

# Introduction to Genetics

## Introduction to Genetics – Study Notes

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### 1. What Is Genetics?

- **Definition:** The branch of biology that studies how traits are inherited from one generation to the next.
  - **Key Question:** How do organisms pass on characteristics such as eye color, blood type, or disease susceptibility?
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### 2. Historical Milestones

| Year            | Scientist                          | Contribution                                                                  |
|-----------------|------------------------------------|-------------------------------------------------------------------------------|
| 1865            | Gregor Mendel                      | Established the laws of inheritance using pea plants; coined “genes” (later). |
| 1900            | Rediscovery of Mendel              | Hugo de Vries, Carl Correns, and Erich von Tschermak revived Mendel’s work.   |
| 1910?<br>1920   | Thomas Hunt Morgan                 | Demonstrated that genes reside on chromosomes (fruit fly experiments).        |
| 1953            | Watson & Crick                     | Described the double helix structure of DNA.                                  |
| 1955?<br>1960   | Erwin Chargaff & Rosalind Franklin | Provided critical data on DNA composition and structure.                      |
| 1970s?<br>1980s | Molecular cloning & PCR            | Enabled manipulation and amplification of specific DNA fragments.             |
| 2003            | Human Genome Project               | Completed the first reference sequence of the human genome.                   |

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### 3. Fundamental Concepts

#### 3.1 Genes & Alleles

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- **Gene:** A discrete unit of heredity located on a chromosome; encodes a functional product (usually a protein or RNA).
- **Allele:** Different versions of a gene that arise by mutation.
  - *Homozygous:* Two identical alleles (AA or aa).
  - *Heterozygous:* Two different alleles (Aa).

## 3.2 Chromosomes

- Linear DNA molecules packaged with proteins (histones) into a compact structure.
- Humans: 46 chromosomes (23 pairs) – 22 autosomes + 1 pair of sex chromosomes (XX or XY).

## 3.3 DNA ? RNA ? Protein (Central Dogma)

1. **Replication** – DNA copies itself before cell division.
2. **Transcription** – DNA ? messenger RNA (mRNA).
3. **Translation** – mRNA ? polypeptide chain (protein).

## 3.4 Types of Inheritance Patterns

| Pattern                     | Key Features                                                               | Example                           |
|-----------------------------|----------------------------------------------------------------------------|-----------------------------------|
| <b>Autosomal Dominant</b>   | One copy of a dominant allele produces phenotype.                          | Huntington disease                |
| <b>Autosomal Recessive</b>  | Two copies of a recessive allele needed.                                   | Cystic fibrosis                   |
| <b>X?Linked Dominant</b>    | Dominant allele on X chromosome; affects both sexes, more severe in males. | Rett syndrome                     |
| <b>X?Linked Recessive</b>   | Recessive allele on X chromosome; males express phenotype more often.      | Hemophilia A                      |
| <b>Mitochondrial</b>        | Inherited only from the mother; genes in mitochondrial DNA.                | Leber hereditary optic neuropathy |
| <b>Polygenic</b>            | Many genes contribute additively.                                          | Height, skin color                |
| <b>Incomplete Dominance</b> | Heterozygote shows intermediate phenotype.                                 | Pink snapdragon flowers (Rr)      |

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| Pattern     | Key Features                    | Example                   |
|-------------|---------------------------------|---------------------------|
| Codominance | Both alleles expressed equally. | ABO blood groups (IA? IB) |

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## 4. Mendelian Laws

### 1. Law of Segregation

- Each individual carries two alleles for a gene; these separate during gamete formation so each gamete receives only one allele.

### 2. Law of Independent Assortment

- Genes located on different chromosomes