BHARATHIDASAN UNIVERSITY

Tiruchirappalli- 620024, Tamil Nadu,
India

Programme: M.Sc., Biomedical Science
(5 Year Integrated Program)

Course Title : Principles of Genetics
Course Code : BM24C4

Unit-I
Introduction to Genetics and Mendelian Laws

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INTRODUCTION TO GENETICS & INHERITANCE

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Structural Hierarchy of Living Organisms

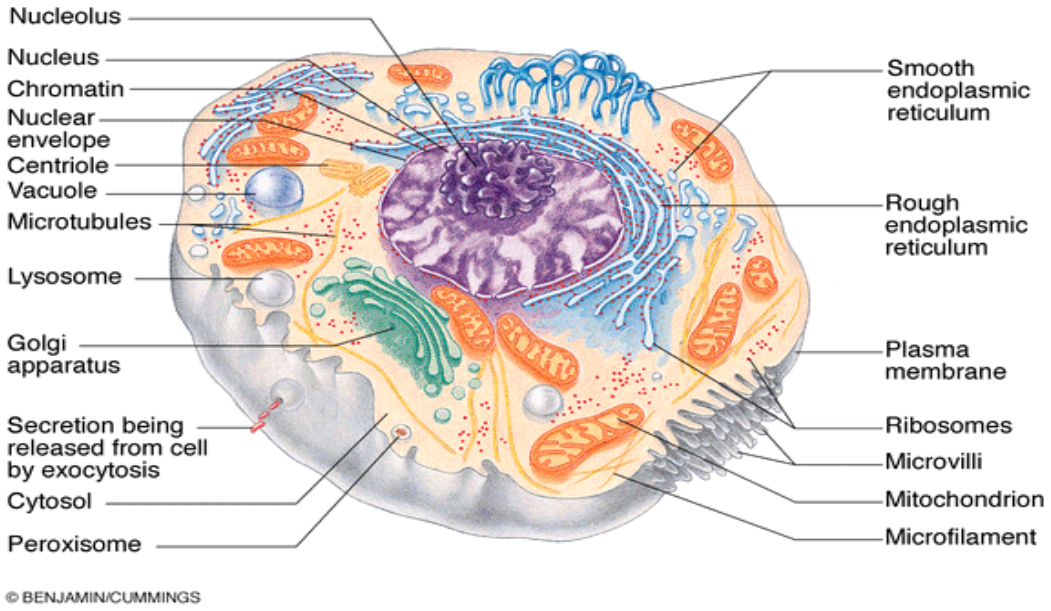
Cell Simplest Structure which can perform all living functions

Tissue Cells working together to perform certain specific functions

Organ Tissue working together to perform specific functions

System Organs working together to perform specific functions

Specificity division of labor



Introduction to Genetics

What is Genetics?

- Study of heredity (how traits are passed from parents to offspring).
- Study of variation (differences among individuals).

Key Terms:

- **Gene** : Unit of heredity (e.g., the gene for eye color).
- **Allele** : Variants of a gene (e.g., blue vs brown eye color alleles).
- **DNA** : Molecule carrying genetic information.
- **Chromosome** : Organized structure of DNA in cells.

Applications:

- Studying genetic diseases (e.g., sickle cell anemia).
- Improving crop yields (e.g., pest-resistant rice).

Mendel and the Laws of Inheritance

Who was Mendel?

- Gregor Mendel (1822–1884), Austrian monk and botanist.
- Conducted experiments on pea plants (Pisum sativum).

Why Pea Plants?

- Distinct traits (e.g., tall vs short plants, yellow vs green seeds).
- Easy to cross-pollinate and reproduce quickly.

Mendel Laws:

Law of Dominance:

- Dominant alleles mask recessive ones.
  Example: In Tt plants, T (tall) is expressed, while t (short) is masked.

Law of Segregation:

- Each organism has two alleles per gene, which segregate during gamete formation.
  Example: Crossing tall (TT) and short (tt) plants yields all tall plants (Tt) in F1.

Law of Independent Assortment:

- Genes for different traits assort independently during gamete formation.
- Example: In dihybrid crosses (seed shape and color), round-yellow and wrinkled-green traits assort independently.

Father of Genetics

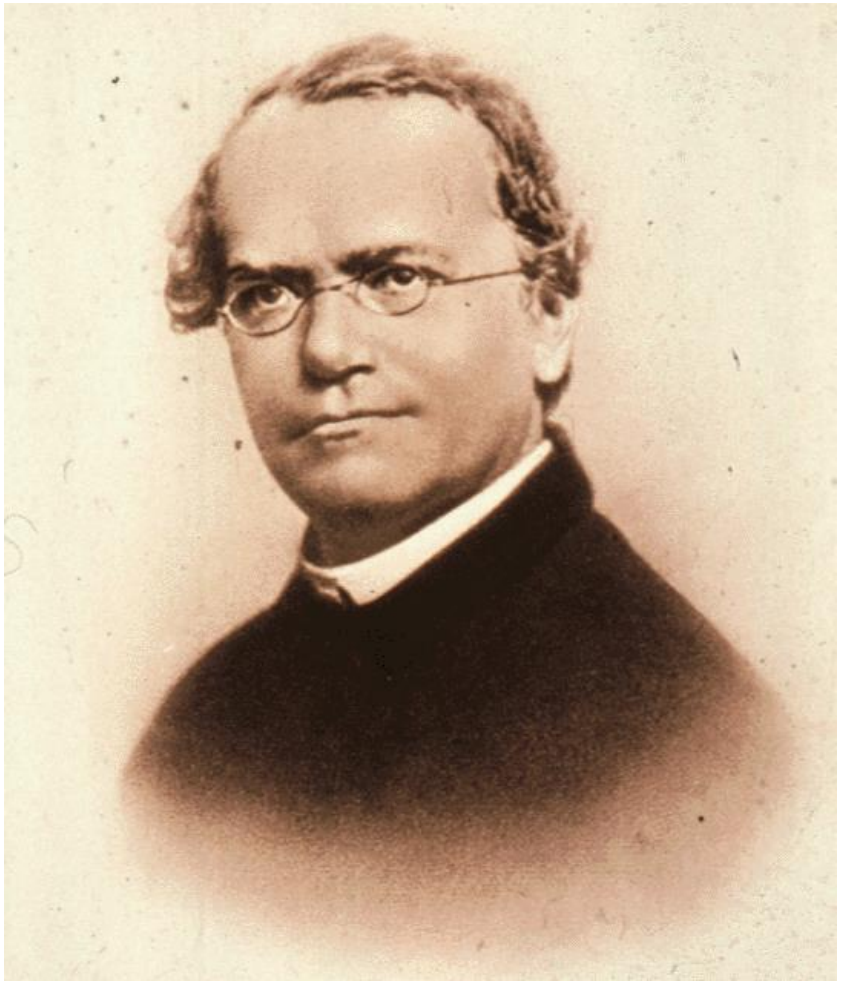
- **Johann Gregor Mendel** (1822 – 1884): An Austrian Monk considered as the pioneer of classical genetics.
- Hr proposed basic laws of genetics in 1866 but no recognition or appreciation was shown until 1900.

- **Reasons**:

  1. Biologists holding onto the Darwin's theory proposed in 1859.

  2. Mendel was ahead of his time in his statistical and experimental methods.

  3. Obscurity of the journal in which his results were published.



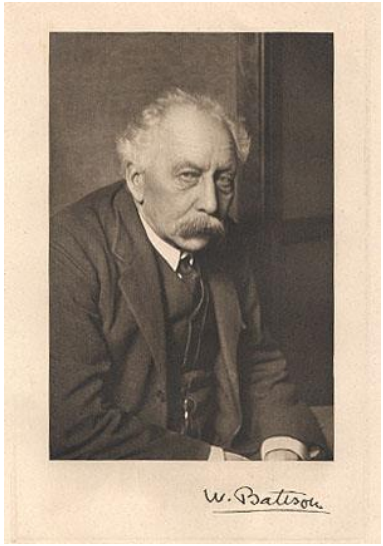
PREMS

Rediscovery

- Three botanists independently derived results similar to that of Mendel in 1900.
- Hugo de Vries *(Oenothera),* Carl Correns (peas, maize and *Xenia*) and Erich von Tschermak (flowering plants).
- Thus Mendel's work was rediscovered and published in the journal called Flora in 1901.
- Bateson conducted hybridization experiments to further confirm Mendel's Work.

Carl Correns, around 1925.

Hugo De Vries, around 1920.
Photo courtesy of
Cold Spring Harbor Laboratory Archives.



Erich von Tschermak-Seysenegg

PREMS

Cellular and Molecular Basis of Inheritance

Chromosome Structure:

- DNA wrapped around proteins (histones) to form chromatin.
- Chromatin condenses to form chromosomes during cell division.
- Visible under a microscope during metaphase.

DNA Structure:

- Double helix, composed of nucleotides (A, T, G, C).
- Stores genetic information (e.g., sequence coding for hemoglobin protein).

Genes and Traits:

- Example: Gene for blood type is located on chromosome 9.
- Determines traits such as height, hair color, and predisposition to diseases.

Mutations and Mutagenesis

What are Mutations?

- Permanent changes in the DNA sequence.
- Can occur naturally (errors in replication) or due to external factors (mutagens).

Types of Mutations:

- **Point Mutation**: Single nucleotide change.
  Example: Sickle cell anemia (GAG → GTG in the hemoglobin gene).
- **Frameshift Mutation**: Insertion or deletion of nucleotides.
  Example: Cystic fibrosis (deletion of three bases in the CFTR gene).
- **Chromosomal Mutations**: Changes in chromosome structure.
  Example: Duplication in Charcot-Marie-Tooth disease.

Mutagenesis:

Physical Mutagens : UV rays causing thymine dimers in DNA.
Chemical Mutagens : Benzopyrene in tobacco smoke.
Biological Mutagens : HPV leading to cervical cancer.

Impact of Mutations:

Positive : Evolutionary adaptations (e.g., antibiotic resistance in bacteria).
Negative : Genetic disorders (e.g., Huntington's disease).

Chromosomes

(Organized structures of DNA, carrying genetic information)

Human Chromosomes:

23 pairs (46 chromosomes): 22 autosomes + 1 pair of sex chromosomes.
Example: Females (XX), Males (XY).

Chromosome Parts:

Centromere: Links sister chromatids, important during cell division.
Telomeres: Protective ends of chromosomes (shortening linked to aging).
Chromatids: Identical copies after DNA replication.

Human Genome Project:

Sequencing all human DNA (completed in 2003).
Identified locations of many genes and their functions.

Chromosomal Nomenclature and Abnormalities

Chromosome Nomenclature:

Numbered based on size and banding patterns (e.g., 1–22, X, Y).
Bands identified via staining techniques (e.g., G-banding).

Chromosomal Abnormalities:

Numerical:

Example: Down Syndrome (Trisomy 21).
Klinefelter Syndrome (47, XXY).

Structural:

Example:

Deletion: Cri du Chat Syndrome (deletion in chromosome 5).
Translocation: Chronic myeloid leukemia (Philadelphia chromosome).

Causes and Effects:

Errors in meiosis or mitosis.
Symptoms vary: from mild developmental delays to severe physical disabilities.

Applications of Genetics

Medicine:

Genetic counseling for hereditary diseases.

Gene therapy for treating genetic disorders (e.g., SCID).

Agriculture:

GMO crops (e.g., Bt cotton, Golden rice).

Improved livestock breeding.

Forensics:

DNA fingerprinting for crime investigations.

Evolution:

Understanding speciation and biodiversity.