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 F_1 plants by cross pollination
    - Self pollination of F_1 plants to obtain F_2 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 F_1 generation while both the traits get expressed in the F_2 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 co-existence 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.

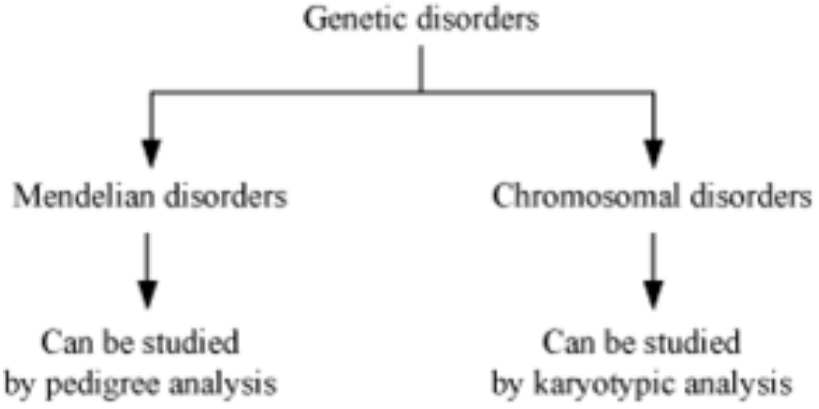
- **Sex determination**
    - **Female heterogamy:** Presence of two kinds of sex chromosomes in the female; only one kind is present in the male; for example, birds (the female has ZW sex chromosome while the male has ZZ sex chromosome).
    - **Male heterogamy:** Presence of two kinds of sex chromosomes 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