Principles of Inheritance and Variation

Genetics: It is the branch of biology which deals with inheritance and variations of characters from parents to offspring.

Inheritance: It is the process of passing characters from parent to progeny. It is the basis of heredity.

Variations: It is the degree by which progeny differs from parents. Variations can be in terms of physiology, morphology and behavioral characteristics of individuals belonging to the same species.

Mendel' s Law Of Inheritance: Mendel conducted hybridization experiments on garden peas for seven years (1856-1863). On the basis of these experiments he proposed the laws of inheritance.

- He selected the characters that have two opposing traits and concluded his hybridization experiments on 14 true-breeding pea plant varieties.

S.No.	Characters	Contrasting Traits
1.	*Stem height*	*Tall/dwarf*
2.	*Flower colour*	*Violet/white*
3.	*Flower position*	*Axial/terminal*
4.	*Pod shape*	*Inflated/constricted*
5.	*Pod colour*	*Green/yellow*
6.	*Seed shape*	*Round/wrinkled*
7.	*Seed colour*	*Yellow/green*

Reasons for selecting garden pea plant:

- Easily available on a large scale.

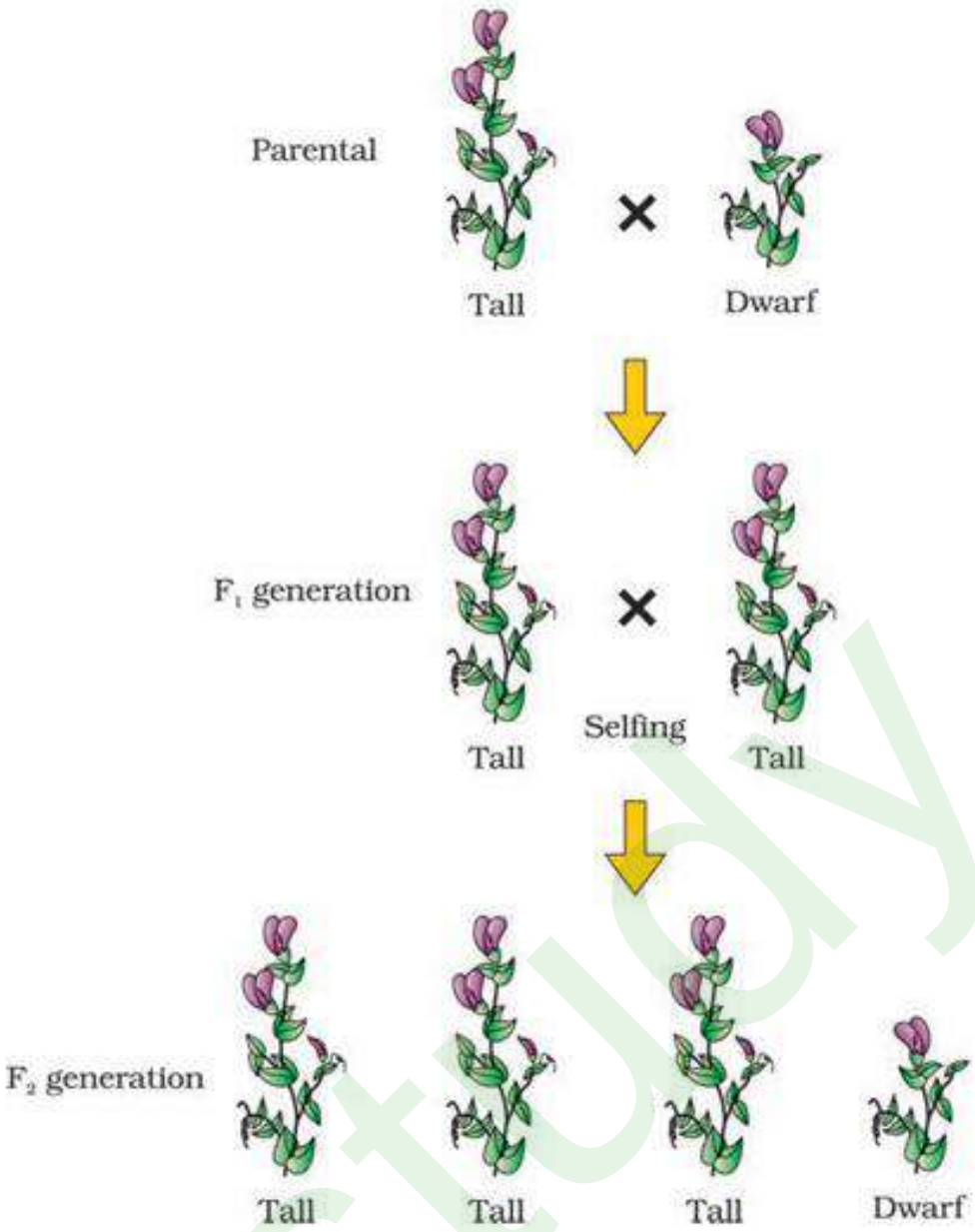
- There are many varieties with distinct characteristics.
- They are self-pollinated and can be cross-pollinated easily.
- They have a short life cycle.

Reason for success of Mendel:

- He studied one character at a time.
- He used available techniques to avoid cross pollination by undesirable pollen grains.
- He applied mathematics and statistics to analyze the results obtained from him.
- Mendel selected seven contrasting characters for the experiment.

Inheritance of one Gene (Monohybrid Cross):-

- Mendel crossed tall and dwarf pea plants and collected the seeds from them. Seeds were used to generate plants of first generation (F1 or Filial progeny).
- Mendel observed that all the first generation plants were tall, none of them were dwarfs. He concluded that the F1 generation resembled either one of the parents.
- He then self-pollinated the tall F1 plants and he observed that some of them were dwarfs.
- In F_2 generation, both the traits were expressed in proportion of 3:1. Dominant trait in F_2 is about thrice of the recessive from.
- Based on these observations, he concluded that something was being stably passed from one generation to the other. He named it ' factors' which are now called as ' genes' .



➢ **Gene:** It is the unit of inheritance. It contains information that is required to express a particular trait in an organism. Genes which code for a pair of contrasting traits are known as ' alleles' . They are slightly different for the same gene.

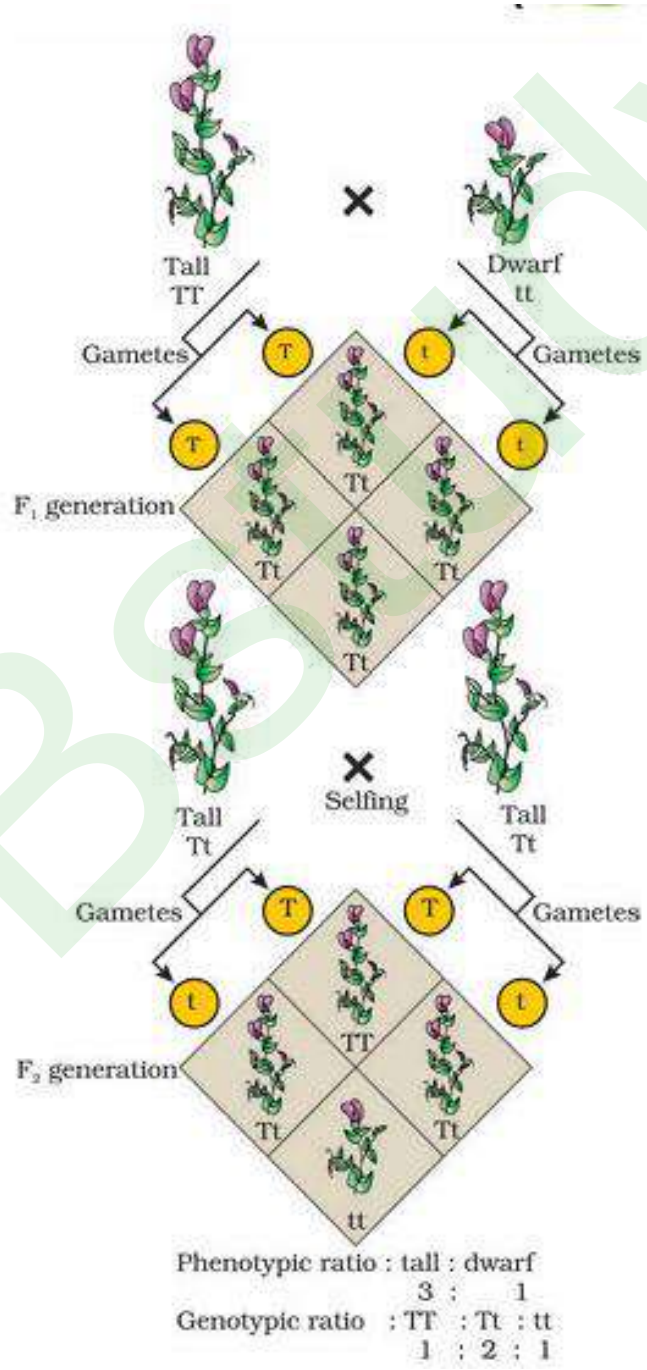
• For representing traits, capital letter is used for the trait expressed at F1 generation and small letter is used for the other one. For example: T for tall trait, t for dwarf.

• T and t are alleles of each other. Pair of alleles for height in the plants are TT, Tt and tt.

• TT and tt are homozygous called genotypes of the plant while the description terms tall and dwarf are phenotype & It represents heterozygous.

• Test cross is the cross between an individual with dominant trait and a recessive organism. It helps us to understand whether the dominant trait is homozygous or heterozygous.

• Punnett square is a graphical representation used to calculate probability of all possible genotypes of offspring in a genetic cross.



Law of Dominance: The dominant allele masks the effect of recessive allele. It explains the expression of only one of the

parental characters in a monohybrid cross in F_1 and expression of both in F_2.

- Characters are controlled by discrete units called factors.
- Factors occur in pairs.
- In a dissimilar pair of factors one member of the pair dominates the other.

For example: the allele of tallness (T) is dominant over the allele of dwarf (t).

Law of Segregation: It states that every individual possess two alleles of a gene and these alleles segregate from each other during gamete formation (at the time of meiosis).

- Alleles do not blend and both the characters are recovered during gamete formation in F2 generation.
- Homozygous individuals produce one type of gametes while heterozygous individuals produce two types of gametes each having one allele with equal proportion.

1) **Incomplete Dominance:** When the experiments were repeated on other traits on other plants, sometimes it was found that F1 progeny does not resemble either of the parent, it was a mixture of two.

- Genotypic ratio was the same as we would expect in Mendelian monohybrid cross but the phenotypic ratio is changed.

For example: Snapdragon or Antirrhinum sp. or dog flower – inheritance of flower colour.

2) **Co- Dominance:** The two alleles are able to express themselves independently when present together.

- I^A, I^B are dominant over i. If I^A and I are present, only I^A expresses. I^A and I^B are present; both of them express each other.

For example: ABO blood grouping in humans is controlled by gene I. It has three alleles IA, IB and i.

ABO blood grouping is also a good example of multiple alleles.

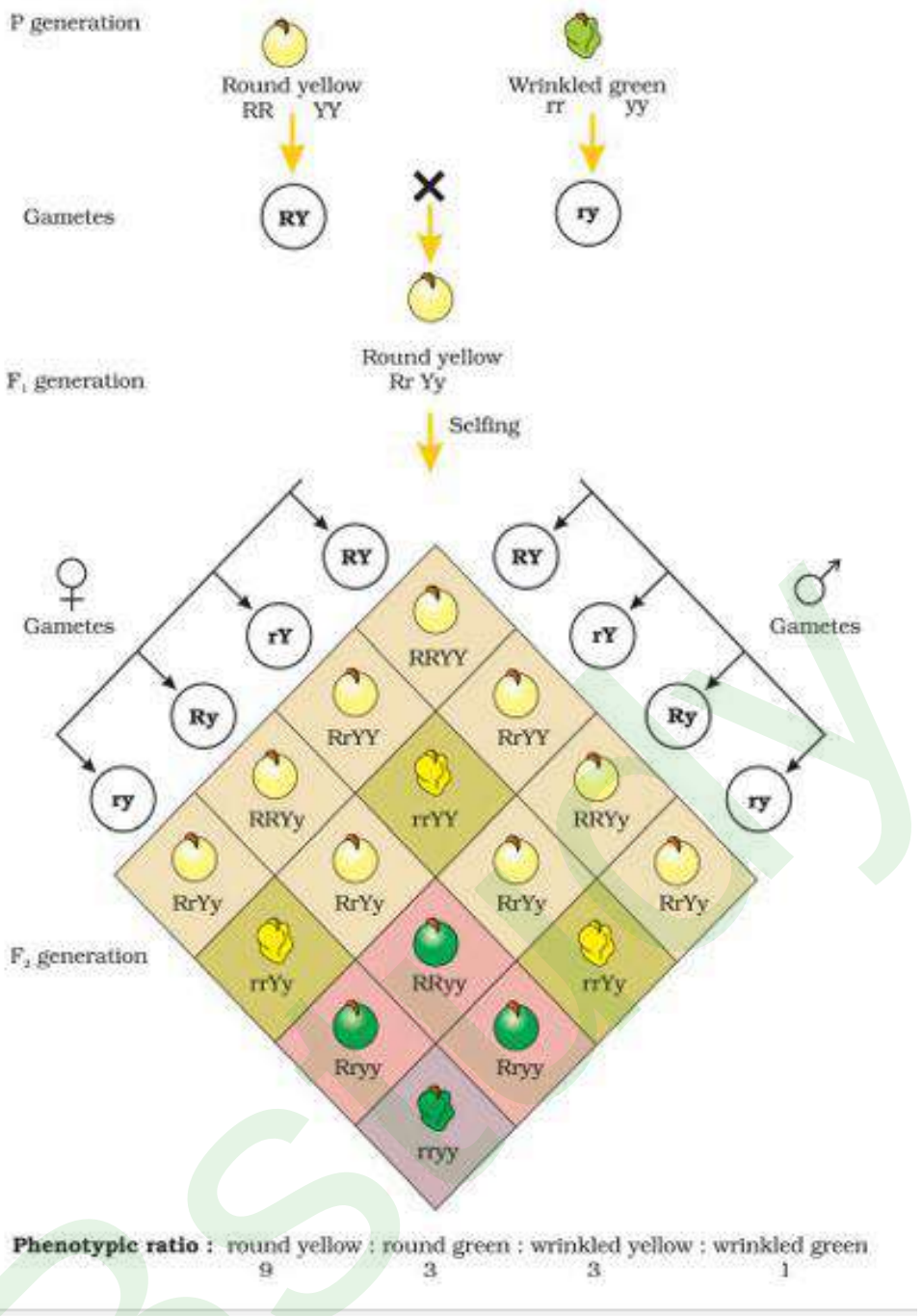
Inheritance of two Gene: Mendel also worked with two characters on pea plants. He chose the color and shape of the seed to explain the inheritance of two genes.

Y - dominant yellow color

y - recessive green color

R - round shape of the seed

r - wrinkled shape of the seed



Phenotypic Ratio– Round yellow : round green : wrinkled yellow : wrinkled green

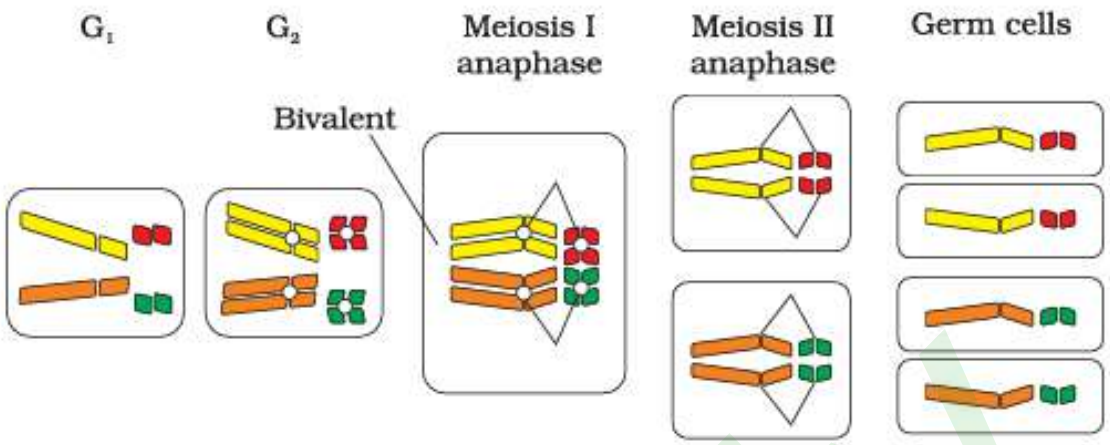
9 : 3 : 3 : 1

Chromosomal Theory of Inheritance: According to this theory–

- Genes are located at specific locations on the chromosomes.
- Chromosomes as well as gene both occur in pairs.
- Homologous chromosomes separate during meiosis.

• Fertilization restores chromosome number to diploid condition.

• Chromosomes segregate as well as assort independently.



Linkage And Recombination: Morgan carried out several dihybrid crosses in Drosophila to study genes that were sex-linked.

• Morgan hybridized yellow-bodied, white-eyed females to brown-bodied, red-eyed males and intercrossed their F1 progeny.

• According to him, two genes did not segregate independent of each other and F2 ratio deviated from 9:3:3:1. This concluded that genes are linked. This process is called linkage.

• Recombination is the rearrangement of genetic material. The generation of non-parental gene combination during dihybrid cross is called recombination.

• When genes are located on same chromosome, they are tightly linked and show less linkage. This is responsible for variation.

Sex Determination: Different organisms have different types of sex determination.

• In 1891, Henking observed specific nuclear structure is located on 50 per cent of sperms only. The discovered X-body but was unable to explain its significance.

• In insects, XO type of sex determination is present. All the eggs have an additional X- chromosome besides the autosomes. Some sperms bear X- chromosome where as some do not.

• Eggs fertilized by sperm having having X- chromosome become females and those fertilized by sperms that do not have an X- chromosome becomes males.

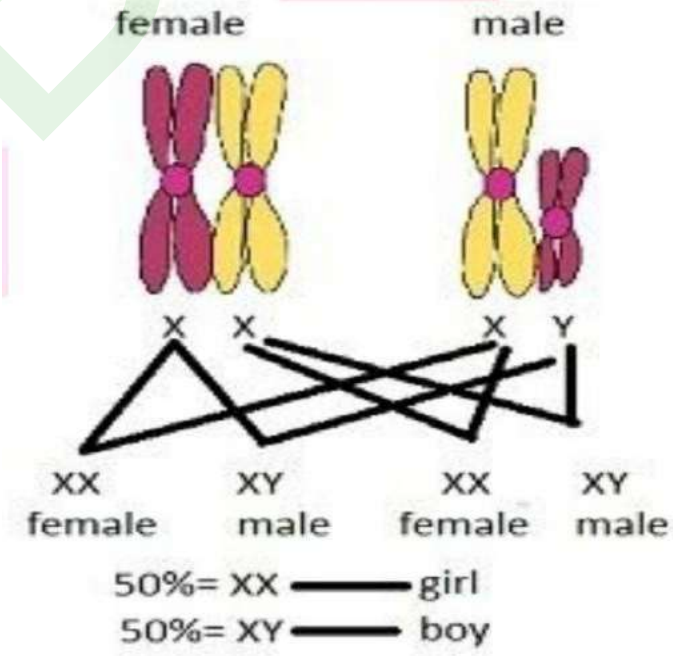
• For example: grasshopper (males have only one X-chromosome besides autosomes and females have a pair of X- chromosomes)

Sex Determination In Humans: XY type of sex determination seen in humans.

• Out of 23 pairs chromosomes present, 22 chromosomes are exactly same in both male and female. These are autosomes.

• Males have (XY) and Females have (XX)chromosomes.

• Drosophila also has XY type of sex determination.



Sex Determination In Birds: ZW type of sex determination is seen in birds.

- Females have ZW and males have ZZ chromosomes.
- In birds sex is determined by type of ovum.
- In birds, females are heterogametic.

Mutation: Mutation is any change in DNA sequence.

- It is a heritable change.
- Mutations can affect genotype as well as phenotype.
- It also leads to variations.

- **Types of mutations:-**

1) **Point Mutations:** Mutations that occur due to change in a single base pair of DNA is called as point mutations. For example: sickle cell anemia

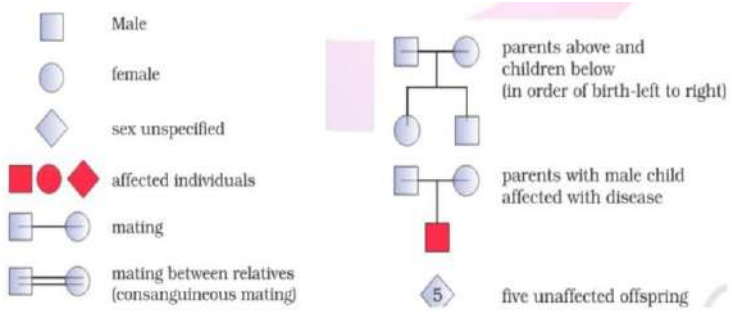
2) **frame- shift mutations:** When there is deletion or insertion of base pairs of DNA, it causes frame- shift mutations.

- Mutagens are the chemical and physical factors that induce mutations. UV rays can also cause mutations.

Genetic Disorders:-

Pedigree Analysis: Analysis of traits in several generations of family is called pedigree analysis.

- Inheritance of a particular trait is represented in the family tree over generations.
- Symbols used in pedigree analysis



Mendelian Disorders: Most common and prevalent Mendelian disorders are Haemophilia, Cystic fibrosis, Sickle-cell anaemia, Colour blindness, Phenylketonuria, Thalassemia, etc.

1) **Haemophilia:** sex-linked recessive disease.

- A single protein that is a part of the cascade of proteins involved in the clotting of blood is affected.
- In affected individual, a simple cut will result in non-stop bleeding.
- Heterozygous female (carrier) can transmit the disease to son.
- Possibility of female becoming a haemophilic is extremely rare.

2) **Sickle-Cell Anaemia:** Autosome linked recessive trait.

- It can be transmitted from parents to the offspring when both the parents are carrier for the gene.
- Disease is controlled by a single pair of allele, HbA and HbS.
- Hb^5Hb^5 homozygous shows the diseased phenotype.
- Heterozygous individuals Hb^5Hb^5 show normal phenotype but they are carrier of the disease.

• The defect is caused due to substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule. It results from single base substitution from GAG to GUG at sixth codon of the beta globin.

• Due to this, mutant haemoglobin is formed. It undergoes polymerization under low oxygen tension causing the change in the shape of the RBCs from biconcave to elongated sickle-like structure.

3) **Phenylketonuria:** Inborn error of metabolism, autosomal recessive trait.

• Affected individual lacks an enzyme that converts the amino acid phenylalanine into tyrosine.

• Due to which, phenylalanine gets accumulated and converted into phenylpyruvic acid and other derivatives.

• This causes mental retardation.

Chromosomal Disorders: It is caused due to absence or excess or abnormal arrangement of one or more chromosomes.

• **Aneuploidy** – failure of segregation of chromatids during cell division cycle results in the gain or loss of chromosome.

• **Polyploidy** – Failure of cytokinesis after telophase stage of cell division results in an increase in a whole set of chromosomes in an organism.

Down's syndrome – gain of extra copy of chromosome 21 (trisomy 21)

• It was first described by Langdon Down (1866).

- Affected individual is short with small round head, furrowed tongue and partially open mouth.
- Broad palm with characteristic palm crease.
- Physical, psychomotor and mental development is retarded.

Klinefelter's syndrome – presence of an additional copy of X– chromosome resulting into karyotype, 47, XXY.
- Sterile individuals are sterile.

Turner's syndrome – loss of an X– chromosome in human females i.e. 45 with XO
- Such females are sterile as ovaries are rudimentary.