parent generation (P_1 generation). The population obtained as the result of crossing the plants with contrasting characters is called the **first filial generation** or F_1 (filial = progeny). The progeny of F_1 plants obtained due to self fertilization represents the second filial generation or F_2. The subsequent generations are known as F_3, F_4, F_5 and so on. Mendel first crossed two plants which differed constantly in one or several characters. The seeds obtained after crossing were used to produce offsprings. The plants obtained from the crossing of two individuals (differing in atleast one set of characters) are known as **hybrids** and the process is called **hybridization**. For example, if a red flowered plant is crossed with a white flowered plant, the seed produced after cross-fertilization is a hybrid and the process of producing hybrid is hybridization.



MENDEL'S LAWS

Based on the observations of his experiments on garden pea, Mendel drew some important conclusions. These conclusions are regarded as Mendel's Laws of Inheritance. These laws are as follows.

1)Law of dominance: Mendel's first experiments were with the varieties of garden pea that differed in only one visible character. These are known as monohybrid experiments.

When Mendel crossed true breeding red flowered peas with true breeding white flowered peas, the first progenies (F_1 generation) formed produced only red flowers.

In offspring, white colour of the flower suppressed and red colour dominated. Mendel, therefore, called such traits as red colour of flowers dominant and white colour recessive. He observed this fact between all the seven pairs of characters selected by him. The pair of contrasting characters is called **allelomorphic** pair or allelic pair (a term coined by **Bateson** in 1906), and each member of the pair can be regarded allele of the other. Thus, redness and whiteness of the flower are alleles to each other.

The law of dominance therefore states that, out of a pair of allelomorphic characters one is dominant and the other recessive.

The hereditary units which are responsible for the appearance of characters in the offsprings were said to be factors or determiners. Later, W. Johannsen (1909) called them **genes**.

2)**Law of segregation (Law of purity of gametes)**:- When pure breeding red and white flowered varieties were crossed, they formed red flowered individuals only in F_1 generation. But, after self-fertilization of F_1 generation plants (red), the individuals of F_2 generation thus formed were red flowered and white flowered in the **ratio 3:1.** This ratio is known as **monohybrid ratio**. Thus, the two contrasting characters have segregated or separated in F_2 generation.

This is known as law of segregation, which states that when a pair of allelomorphs are brought together in the hybrid (F_1), they remain together in the hybrid without blending but separate complete and pure during gamete formation. This law is also known as Mendel's second law of purity of gametes. It is sometimes said to be the law of the splitting of hybrids.

The law of segregation can be explained as follows : pure breeding red (RR) and white (rr) varieties when crossed in F_1 generation form hybrid red (Rr). During the gamete formation segregation takes place and only one factor of the pair of alleles alone enters one gamete and the other enters to the other gamete. Thus, a gamete can be only either R or r. The F_1 hybrid (Rr) in the above cross produces two types of gametes R and r. After self-pollination they result in RR (red), Rr (Red), Rr (red) and rr (white) plants in F_2 generation. Each parent thus process two types of gametes which have equal chances of combination together.

The law of segregation is Mendel's most important contribution to biology because it introduced the concept of hereditary factors as discrete, physical entities that do not become blended or altered when present together in the same individual. He disproved the blending theory by showing that although traits caused by recessive alleles disappear in the F_1 generation, they reappear unchanged in the F_2 generation

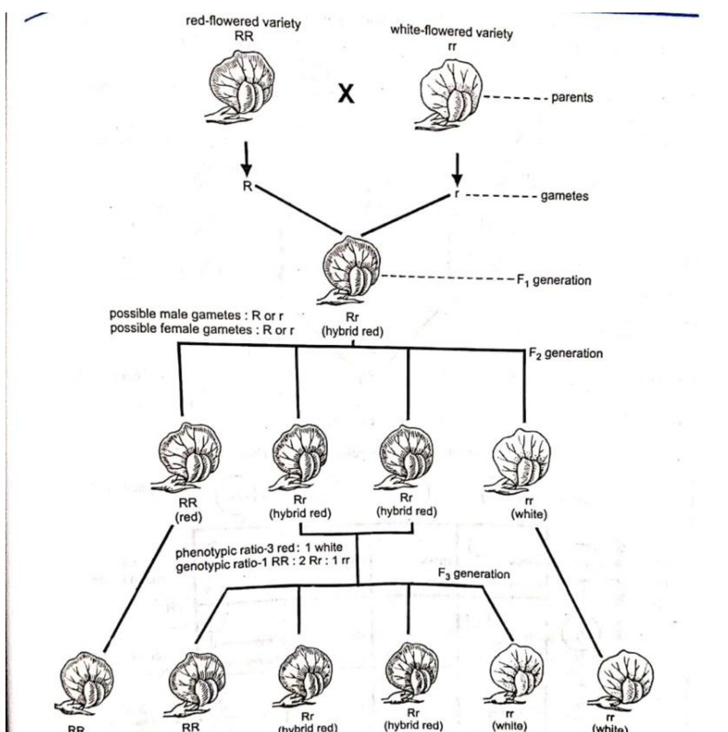


Fig. 6. Monohybrid cross to explain law of segregation.

Limitations of the law of segregation. The law of segregation applies only to diploid organisms that form haploid gametes to reproduce sexually. It further applies only to traits controlled exclusively by a single gene pair in which of the two alleles one is dominant over the other. The law therefore does not apply to

- Alleles that are incompletely dominant or codominant.
- Genes that collaborate or vary in their expressivity or penetrance.
- Genes that are pleiotropic (i.e., each gene having two or more phenotypic effects), complementary, or influenced by epistasis (suppression of one gene's effects by another).
- Traits caused by many gene pairs.

3)Law of Independent assortment :- Mendel investigated not only those crosses in which the parent differed in single pair of characters, but also others in which the parent differed in two pairs. Such a cross which involves two pairs of contrasting characters simultaneously is called **dihybrid cross**.

For a dihybrid experiment, Mendel crossed two parent pea plants, one of which was homozygous for round and yellow seed (RRYY) and the other for wrinkled and green seed (rryy). Round shape and yellow colour of the seed is dominant over the wrinkled shape and green colour. During gamete formation, each pair of alleles segregates independently of each other and produces only RY or ry gametes. All the plants produced in the F_1 generation were with round and yellow seeds (RrYy), which were **heterozygous** for both the alleles and are thus called dihybrid.

In F_2 generaiton, the F_1 hybrid produces four kinds of gametes in equal numbers : RY, Ry, rY and ry. As there are four kinds of pollen grains and four kinds of egg cells, there are chances of 16 possible types. The union of gametes in fertilization is random thus resulting in nine types of genotypes and four types of phenotype.

The plants with round yellow and wrinkled green seeds are parental combinations, but round green and wrinkled yellow are new combinations (called recombinants). The ratio of F2 generation 9:3:3:1 is called dihybrid ratio. Thus, each pair of alleles segregates strictly independent of each other, and this demonstrates the law of independent assortment.

Mendel's independent assortment can be defined as that when two pairs of independent alleles are brought together in the hybrid F1, they show independent dominant effects. In the formation of gametes the law of segregation operates, but the factors assort themselves independently at random and freely. It is immaterial whether both the dominant characters enter the hybrid from the same or two different parent but the segregation and assortment remain the same.

Limitations of law of independent assortment. The law of independent assortment applies only to gene pairs on different pairs of homologous chromosomes – one gene pair on one pair of homologous chromosomes and the other gene pair on another pair of homologous chromosomes. Although Mendel was not aware of chromosomes, all the traits he studied were on different pairs of homologous chromosomes. That is why he could conclude that gene pairs segregate independently of one another.

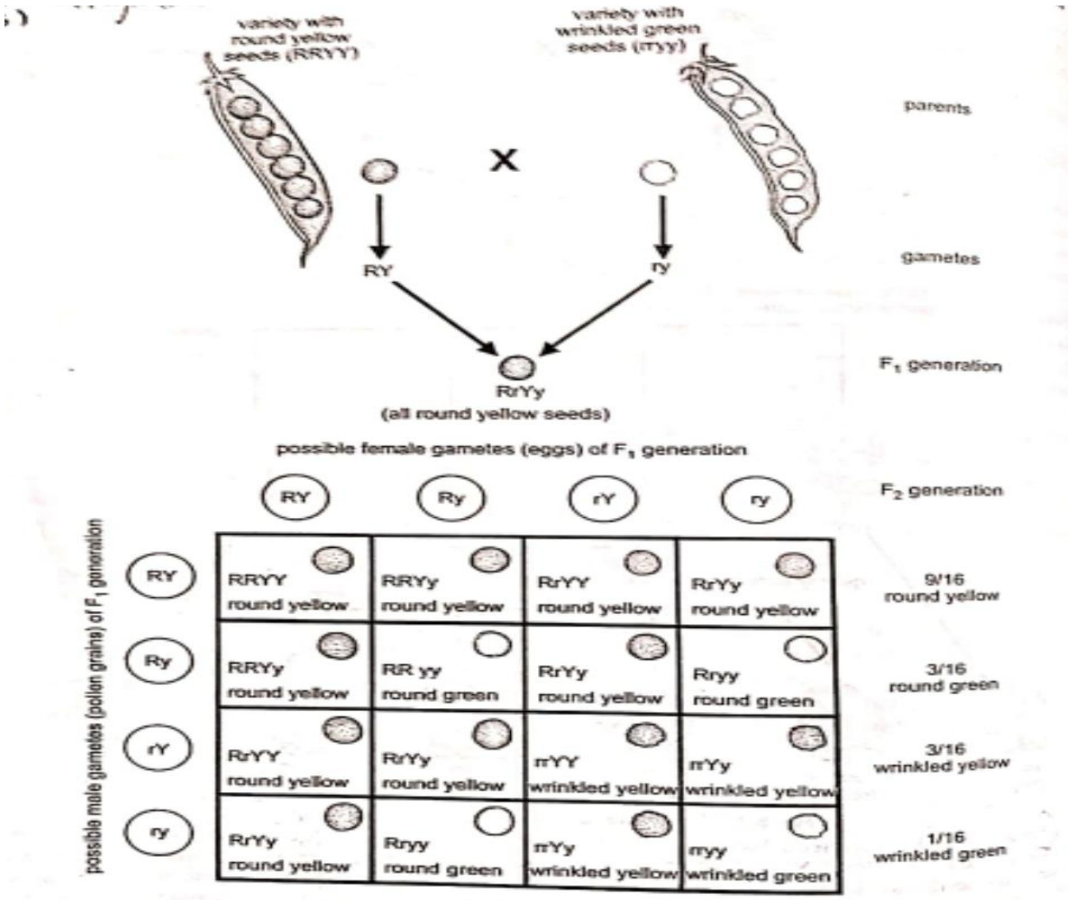


Fig. 7. Dihybrid cross to explain law of independent assortment.

(Round yellow	:	Round green	:	Wrinkled yellow	:	Wrinkled green)
9/16	:	3/16	:	3/16	:	1/16

Reasons of Mendel's Success

The following are the main reasons for the success of Mendel's experiments.

- His choice of pea plants for his breeding experiments was excellent.
- He kept complete records of every cross and applied statistical methods and laws of probability for computing his results. He thus could know the pedigree of the progenies.
- He took one or two traits at one time for his experiments. His predecessors usually studies many traits simultaneously that caused confusion and made matters complicated.
- He was fortunate enough that the characters he chose for his experiments did not show linkage, incomplete dominance, gene interactions, etc.
- He took utmost care to check contamination from foreign pollen at the time of hybridization.

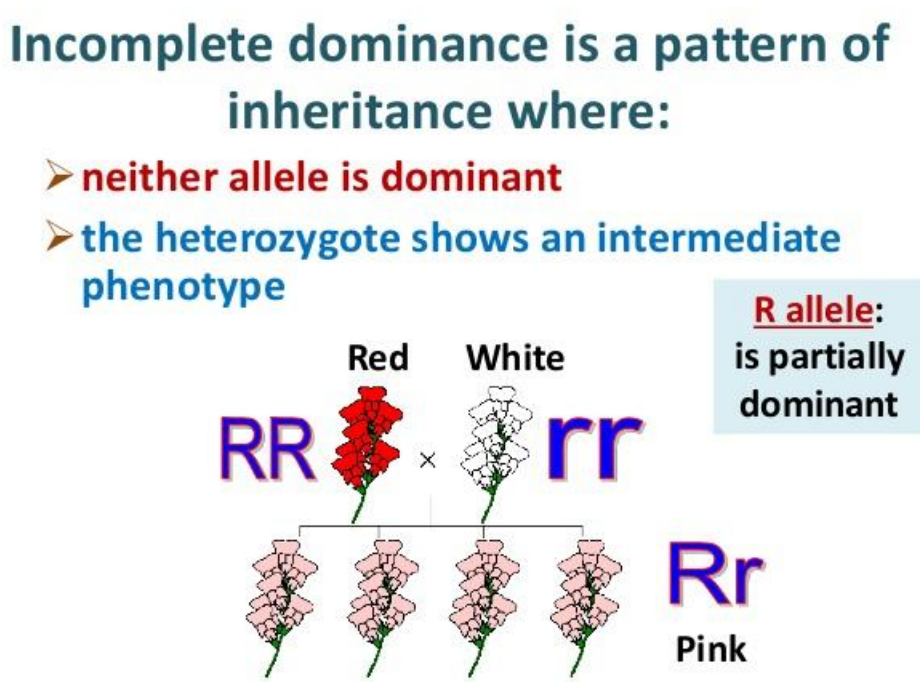
OTHER PATTERNS OF INHERITANCE AND GENOTYPIC EXPRESSION OR NON MENDELISM

INTRODUCTION

Mendel explained inheritance in terms of discrete hereditary determiners (factors), now called **genes**. What was shown by him in pea was later shown to be generally true for a variety of traits in many plants and animals. But all patterns of inheritance could not be explained exclusively on the basis of Mendel's original principle s alone and complexities were observed by subsequent workers.

A) INCOMPLETE DOMINANCE

Mendel's law of dominance states that one allelomorph dominates over the other and as such F_1 exhibits one of the two alternative phenotypes present in parents. However, there are some characters which are exceptions to this rule and show incomplete or partial dominance. In such cases, dominance is absent and the hybrid individuals resemble neither parent and are **intermediate** between the two parents. This phenomenon is known as **incomplete dominance**. For example, the F_1 progeny from a cross between red and white flowered four O'clock plant (*Mirabilis jalapa*) and in *Lathyrus odoratus* bear pink flowers. In the F_2 generation, red, pink and white flowered plants occur in 1:2:1 ratio.



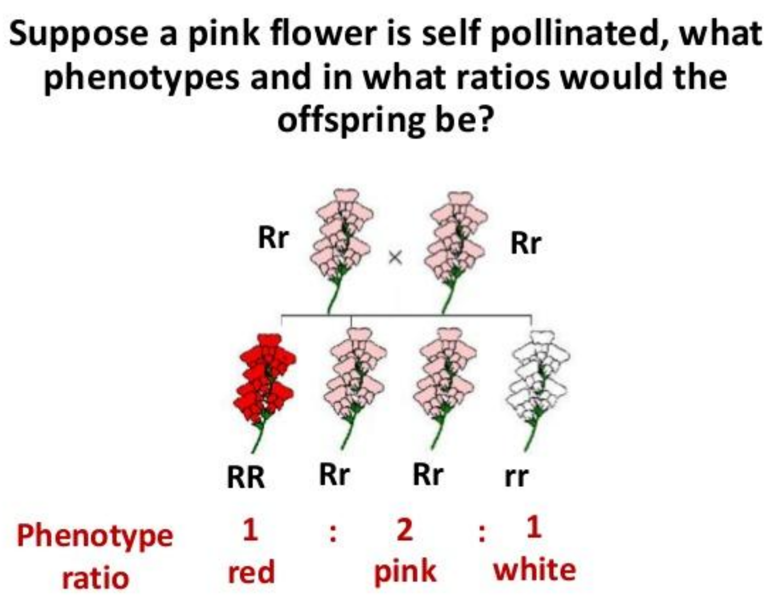
RR (red) x rr (white) --------- Parents

R r ----------------gametes

Rr (pink) ------------------- F_1 Generation

1 RR (Red) : 2 Rr (Pink) : 1 rr (white) ------------- F_2 Generation

Incomplete dominance in *Lathyrus odoratus*



Examples of incomplete dominance are also met within the animal kingdom. In the blue Andalusian fowl, the F_1 progeny of a black and white cross has a mixture of black and white in their feathers, i.e. they were slate-blue in appearance. In the F_2 generation three kinds of offsprings are produced in the ratio of 1:2:1, i.e. 1 black, 2 state blue and 1 white. The black and white breed true, while the slate blues on inbreeding invariably produce three kinds like their F_1 ancestors. As a matter fo fact, they are heterozygous and never breed true.

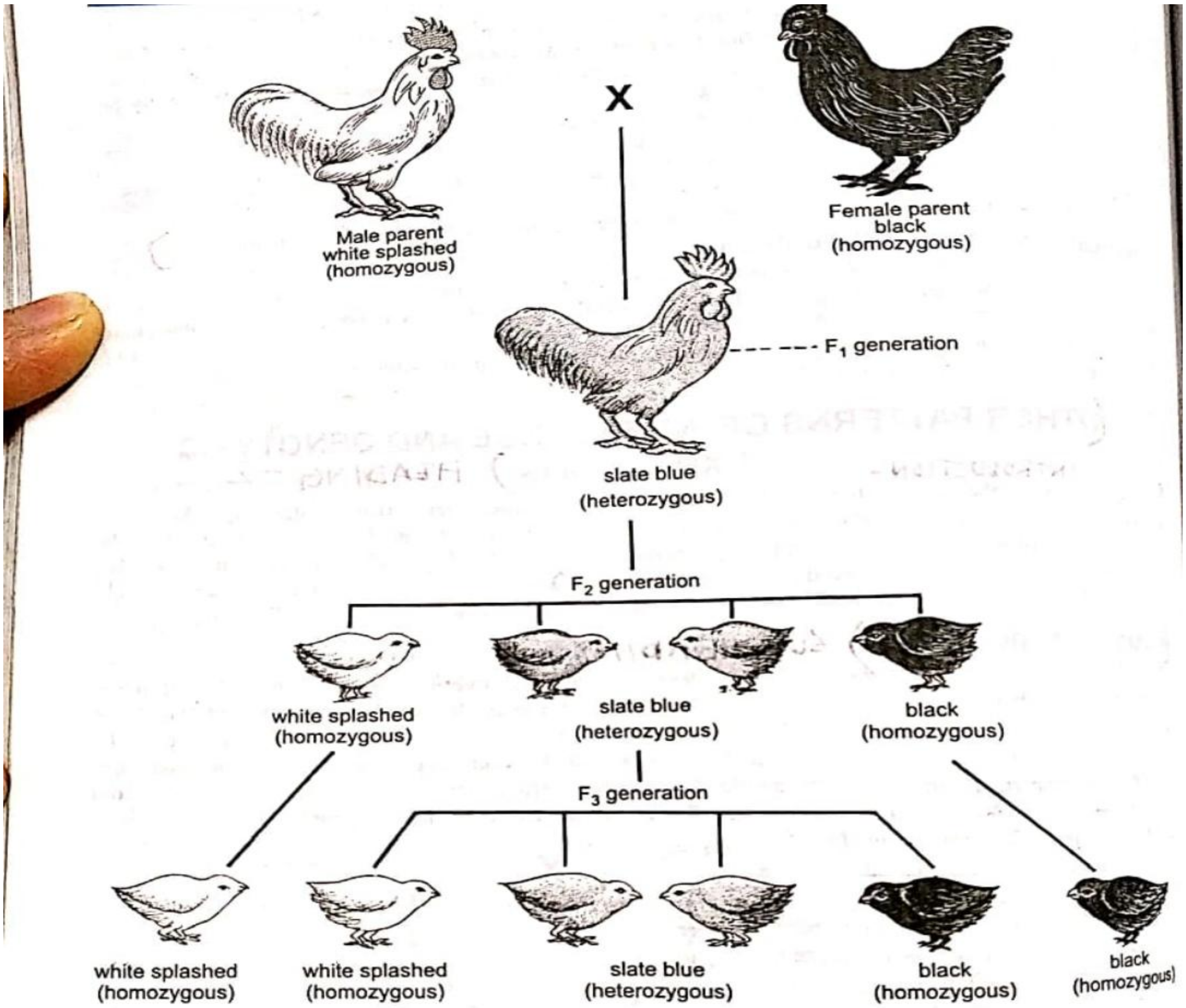
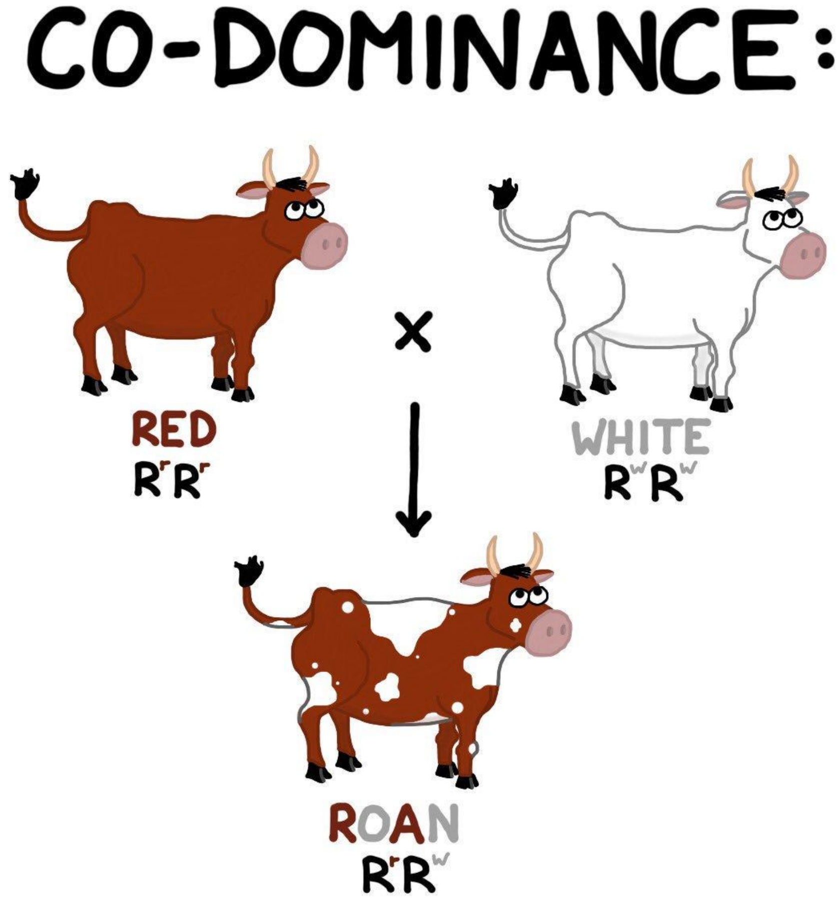


Fig. 11. Incomplete dominance in Andalusian fowl.

The phenomenon of incomplete dominance can be explained on the basis of Mendelian segregation. In case of complete dominance, the recessive factor (gene) fails to show its effect or is entirely ineffective in the presence of the dominant factor. But in incomplete dominance, both alleles have almost equal effect on the phenotype and this results in an **intermediate** character of the hybrid.

B) CO-DOMINANCE

In co-dominance, both allelic genes of a genetic trait are equally expressed, i.e. the dominant character not able to suppress the recessive character and thus both the characters appear side by side in F1 hybrids. For example, if a cattle with black coat is crossed to a cattle with white coat, the F1 hybrids possess neither black or white coat colour, but have roan coat colour, where black and white patches appear separately. Thus, the alleles which are able to express themselves **independently** when present together are called co-dominant alleles.



Incomplete dominance	Codominance
1. When none of the two alleles is dominant, but these alleles mix up to form a new trait, then it is termed as incomplete dominance.	1. When both the alleles are dominant in nature, and the traits for both the alleles are expressed equally, then its is termed as codominance.
2. Although both the alleles mix up, but only one allele's effect is seen.	2. In codominance, both the alleles mix up equally and their effects are also seen equally.
3. Incomplete dominance always lead to the formation of a new phenotype.	3. In codominance, no new phenotype is formed.
4. Examples are snapdragon and mirabilis Jalapa.	4. Examples are Roan character is cattle, and blood groups in human.

CO DOMINANCE IN HUMAN BLOOD GROUPS:

Whether a person has type A, type B, type AB or type O blood depends on the presence or absence of specific substances on the red blood cells. There are two of these substances – antigens A and B. Thus a person with antigen A is considered to be with blood group A, a person who is with blood group O has neither of the antigens and a person with antigen B, blood group is called B. If both the antigens A and B are present the blood group is called AB. The blood group character is controlled by set of three alleles. The gene which produces antigen A is denoted by I^A the gene for antigen B by I^B and gene for absence for both the antigens by i. Genes I^A and I^B are both dominant over gene I^O or i, but not over each other. Any person carries two of these alleles, one from each parent. I^A determines the formation of glycoprotein A and I^B determines the formation of glycoprotein B. The dominant relationship of three alleles are interesting. Both I^A and I^B are fully expressed in the presence of the other. Both I^A and I^B are absent (i.e. with genotype ii) person is with blood group O. Since a person with genotype I^A I^B produces both the glycoproteins alleles I^A or I^B are said to be **co-dominant**.

Phenotype	Genotype	Antigen
Type A	I^AI^A or I^Ai	A
Type B	I^BI^B or I^Bi	B
Type AB	I^AI^B	A and B
Type O	Ii	none

Blood groups are inherited in a simple Mendel's pattern. Children with all the four type of blood groups are possible in a cross between two persons heterozygote for blood groups A and B.