Chapter 1: Overview of Genetics

Student Learning Objectives

Upon completion of this chapter the student should be able to:

1. Understand the key biological molecules that are associated with the study of genetics.
2. Understand the relationships between genes and traits and the types of traits that are studied by geneticists.
3. Recognize the three major fields of genetics (transmission, molecular, and population) and the general characteristics of each field.
4. Discuss how genetics is an experimental science, and outline different strategies for solving problems in genetics.

Key Terms

Alleles
Amino acid
Behavioral traits
Biological evolution / Evolution
Carbohydrates
Cellular level
Chromosomes
Deoxyribonucleic acid (DNA)
Diploid
Discovery-based science
Environment
Enzymes
Gametes
Gene
Gene expression
Gene mutations
Genome
Genetic approach
Genetic code
Genetic cross
Genetics
Genetic variation
Haploid
Homologs
Hypothesis testing
Lipids
Loss-of-function allele
Loss-of-function mutation
Macromolecules
Messenger RNA (mRNA)
Model organisms
Molecular level
Morphological traits
Morphs
Natural selection
Norm of reaction
Nucleic acids
Nucleotides
Organelle
Organism level
Phenylketonuria (PKU)
Physiological traits
Polypeptides
Proteins
Proteome
Population level
Ribonucleic acid (RNA)
Scientific method
Somatic Cells
Species
Traits
Transcription
Translation

Chapter Outline

Introduction

1. The Human Genome Project began in 1990 as a project between the National Institutes of Health (NIH) and the Department of Energy (DOE). The project was completed in 2003 (Figure 1.1).
   a. human genome contains approximately 3 billion nucleotides
   b. project is 99.99% accurate (1 in 10,000 nucleotides)
2. Human Genome Project enables scientists to:
   a. determine the number of genes in humans
   b. examine the relationship between genes and living cells
   c. study the evolution of species
   d. understand developmental genetics
   e. explore the relationship between genetic mutations and disease
   f. develop new technologies for genetic studies
3. Modern genetic studies are a result of developments associated with the Human Genome Project.
   a. The development of new medicines, such human insulin manufactured by *E. coli* bacteria.
   b. Cloning of mammals, such as Dolly the sheep by Ian Wilmut and associates (1997) (Figure 1.2).
   c. production of transgenic organisms, such as those that contain an introduced GFP gene from jellyfish (Figure 1.3)

1.1 The Molecular Expression of Genes

1. Genes are the basic units of heredity, while traits are the characteristics of an organism. Genes provide the blueprint for an organism's traits.

Living Cells Are Composed of Biochemicals

1. Organic molecules provide energy for cellular functions as well as the building blocks for larger molecules.
2. Categories of biologically important organic molecules:
   a. nucleic acids
   b. proteins
   c. carbohydrates
   d. lipids
3. Molecules that are made of repetitive subunits are called macromolecules.
4. Larger cellular structures, such as chromosomes, are built from combinations of micromolecules and macromolecules (Figure 1.4).

Each Cell Contains Many Different Proteins That Determine Cell Structure and Function

1. Cellular characteristics are determined primarily by proteins.
2. All of the proteins that a cell or organism makes at a given time is called the proteome.
3. General roles of proteins include: cellular support, transport across the cell membrane, biological motors, cell-to-cell recognition, and cell signaling.
4. Enzymes are primarily proteins that accelerate a chemical reaction.

DNA Stores the Information for Protein Synthesis

1. DNA stores the information needed for synthesis of cellular proteins.
2. DNA is made of nucleotides. Each nucleotide includes a nitrogen-containing base, which is either adenine (A), thymine (T), cytosine (C), or guanine (G).
3. The information in the DNA encodes the sequence of amino acids in a protein.
    a. the genetic code relates the genetic information to the correct amino acid.
4. In cells, DNA is found in chromosomes. The information on a chromosome is organized as genes (Figure 1.5).
    a. on average, a human chromosome contains about 1,000 genes

The Information in DNA is Accessed During the Process of Gene Expression

1. Gene expression refers to the use of the genetic information to synthesize a cellular protein.
2. Process includes two steps (Figure 1.6):
    a. information in the DNA is copied into RNA by transcription
    b. the RNA is then translated into a functional protein
3. The distinctive structure of a protein determines is cellular function.

1.2 The Relationship Between Genes and Traits

1. A trait is the displayed characteristic of an organism.
2. Morphological traits are associated with the appearance of an organism (eye color, height, etc.).
3. Physiological traits are associated with the ability of the organism to function, such as metabolic functions.
4. Behavioral traits are associated with how an organism responds to its environment.

The Molecular Expression of Genes Within Cells Leads to an Organism's Traits

1. Genetics spans four levels of biological organization (Figure 1.7):
    a. molecular level – the processes of transcription and translation
    b. cellular level – the function of a protein within the cell
    c. organism level – the observed traits of an organism
    d. population level – the occurrence of the trait in a population
2. Forms of a gene are called alleles.
    a. Different alleles of a gene have different DNA sequences.
    b. An example is eye color in humans. The gene is for eye pigmentation, the alleles of the gene determine the color, and different eye colors are produced by different alleles.

Inherited Differences in Traits Are Due to Genetic Variation

1. Genetic variation is the differences in inherited traits among individuals of a population.
2. For species that occupy wide geographic ranges, these differences may be drastic enough so that scientists may consider the organisms to be different species. These are called morphs (Figure 1.8).
3. Genetic variation is due to changes in the nucleotide sequence of the DNA. These variations may be caused by:
    a. gene mutations at the nucleotide level
    b. major structural changes in a chromosome
    c. variation in the total number of chromosomes (Figure 1.9)

Traits Are Governed by Genes and by the Environment

1. The external environment may influence an organism's traits.
2. The norm of reaction refers to the effects of environmental variation on an individual's traits.
3. Example is the human disease phenylketonuria (PKU), which encodes a gene called phenylalanine hydroxylase. This gene allows for the metabolism of the amino acid phenylalanine.
    a. mutations causing a defect in this gene mean that toxic levels of phenylalanine accumulate in the blood, causing mental retardation
    b. defective gene present in 1 in 8,000 births in the U.S.
    c. By eating a diet free of phenylalanine, an individual with a defective PKU allele can avoid the symptoms of the disease (Figure 1.10).

During Reproduction, Genes Are Passed from Parent to Offspring

1. Gregor Mendel first established that genetic information was passed from parent to offspring as discrete units (genes).
2. Sexually reproducing species are usually diploid (two copies of each chromosome, or 2n) (Figure 1.11a).
3. The copies of each chromosome are called homologs.
4. Gametes are haploid (one copy of each chromosome, also known as the n number) (Figure 1.11b).
5. Sexual reproduction increases genetic variation in a population.
6. Somatic cells, also known as body cells, are diploid.

The Genetic Composition of a Species Evolves over the Course of Many Generations

1. The change in the genetic composition of a species over time is called biological evolution.
2. Charles Darwin proposed the theory of natural selection as the mechanism for biological evolution (Figure 1.12).

1.3 Fields of Genetics

1. Genetics is a broad discipline encompassing molecular, cellular, organism, and

population biology.
2. The study of model organisms (organisms studied by many different researchers so they can compare their results and determine scientific principles that apply more broadly to other species) have led to our understanding of genes in all species.

Transmission Genetics Explores the Inheritance Patterns of Traits as They Are Passed from Parents to Offspring

1. Framework provided by Gregor Mendel who suggested that genetic determinants (genes) were discrete units that were passed from generation to generation.
2. Studies of transmission genetics rely on the genetic cross to examine how traits are passed from parents to offspring.

Molecular Genetics Focuses on a Biochemical Understanding of the Hereditary Material

1. The goal is to understand the workings of the genetic material at the molecular level.
2. Molecular geneticists use a genetic approach to study mutant genes with an abnormal function.
    a. Loss-of-function alleles (mutations) help geneticists understand the role of the gene in the organism.

Population Genetics Is Concerned with Genetic Variation and Its Role in Evolution

1. Involves the use of mathematical theories to explain the prevalence of certain genes in a population.
2. Provides a link between the study of transmission genetics (Mendel) and natural selection (Darwin).
3. Population geneticists study the relationship between genetic variation and the environment that an organism inhabits.

1.4 The Science of Genetics

Genetics Is an Experimental Science

1. Uses hypothesis testing, also known as the scientific method, and discovery-based science.
2. Hypothesis testing involves gathering of data to support or refute a putative conclusion.
3. Discovery-based science involves gathering of data without a preconceived hypothesis.

Genetics TIPS Will Help You to Improve Your Proble-Solving Skills

1. Students' learning will involve two general goals:
    a. Foundational knowledge – Learn to describe core concepts in genetics.
    b. Problem-solving skills – Be able to apply that knowledge in different ways.
2. The boxed feature in the textbook chapters called Genetics TIPS will help students develop these skills
    a. Topic, Information, and Problem-solving Strategy

List of Key Investigators

Chalfie, Martin – received Nobel Prize for the discovery and development of GFP.

Crisanti, Andrea – altered mosquitoes to express GFP only in the gonads of males, to allow them to sort males from females.

Darwin, Charles – Proposed the theory of natural selection as the mechanism for biological evolution.

Mendel, Gregor – Determined that genetic information was from generation to generation in discrete units.

Shimomura, Osamu – received Nobel Prize for the discovery and development of GFP.

Tsien, Roger – received Nobel Prize for the discovery and development of GFP.

Wilmut, Ian – Created Dolly, the first mammalian clone from an adult animal.