Genetics

Chapter 25: GENETICS: THE SCIENCE OF HEREDITY

Learning Objectives: Upon completion of this chapter, students should be able to:

- Provide an introduction to genetics.
- Explain the transmission and expression of characters (traits) in organisms.
- Discuss how characters get transmitted and behave from generation to generation.
- Describe Mendel's work in genetics.
- Explain single-factor inheritance (monohybrid cross).
- Explain dihybrid cross.
- Define breeding true.
- Understand the concept of probability in genetics.
- Describe the structure of chromosomes and their location.
- Explain DNA replication and its role in heredity.
- Discuss the role of chromosomes in transmitting hereditary characters.
- Identify amendments to Mendelian genetics (e.g., incomplete dominance, co-dominance, linkage, multiple alleles, polygenic inheritance).
- Discuss sex determination.
- Explain sex-linked characters.
- Describe mutations.
- Discuss non-disjunction.
- Explain the application of genetics in medicine (genetic disorders, Rhesus factor) and agriculture (inbreeding, outbreeding, animal breeding, plant breeding).
- Briefly touch on hybridoma technology (biotechnology genetic engineering).

25.1 Introduction to Genetics

- **Genetics:** The scientific study of heredity and variation in living organisms. It explains how traits are inherited from parents to offspring.
- **Heredity:** The passing of traits from parents to their offspring.
- **Characters (Traits):** Specific characteristics or features of an organism that are determined by genes (e.g., eye color, plant height, blood group).
- **Transmission and Expression of Characters:** Genetic information (genes) is passed from one generation to the next. These genes then direct the synthesis of proteins, which ultimately determine the expression of traits.

25.2 Mendelian Genetics

- **Gregor Mendel:** An Austrian monk, often called the "father of genetics," who performed experiments with pea plants in the mid-19th century. His work laid the foundation for modern genetics.
- **Mendel's Work:** He studied the inheritance of single traits, observing consistent patterns that led to his laws of inheritance.
- **Breeding True (Purebred):** An organism that, when self-pollinated or bred with another organism of the same type, produces offspring identical to the parent for a particular trait. This indicates homozygosity for the trait.
- **Single-Factor Inheritance (Monohybrid Cross):**
    - **Definition:** The study of the inheritance of a single gene controlling one trait.
    - **Key Terms:**
        - **Gene:** A segment of DNA that codes for a specific trait.
        - **Allele:** Different forms of a gene (e.g., for pea plant height, there's an allele for tall and an allele for dwarf).
        - **Dominant Allele:** An allele that expresses its trait even when only one copy is present (represented by a capital letter, e.g., T for tall).
        - **Recessive Allele:** An allele whose trait is only expressed when two copies are present (represented by a lowercase letter, e.g., t for dwarf).
        - **Genotype:** The genetic makeup of an organism (e.g., TT, Tt, tt).
        - **Phenotype:** The observable physical characteristics of an organism, determined by its genotype (e.g., Tall, Dwarf).
        - **Homozygous:** Having two identical alleles for a trait (e.g., TT or tt).
        - **Heterozygous:** Having two different alleles for a trait (e.g., Tt).
    - **Mendel's Law of Segregation:** Alleles for a trait separate during gamete formation, so each gamete carries only one allele for each gene.
    - **Example (Pea plant height):** Cross between homozygous tall (TT) and homozygous dwarf (tt) produces F1 generation all heterozygous tall (Tt). Self-pollinating F1 produces F2 generation with a phenotypic ratio of 3 Tall : 1 Dwarf and a genotypic ratio of 1 TT : 2 Tt : 1 tt.
- **Dihybrid Cross:**
    - **Definition:** The study of the inheritance of two different traits simultaneously.
    - **Mendel's Law of Independent Assortment:** Alleles for different genes assort independently of each other during gamete formation, as long as they are on different chromosomes.
    - **Example:** Cross between pea plants pure for round, yellow seeds (RRYY) and wrinkled, green seeds (rryy) produces F1 all RrYy (round, yellow). Self-pollinating F1 produces F2 with a phenotypic ratio of 9:3:3:1 (e.g., 9 round yellow : 3 round green : 3 wrinkled yellow : 1 wrinkled green).
- **Probability in Genetics:**
    - Genetic crosses can be analyzed using probability to predict the likelihood of offspring having specific genotypes and phenotypes. Punnett squares are a tool for visualizing these probabilities.

25.3 Chromosomes and DNA

- **Chromosomes:** Thread-like structures found in the nucleus of eukaryotic cells, composed of DNA tightly coiled around proteins (histones).
    - **Location:** Primarily in the nucleus of eukaryotic cells.
    - **Role in Heredity:** Chromosomes carry genes, which are the units of hereditary information. They ensure the accurate transmission of genetic material during cell division (mitosis and meiosis).
- **DNA (Deoxyribonucleic Acid):** The genetic material of all living organisms. It is a double helix structure.
    - **DNA Replication:** The process by which DNA makes an identical copy of itself before cell division. This ensures that each new cell receives a complete set of genetic instructions.
    - **Importance in Heredity:** Faithful replication of DNA ensures that genetic information is accurately passed from parent cells to daughter cells and from parents to offspring.

25.4 Amendments to Mendelian Genetics (Non-Mendelian Inheritance)

Not all inheritance patterns follow Mendel's simple dominant-recessive model.

- **Incomplete Dominance:** Neither allele is completely dominant, resulting in a heterozygous phenotype that is intermediate between the two homozygous phenotypes.
    - *Example:* Red flower (RR) x White flower (WW) = Pink flower (RW).
- **Co-dominance:** Both alleles are fully expressed in the heterozygous phenotype.
    - *Example:* ABO blood groups (A and B alleles are co-dominant, leading to AB blood type where both A and B antigens are present).
- **Linkage:** Genes located on the same chromosome tend to be inherited together because they are "linked". This violates Mendel's law of independent assortment. Crossing over can separate linked genes.
- **Multiple Alleles:** More than two alleles exist for a given gene within a population (though an individual only has two).
    - *Example:* ABO blood group system (A, B, O alleles).
- **Polygenic Inheritance:** A single trait is controlled by two or more genes, often resulting in a continuous range of phenotypes.
    - *Examples:* Human height, skin color, intelligence.

25.5 Sex Determination and Sex-Linked Characters

- **Sex Determination:** The biological system that determines the development of sexual characteristics in an organism. In humans, it's typically determined by sex chromosomes.
    - **Humans:** Females have two X chromosomes (XX), males have one X and one Y chromosome (XY). The presence of a Y chromosome (specifically the SRY gene) determines maleness.
- **Sex-Linked Characters:** Traits whose genes are located on the sex chromosomes (usually the X chromosome in humans).

    - Since males have only one X chromosome, they express recessive sex-linked traits more often than females (who have a second X chromosome that can mask the recessive allele).
    - *Examples:* Red-green color blindness, hemophilia (bleeding disorder).

25.6 Mutations

- **Definition:** A permanent, random change in the DNA sequence of an organism's genome.
- **Causes:** Can occur spontaneously (errors during DNA replication) or be induced by mutagens (e.g., radiation, certain chemicals).
- **Effects:** Can be harmful (leading to genetic disorders or cancer), neutral (no observable effect), or rarely beneficial (providing new variation for evolution).
- **Significance:** Mutations are the ultimate source of new genetic variation in a population, driving evolution.

25.7 Non-Disjunction

- **Definition:** The failure of homologous chromosomes or sister chromatids to separate properly during cell division (meiosis or mitosis).
- **Consequences:** Results in gametes or cells with an abnormal number of chromosomes (aneuploidy).
    - *Example:* Down syndrome (Trisomy 21), where an individual has three copies of chromosome 21 instead of two.

25.8 Application of Genetics

Genetics has vast applications in various fields:

- **A. Medicine:**
    - **Genetic Disorders:** Understanding inheritance patterns helps diagnose, counsel, and potentially treat genetic diseases caused by mutations or chromosomal abnormalities (e.g., sickle cell anemia, cystic fibrosis, Huntington's disease).
    - **Rhesus (Rh) Factor:** A blood group system. Incompatibility between Rh-negative mothers and Rh-positive fetuses can lead to hemolytic disease of the newborn, which can be prevented with medical intervention.
    - **Genetic Counseling:** Providing information and support to families affected by or at risk of genetic disorders.
    - **Gene Therapy:** Experimental technique using genes to treat or prevent disease.
- **B. Agriculture:**
    - **Inbreeding:** Mating of closely related individuals to maintain desirable traits and homozygosity. Can lead to inbreeding depression (reduced vigor due to increased homozygosity of deleterious recessive alleles).
    - **Outbreeding (Cross-breeding):** Mating of unrelated individuals, often to introduce new genes or increase genetic diversity. Can lead to hybrid vigor (heterosis).
    - **Animal Breeding:** Selective breeding to improve desirable traits in livestock (e.g., milk yield, meat quality, disease resistance).
    - **Plant Breeding:** Selective breeding to develop new crop varieties with improved yield, disease resistance, nutritional content, or stress tolerance.

    - **Genetically Modified Organisms (GMOs):** Using genetic engineering to introduce new traits into crops or animals (e.g., Bt corn for pest resistance).
- **C. Biotechnology and Genetic Engineering:**
    - **Hybridoma Technology:** A technique used to produce large quantities of specific antibodies (monoclonal antibodies) by fusing antibody-producing B cells with myeloma (cancer) cells. Used in diagnostics and therapies.
    - **Recombinant DNA Technology:** Involves cutting and pasting DNA sequences to create new genetic combinations. Basis for gene cloning, genetic modification, and gene therapy.
    - **CRISPR-Cas9:** A powerful gene-editing tool for precise modification of DNA.