**Chapter 1: Overview of Genetics**

***Key Terms***

Alleles

Amino acid

Behavioral traits

Biological evolution

Carbohydrates

Cellular level

Chromosomes

Deoxyribonucleic acid (DNA)

Diploid

Discovery-based science

Environment

Enzymes

Evolution

Gametes

Gene

Gene expression

Gene mutations

Genetic approach

Genetic code

Genetic cross

Genetic variation

Genome

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

Organismal level

Phenylketonuria (PKU)

Physiological traits

Proteins

Population level

Proteome

Ribonucleic acid (RNA)

Scientific method

Species

Somatic Cells

Traits

Transcription

Translated (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.

a. human genome contains approximately 3 billion nucleotide base pairs (Figure 1.1)

b. project is 99.99% accurate (1 error in 10,000 nucleotides)

2. Human Genome Project should enable scientists to:

a. determine the number of genes in humans

b. examine the relationship between genes and living cells

c. study the evolution of the 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 result in developments that affect our daily lives.

a. The development of new medicines such as Humulin. Humulin is human insulin manufactured by *E. coli* bacteria.

b. DNA fingerprinting for forensic studies

c. Cloning of mammals, such as Dolly the sheep by Ian Wilmut and associates (1997) (Figure 1.2)

d. production of transgenic organisms (Figure 1.3)

*1.1 The Molecular Expression of Genes*

***Learning Outcomes:***

1. Describe the biochemical composition of cells.

2. Explain how proteins are largely responsible for cell structure and function.

3. Outline how DNA stores the information to make proteins.

1.Genes represent the basic unit of heredity, while a trait represents the characteristics of an organism. Genes provide the blueprint that determines an organism’s traits.

Living Cells Are Composed of Biochemicals

1. Small organic molecules provide energy for cellular functions as well as the building blocks for larger molecules.

2. There are Four categories of biologically important organic molecules that are made of repetitive subunits. These organic molecules are called macromolecules.

a. nucleic acids

b. proteins

c. carbohydrates

d. lipids

3. Larger cellular structures, such as chromosomes, are built from combinations of macromolecules and smaller molecules (Figure 1.4).

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

1. Cellular characteristics are determined primarily by proteins.

2. A cell’s proteome consists of all of the proteins that a cell makes at a given time.

3. General roles of proteins include: enzymatic functions, cellular support, transport across the cell membrane, biological motors, cell-to-cell recognition, and cell signaling.

4. Most enzymes are proteins that accelerate a chemical reaction (catalyst).

a. catabolic enzymes break down molecules and release energy

b. anabolic enzymes synthesize larger molecules

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 nitrogenous base, which is either adenine (A), thymine (T), cytosine (C), or guanine (G).

3. The information in the DNA contains the information the sequence of amino acids in a protein.

a. the genetic code is used to relate the genetic information to the correct amino acid

4. In cells, DNA is found in chromosomes (Figure 1.5). The information on a chromosome is organized as genes.

a. on average, a human chromosome contains approximately1000 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 affect the characteristics of cells.

2. Process includes two steps (Figure 1.6):

a. information in the DNA is copied into RNA by transcription (genes that contain the information for the synthesis of proteins are copied into messenger RNA (mRNA)).

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*

***Learning Outcomes:***

1. Explain how the expression of genes leads to an organism’s traits.

2. Define genetic variation.

3. Discuss the relationship between genes and traits.

4. Describe how genes are transmitted in sexually reproducing species.

5. Outline the process of evolution.

The Molecular Expression of Genes Within Cells Leads to an Organism’s 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.

5. 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. organismal level – the observed traits of an organism

d. population level – the occurrence of the trait in a population under given conditions

6. Alternate forms of a gene are called alleles. Different alleles of a gene have different DNA sequences.

Inherited Differences in Traits Are Due to Genetic Variation

1. Genetic variation is the differences in inherited traits among individuals of a population (Figure 1.8).

2. For species that occupy wide geographic ranges, these differences may be drastic. These are very distinct forms of a single species are called morphs.

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 in an organism

Traits Are Governed by Genes and by the Environment

1. The external environment in which an organism exists may influence that organism’s traits.

2. The term 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 hydrolase. This gene allows for the metabolism of the phenylalanine amino acid.

a. Mutations causing a defect in this gene mean that toxic levels of phenylalanine accumulate in the blood, causing mental impairment and developmental problems.

b. The defective allele is present in 1 in 8,000 individuals in the U.S.

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).

3. The copies of each chromosome are called homologs (Figure 1.10a).

4. Gametes are haploid (one copy of each chromosome, also known as the n number) (Figure 1.10b).

5. Sexual reproduction increases genetic variation in a population.

6. Somatic cells, also known as body cells, are diploid in a diploid organism.

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, or simply evolution.

2. Charles Darwin proposed the theory of natural selection as the mechanism for biological evolution.

3. Over a long period of time, the accumulation of many genetic changes may lead to rather striking modifications in a species’ characteristics (Figure 1.11).

*1.3Fields of Genetics*

***Learning Outcomes:***

1. Compare and contrast the three major fields of genetics: transmission, molecular, and population genetics.

2. Discuss how genetics is an experimental science.

1. Genetics is a broad field encompassing molecular, cellular, organism, and population biology.

2. Experimentally, geneticists often focus their efforts on 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.

3. Figure 1.12 shows some common model genetic organisms.

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 controlled genetic crosses and pedigree analyses to examine how traits are passed from parents to offspring.

Molecular Genetics Focuses on a Biochemical Understanding of the Hereditary Material

1. 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. These loss of function mutations help geneticists understand the normal 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 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.

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.