

Principles of Inheritance and Variation

- Genetics: It is the branch of science that deals with the principles of inheritance and its practices.
- Mendel was the first to carry out the study on the transmission of characteristics from parents to offspring.
- Mendel proposed that heredity is controlled by genes.
- **'s law of inheritance**
- Mendel experimented on garden pea plant (*Pisum sativum*) having many visible contrasting characters.
- He used **seven** contrasting pairs of characters or traits in garden pea.

Trait	Dominant trait	Recessive trait
Seed shape	Round	Wrinkled
Seed colour	Yellow	Green
Flower colour	Violet	White
Pod shape	Full	Constricted
Flower position	Axial	Terminal
Stem height	Tall	Dwarf
Pod colour	Green	Yellow

- Mendel crossed pea plants having these seven pairs of contrasting characters/traits and produced offspring from them.
- **His experiments included three steps -**
 - Selection of true breeding plants
 - Obtaining F₁ plants by cross pollination
 - Self pollination of F₁ plants to obtain F₂ generation
- **Monohybrid cross:** It is a cross between two parents that have one pair of contrasting characters; for example, a cross between tall (TT or Tt) and dwarf (tt) plants.
- The **phenotypic ratio** obtained in monohybrid cross is 3:1 while **genotypic ratio** is 1:2:1.
- Based on observations on monohybrid crosses, two laws were proposed -

- **First law or law of dominance:** It states that only one parental trait gets expressed in the F₁ generation while both the traits get expressed in the F₂ generation.
- **Law of segregation:** It states that two alleles segregate from each other when characters are transferred from parents to offspring during reproduction.
- **Test cross:** It is a cross between organisms with unknown genotype and recessive parents. This cross is used for determining whether the given individual has homozygous or heterozygous genotype.

Incomplete dominance

- It is the phenomenon where one allele is incompletely dominant over the other member of the allelic pair.
- Both **phenotypic and genotypic ratios** are the same in the case of incomplete dominance, i.e., 1:2:1.
- **Dominant:** It is the character/trait that is able to express itself over another contrasting trait; for example, tall plant is dominant over dwarf plant.
- **Recessive:** It is the character/trait that is unable to express itself over another contrasting trait.

Co-dominance

- It is the phenomenon where both the alleles of a gene are equally dominant and get expressed together in heterozygous condition; for example, ABO blood group in humans.
- Blood group ABO is an example of multiple alleles.
- **Dihybrid cross:** It is the cross between two parents that have two pairs of contrasting characters; for example, the cross between **round yellow seed** and **wrinkled green seeds**.
- The **phenotypic ratio** obtained in dihybrid cross is **9:3:3:1**.
- On the basis of observation of dihybrid cross, the law of independent assortment was proposed.
- **Law of independent assortment:** It states that the members of different pairs of alleles assort independently into gametes.

Chromosomal theory of inheritance

- It was proposed by Sutton and Boveri.
- Mendel's law was extended as chromosomal theory of inheritance after it was known that genes are located on the chromosomes.

Polygenic Inheritance

- In polygenic inheritance is also known as quantitative inheritance.
- The expression of quantitative traits is controlled by more than one pair of genes and the environment also contributes towards such type of inheritance.
- The most common example of polygenic inheritance is observed in the inheritance of skin colour in human.
- It was first studied by C.B. Davenport (1913).
- **Morgan** worked on *Drosophila* and found that genes are linked.
- **Linkage:** It is the coexistence of two or more genes on the same chromosome. If the genes lie together, they are inherited together and are said to be linked genes.
- **Recombination:** It is the mixing of the maternal and paternal characters in a sexually reproducing organism so as to bring genetic variation in the offspring.
- **Sex determination**
 - **Female heterogamy:** Presence of two kinds of sex chromosomes in the female; only one kind is present in the male; for example, birds (the female has ZW sex chromosome while the male has ZZ sex chromosome).
 - **Male heterogamy:** Presence of two kinds of sex chromosomes in the male; only one kind is present in the female; for example, humans, *Drosophila* (the female has XX sex chromosome while the male has XY sex chromosome).
 - **In humans**, the genetic make up of the sperm determines the sex of the baby.
- **Sex determination in honey bees**
 - Show a special mechanism of sex determination called the haplo-diploidy.
 - Unfertilized eggs develop into males.
 - Fertilized eggs develop into females.
- **Sex-Linked Inheritance**
 - The appearance of a trait because of the presence of an allele either on X chromosome or Y chromosome is called Sex-linked Inheritance.
- **Diseases observed in X-linked Inheritance**
 - Haemophilia

- Colour-Blindness

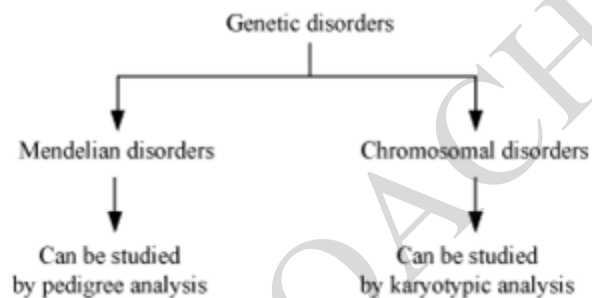
- **Criss-Cross Inheritance**

- The transfer of a gene from mother to son or father to daughter is called as criss-cross inheritance. For e.g. in X-chromosome linkage

Mutation

- It is the sudden change in genotype due to the alteration in DNA sequences.
- **Mutation and recombination** brings variation in DNA.
- **Point mutation** arises due to the change in a single base pair in DNA; for example, sickle-cell anaemia.
- **Frame shift mutation** arises due to deletion and insertion of base pairs.
- **Mutagens** are factors that induce mutations; for example, UV radiation.
- Inheritable mutations can be studied by pedigree analysis

Genetic Disorders



- **Examples of Mendelian disorder:**

1. **Haemophilia – Sex-linked recessive disorder**
2. **Sickle-cell anaemia – Autosome-linked recessive disorder**
3. **Phenylketonuria – Inborn error of metabolism; autosomal-recessive disorder**

- **Examples of chromosomal disorder:**

1. **Aneuploidy is the presence of abnormal number of chromosomes in an individual.**

- **Down's syndrome** – Characterised by trisomy of the 21st chromosome; chromosomes increase from 46 to 47
- **Klinefelter's syndrome** – Characterised by the presence of an additional X-chromosome; Karyotype – 47, XXY
- **Turner's syndrome** – Characterised by monosomy of sex chromosomes. Karyotype – 45, XO