



الوراثة الطبية 1 / الكورس الاول

INTRODUCTION OF MEDICAL GENETICS

Sawa University

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Department of Medical Laboratories

قسم تقنيات المختبرات الطبية

Third Stage

المرحلة الثالثة

محاضرة رقم 1

الجانب النظري : الوراثة الطبية

Lecture No. 1

Theoretical : Medical Genetics

تدريسي المادة : م. د عباس علي

Learning Objectives

By the end of this lecture, you will be able to:

- Define genetics, heredity & variation.
- Outline major historical milestones.
- Use basic genetic terminology correctly.
- Distinguish major inheritance patterns.
- Give real-world examples of genetic applications.

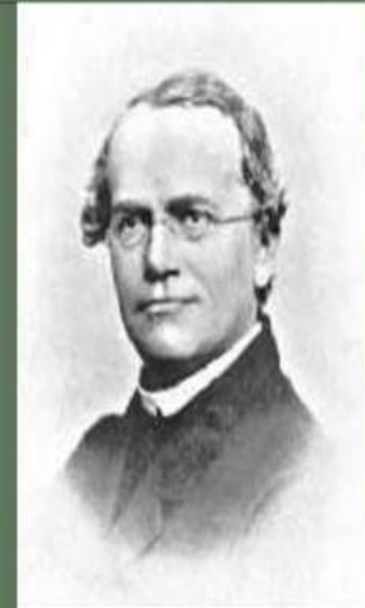
What is Genetics?

- **Genetics** is the branch of biology that studies heredity (**how traits are passed from parents to offspring**) and variation (**differences between individuals**).
- It explains how DNA, genes, and chromosomes determine physical and biological characteristics.
- The word “**genetics**” was first used by William Bateson in 1905 and derived from the Greek word “**Gene**” which mean to become or to grow.
- Genetics connects molecular biology, evolution, medicine, and biotechnology by explaining how traits are inherited and how new traits arise.

Historical Background

- **Gregor Mendel (1866):**
 - - Known as the **Father of Genetics**.
 - - Discovered basic laws of inheritance through pea plant experiments, establishing concepts of dominant and recessive traits
- **Other milestones:**
 - -. **Friedrich Miescher (1869):** First isolated “nuclein” (later known as **DNA**) from cell nuclei.
 - Early 1900s: Chromosome theory of inheritance linked Mendel’s factors to physical structures in cells.
 - **Thomas Hunt Morgan (1910):** Demonstrated that genes reside on chromosomes using fruit flies.

History of Genetics



Gregor Mendel the “Father of Genetics”

Historical Background

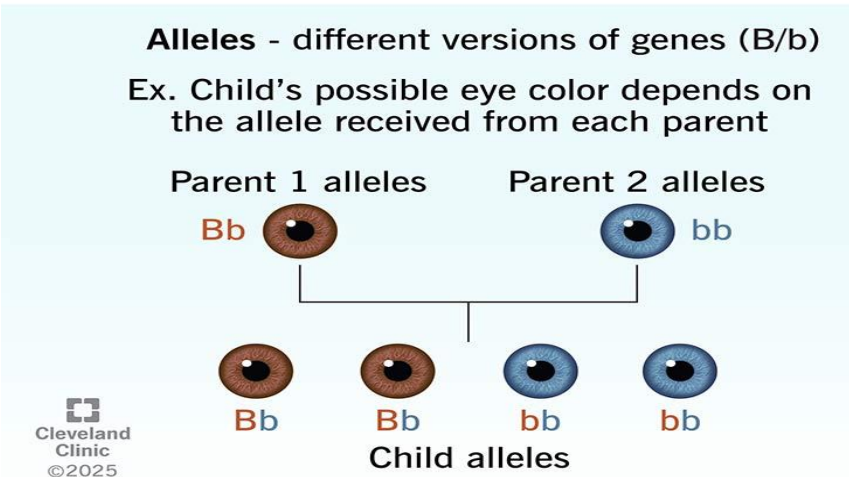
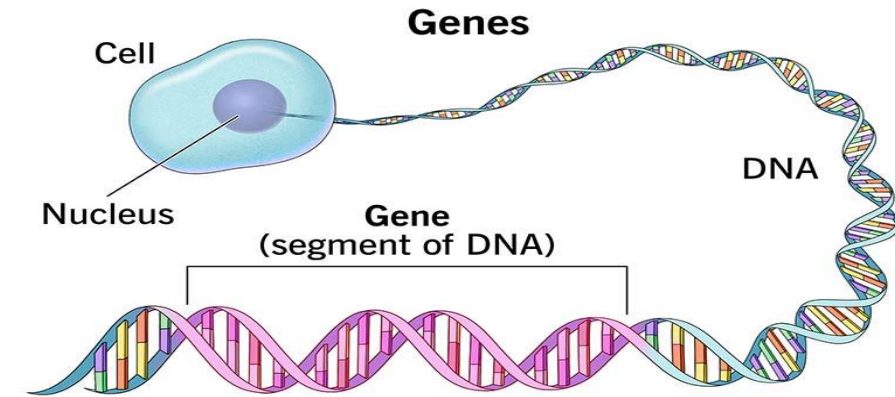
- **Oswald Avery (1944)**: Proved that DNA carries genetic information.
- **Hershey & Chase (1952)**: Confirmed that DNA is the genetic material (not protein) by using bacteriophage experiments.
- **Watson & Crick (1953)**: Proposed the double-helix model of DNA
- **Human Genome Project (2003)**: Completed the sequencing of the entire human genome, paving the way for genomic medicine.



Watson & Crick

Basic Genetic Concepts

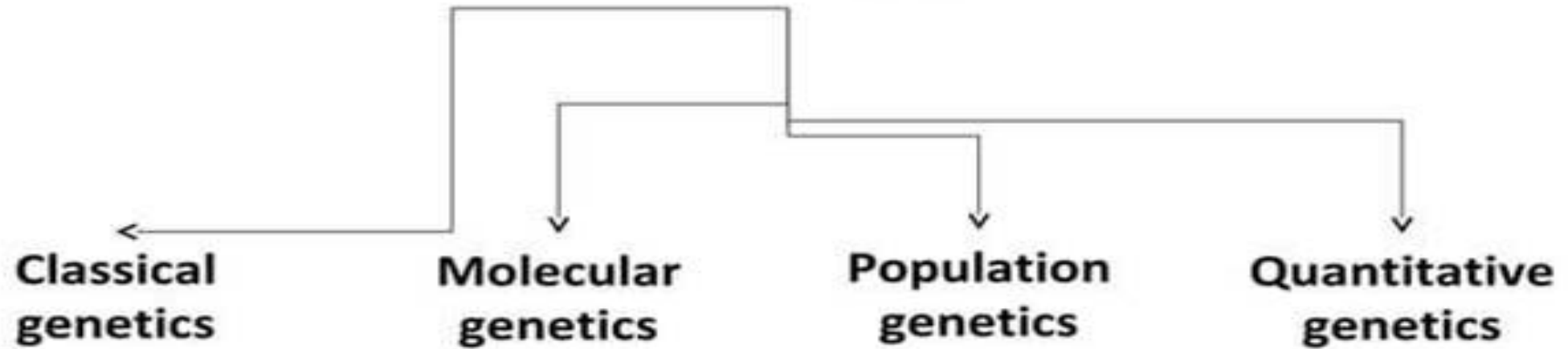
- **Gene:** A segment of DNA that codes for a specific trait or protein.
- **Alleles:** Different forms of a gene (dominant or recessive).
- **DNA:** Deoxyribonucleic acid, the chemical molecule that carries genetic instructions.
- **Chromosome:** A thread-like structure of DNA carrying genetic information.
- **Genome:** The complete set of an organism's DNA.
- **Mutation:** A change in DNA sequence that may affect traits.



Applications of Genetics

- - **Medical Genetics**: Diagnosis and treatment of genetic diseases.
- - **Agricultural Genetics**: Production of disease-resistant crops and animals.
- - **Forensic Science**: DNA fingerprinting for crime investigation.
- - **Biotechnology**: Genetic engineering, cloning, and CRISPR technology.
- - **Evolutionary Studies**: Understanding species evolution and adaptation.

The four major subdivisions Genetics



Types of Genetics

- **Classical (Mendelian) Genetics:** Study of inheritance patterns using crosses and observable traits.
- **Molecular Genetics:** Focus on the structure and function of genes at the DNA and RNA levels.
- **Population Genetics:** Study gene frequency changes in populations over time under forces like natural selection and genetic drift.
- **Quantitative Genetics:** Analysis of complex traits controlled by multiple genes and environmental factors.
- **Genomics:** Comprehensive study of whole genomes, including sequencing, mapping, and bioinformatics.
- **Epigenetics:** Investigation of heritable changes in gene expression without alterations in DNA sequence (e.g., DNA methylation).

Mendelian Genetics: Laws of Inheritance

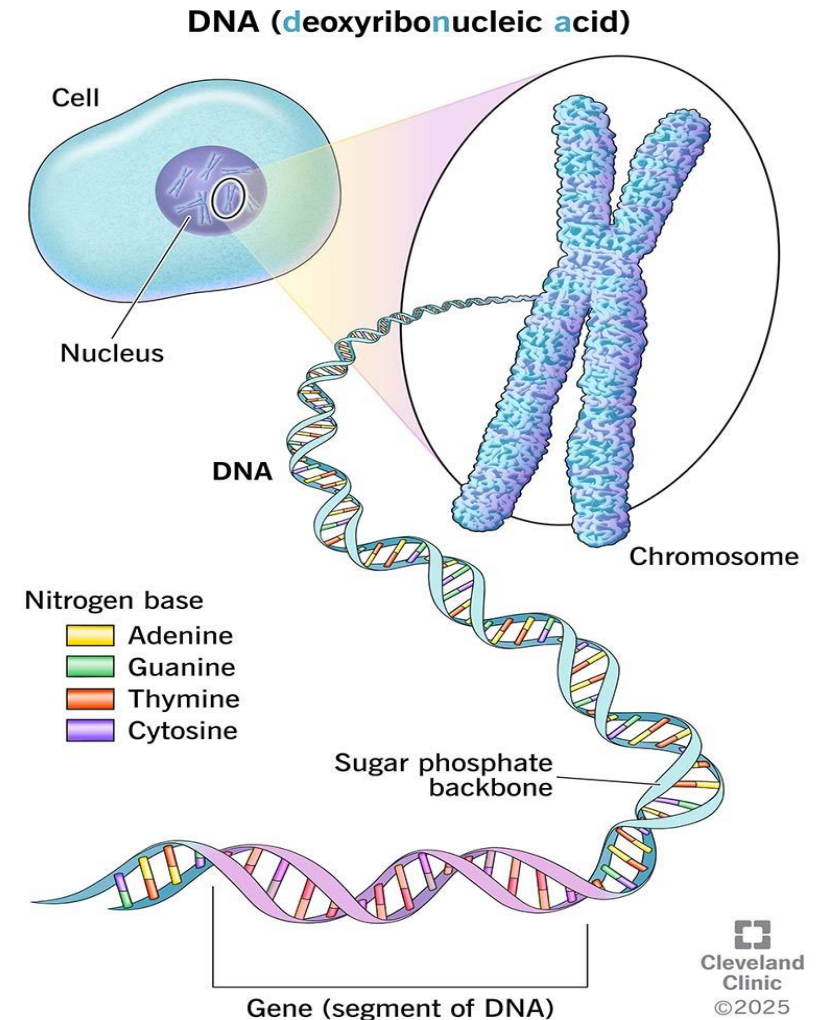
- **Law of dominance:** Some alleles are dominant while others are recessive; an organism with at least one dominant allele will display the effect of the dominant allele.
- **Law of segregation:** During gamete formation, the alleles for each gene segregate from each other so that each gamete carries only one allele for each gene.
- **Law of independent assortment:** Genes of different traits can segregate independently during the formation of gametes.

Patterns of Inheritance

- - **Autosomal Dominant**: Trait expressed if one dominant allele is present.
- - **Autosomal Recessive**: Trait expressed only when two recessive alleles are present.
- - **X-linked** (Sex-linked): Trait linked to genes on the X chromosome.
- - **Incomplete Dominance**: A blending of traits (e.g., pink flowers from red $\times$ white).
- - **Codominance**: Both alleles are expressed equally (e.g., AB blood group).
- - **Polygenic Inheritance**: Multiple genes influence a single trait (e.g., height, skin color).

Structure of DNA

- **Components:** Nucleotides consisting of a phosphate group, deoxyribose sugar, and a nitrogenous base (A, T, G, C).
- **Base Pairing:** Adenine pairs with Thymine; Cytosine pairs with Guanine through hydrogen bonds.
- **Double Helix:** Two antiparallel strands twist around each other to form a stable helical structure.
- **DNA vs RNA:** RNA contains ribose sugar, uracil instead of thymine, and is usually single-stranded.
- The sequence of bases encodes genetic instructions for building proteins.

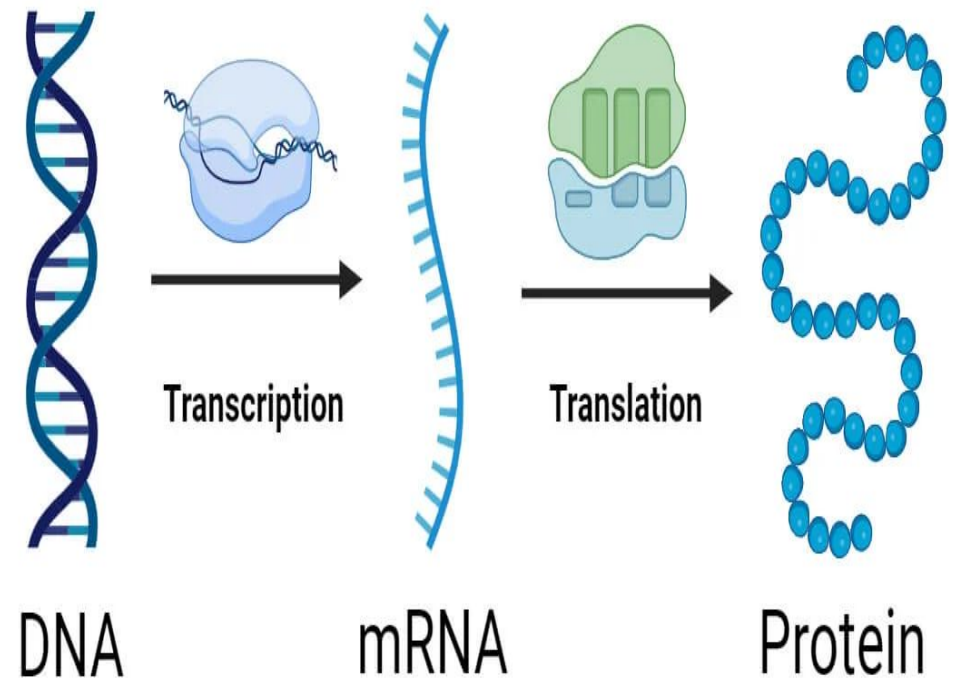


Central Dogma of Molecular Biology

- **DNA → RNA → Protein**

- **Transcription:** DNA is copied into messenger RNA (mRNA) inside the nucleus.
- **Translation:** mRNA is decoded by ribosomes to synthesize proteins in the cytoplasm.
- Proteins determine cellular structure, metabolic activity, and visible traits.
- **Gene regulation** allows cells to respond to environmental changes and differentiate into various tissue types.

Gene Expression



Reference:

Jorde, L.B.; Carey, J.C & Bamshad, M.J. (2019). Medical genetics. Sixth Edition, June 1.

Any questions?

*Thank
You*