16. Heredity and Variation

- **Nucleus**
  - It controls all the cellular activities of cell.
  - It consists of the following.
    a. **Nuclear membrane –** It has perforations called nuclear pores.
    b. **Nucleoplasm**
    c. **Nucleolus**
  - **Nucleolus** is the site of ribosomal RNA formation.
  - Network of nucleoprotein fibres are called the chromatin.
  - Chromatin contains DNA, histones (basic proteins), non-histone proteins, and RNA.
  - Chromatin threads condense and organize to form chromosome.
  - The primary constriction in the chromosome is called centromere.

- Based on the position of centromere, chromosomes are of four types:
  - **Metacentric:** Centromere is located at the middle of chromosome.
  - **Sub-metacentric:** Centromere is slightly away from the middle of chromosome.
  - **Acrocentric:** Centromere is situated close to the end of chromosome.
  - **Telocentric:** Centromere is located at the terminal end.

- A small chromosomal segment separated from the main body of the chromosome by a secondary constriction is called satellite.
- **Structure of DNA**
  - It has a double-helix structure, similar to ladder.
  - It is made up of nucleotides.
  - Nuclotides are made of sugar, phosphate groups and nitrogen bases
- **Components of DNA**
  - Sugar
  - Phosphate groups
  - Nitrogen bases
- **Genes**
  - It is a unit of DNA.
  - Located on Chromosomes.
  - Controls the development of one or more traits.
  - It is the basis of Inheritrance.
  - It can acquire mutation leading to variation.

RNA

- There are three types of RNA –
  - **Messenger RNA (mRNA):** Provides template to initiate translation
  - **Transfer RNA (tRNA)**: Brings amino acids to mRNA; also known as adapter molecule
  - **Ribosomal RNA (rRNA)**: Has structural and catalytic role during translation

- **Heredity-** transmission of characteristics or traits from parents to offsprings.
- **Variations-** difference among individuals of a species and also among offsprings of same parents.
- Variations are of two types- heritable and non-heritable.
- **Basis of heredity-** each trait is influenced by both maternal and paternal DNA.

Mendel's work

- Proposed- heredity is controlled by factors. Factors are now called genes.
- Performed experiments on garden pea (*Pisum sativum*)
- Used seven contrasting pairs of characters or traits to study heredity.
- **Dominant trait-** able to express itself over another contrasting trait
- **Recessive trait-**unable to express its effect in the presence of a dominant trait
- Mendel represented- dominant trait as upper case (e.g., T for tallness) and recessive trait as lower case (e.g., t for shortness)
- **Homozygous-** when the factors or genes of a trait are similar e.g., TT or tt
- **Heterozygous-** when the factors or genes of a trait are different e.g., Tt
- **Genotype-**genetic constitution of an organism e.g., pure tall- TT
- **Phenotype-**observable traits or characteristics of an organism e.g., tallness, shortness etc.
- **Genotypic ratio-**expected ratio of genotypes produced by a particular cross
- **Phenotypic ratio-**expected ratio of phenotypes produced by a particular cross
- **Monohybrid cross-**involves only one pair of contrasting characters
- **Phenotypic ratio in monohybrid cross is 3:1**
- **Dihybrid cross-**involves two pairs of contrasting characters
- **Phenotypic ratio in dihybrid cross is 9:3:3:1**

Stages of Mendel's experiment

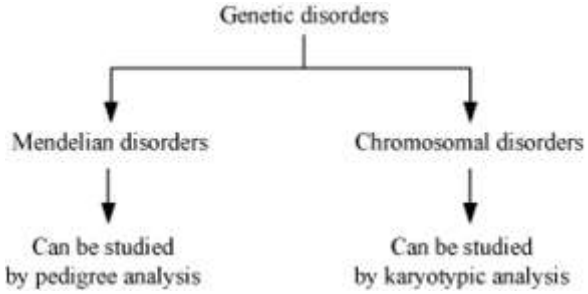
- Selection of parents- true breeding with contrasting pairs of traits e.g., pure tall (TT) and pure dwarf (tt) pea plants were selected
- Obtaining F_1 plants- F_1 generation is the first filial generation, formed after crossing desirable parents e.g., crossing pure tall (TT) and dwarf (tt) plants gives heterozygous tall (Tt) F_1 plants
- Self-pollination of F_1 plants- involves crossing F_1 plants to obtain F_2 plants

- **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.

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