Biology for Non-Majors I

Lumen Learning, Houston Community College

CONTENTS

Course Contents 7
- About this Course 7
- Course Contents at a Glance 8
- Learning Outcomes 10

Module 1: Introduction to Biology 14
- Why It Matters: Introduction to Biology 14
- Introduction to Characteristics of Life 15
- Properties of Life 15
- Levels of Organization of Living Things 18
- Introduction to Taxonomy 21
- The Diversity of Life 21
- Phylogenetic Trees 24
- Taxonomy 27
- Introduction to the Branches of Biology 29
- The Branches of Biology 29
- Introduction to the Process of Science 31
- Scientific Inquiry 32
- Basic and Applied Science 36
- Summary: The Process of Science 38
- Putting It Together: Introduction to Biology 39

Module 2: Chemistry of Life 41
- Why It Matters: The Chemistry of Life 41
- Introduction to Atoms and Elements 42
- Elements in Biological Matter 42
- Atoms 43
- Properties of Elements 44
- Isotopes 47
- Introduction to Atomic Bonds 48
- Chemical Bonding 48
- Ionic Bonds 51
- Covalent Bonds 52
- Weaker Bonds in Biology 55
- Why Life Depends on Water 57
- Introduction to the pH Scale 61
- Buffers, pH, Acids, and Bases 61
- Putting It Together: Chemistry of Life 63

Module 3: Important Biological Macromolecules 65
- Why It Matters: Important Biological Macromolecules 65
- Introduction to Carbon 66
- Carbon and Carbon Bonding 66
- Introduction to Carbohydrates 68
- Structure and Function of Carbohydrates 68
- Introduction to Lipids 75
- Lipids 75
- Introduction to Proteins 80
- Components of Proteins 81
- Protein Structure 82
- Function of Proteins 85
- Introduction to Nucleic Acids 86
- Structure of Nucleic Acids 87

- DNA and RNA 89
- Introduction to Comparing Biological Macromolecules 90
- Different Types of Biological Macromolecules 91
- Putting It Together: Important Biological Macromolecules 92

Module 4: Cellular Structure 93
- Why It Matters: Cellular Structure 93
- Introduction to Cell Theory 93
- Cell Theory 94
- Introduction to Prokaryotes and Eukaryotes 95
- Comparing Prokaryotic and Eukaryotic Cells 96
- Eukaryotic Origins 98
- Introduction to Organelles 104
- Cytoplasm 105
- The Endomembrane System 105
- Ribosomes, Mitochondria, Vesicles, and Peroxisomes 109
- The Cytoskeleton, Flagella and Cilia, and the Plasma Membrane 111
- Animal Cells versus Plant Cells 113
- Summary: Organelles 119
- Putting It Together: Cellular Structure 121

Module 5: Cell Membranes 124
- Why It Matters: Cell Membranes 124
- Introduction to Structure of the Membrane 124
- Structure of the Cell Membrane 125
- Introduction to Kinds of Transport 128
- Passive Transport 128
- Active Transport 135
- Membranes and Transport 140
- Introduction to Endocytosis and Exocytosis 140
- Endocytosis 141
- Exocytosis 144
- Putting It Together: Cell Membranes 146

Module 6: Metabolic Pathways 148
- Why It Matters: Metabolic Pathways 148
- Introduction to Energy and Metabolism 148
- Metabolic Pathways 149
- Thermodynamics 151
- Energy 154
- Enzymes 156
- Summary: Energy and Metabolism 159
- Introduction to ATP in Living Systems 161
- ATP in Living Systems 162
- Introduction to Cellular Respiration 165
- Glycolysis 166
- Citric Acid Cycle and Oxidative Phosphorylation 169
- Summary: Cellular Respiration 173
- Introduction to Fermentation 175
- Types of Fermentation 175
- Introduction to Photosynthesis 180
- An Overview of Photosynthesis 181
- Light Energy 187
- The Light-Dependent Reactions of Photosynthesis 190
- The Calvin Cycle 193
- Summary: Photosynthesis 198

- Introduction to Connections to Other Metabolic Pathways 198
- Connections to Other Metabolic Pathways 199
- The Energy Cycle 201
- Putting It Together: Metabolic Pathways 203

Module 7: Cell Division 204
- Why It Matters: Cell Division 204
- Introduction to Chromosomes and DNA Packaging 205
- DNA and Chromosomes 205
- Chromosome Structure 207
- Introduction to the Cell Cycle 210
- Interphase 211
- Mitosis 212
- Cytokinesis 214
- The Complete Cell Cycle 215
- Introduction to Cell Cycle Checkpoints 216
- Control of the Cell Cycle 217
- Cancer and the Cell Cycle 219
- Introduction to Sexual Reproduction 220
- Sexual Reproduction 220
- Introduction to Meiosis 225
- Stages of Meiosis 225
- Meiosis I 226
- Meiosis II 231
- Meiosis: The Complete Cycle 234
- Introduction to Genetic Diversity 234
- Genetic Variation in Meiosis 235
- Mitosis, Meiosis, and Sexual Reproduction 236
- Introduction to Errors in Chromosome Number 237
- Karyotypes 237
- Common Disorders 239
- Putting It Together: Cell Division 244

Module 8: DNA Structure and Replication 246
- Why It Matters: DNA Structure and Replication 246
- Introduction to Storing Genetic Information 247
- Structure of DNA 248
- Genetic Information 250
- Introduction to DNA Replication 253
- Basics of DNA Replication 253
- Major Enzymes 256
- Proofreading DNA 261
- Introduction to Virus Replication 263
- Viral Morphology 263
- Viral Infectious Cycles 266
- Prions and Viroids 270
- Putting It Together: DNA Structure and Replication 271

Module 9: DNA Transcription and Translation 273
- Why It Matters: DNA Transcription and Translation 273
- Introduction to Transcription 274
- Steps of Transcription 274
- pre-RNA and mRNA 277
- Introduction to Translation 278
- Requirements for Translation 279
- Genetic Code 281

- Steps of Translation 282
- Introduction to the Central Dogma 285
- The Central Dogma 286
- Introduction to DNA Mutations 286
- What is a Mutation? 287
- Major Types of Mutations 288
- Putting It Together: DNA Transcription and Translation 290

Module 10: Gene Expression 291
- Why It Matters: Gene Expression 291
- Introduction to Regulation of Gene Expression 291
- Expression of Genes 292
- Prokaryotic and Eukaryotic Gene Regulation 295
- Introduction to Prokaryotic Gene Regulation 297
- Gene Regulation in Prokaryotes 298
- Introduction to Eukaryotic Gene Regulation 300
- Eukaryotic Epigenetic Gene Regulation 300
- Eukaryotic Transcription Gene Regulation 303
- Post-Translational Control of Gene Expression 306
- Putting It Together: Gene Expression 308

Module 11: Trait Inheritance 310
- Why It Matters: Trait Inheritance 310
- Introduction to the Father of Genetics 311
- Mendel's Experiments and Heredity 311
- Characteristics and Traits 315
- Laws of Inheritance 322
- Heredity 328
- Introduction to Beyond Dominance and Recessiveness 329
- Non-Mendelian Inheritance 329
- Non-Mendelian Punnett Squares 334
- Multiple Alleles 335
- Introduction to Heredity and Disease 337
- Pedigrees and Disease 338
- Genetic Disorder and Pedigrees 341
- Polygenic Inheritance and Environmental Effects 341
- Introduction to Genetics and the Environment 343
- Effect of the Environment 343
- Pleiotropy and Human Disorders 345
- Putting It Together: Trait Inheritance 346

Module 12: Theory of Evolution 348
- Why It Matters: Theory of Evolution 348
- Introduction to Charles Darwin 349
- Darwin and Descent with Modification 349
- Darwin and the Theory of Evolution 352
- Introduction to Evidence for Evolution 354
- Physical Evidence 354
- Biological Evidence 357
- Misconceptions of Evolution 358
- Introduction to Mutations and Evolution 360
- Population Genetics 361
- Selective and Environmental Pressures 362
- Genetic Variation and Drift 369
- Introduction to Phylogenetic Trees 372
- Scientific Classification 373

- Structure of Phylogenetic Trees 374
- Limitations of Phylogenetic Trees 375
- The Taxonomic Classification System 376
- Putting It Together: Theory of Evolution 380

Module 13: Modern Biology 382
- Why It Matters: Modern Biology 382
- Introduction to Key Technologies 382
- Manipulating Genetic Material 383
- DNA Sequencing 388
- Cloning 390
- Genetic Engineering 396
- Introduction to Biotechnology Applications 397
- Medicinal Biotechnology 397
- Agricultural Biotechnology 399
- Introduction to Risks and Benefits of Genomic Science 401
- Genetic Information Used for Identification 401
- Medical Uses of Genetic Information 402
- Genomics in Agriculture 404
- Putting It Together: Modern Biology 405

COURSE CONTENTS

ABOUT THIS COURSE

Biology for Non-Majors I introduces students to the basics of the scientific process, the chemical foundations of life, cell structure and function, photosynthesis, cellular respiration, genetics and inheritance, and evolution. Designed for non-life science majors, this course is the first in a two-part series that completes a survey of biological principles.

This course was developed by Lumen Learning, with contributing work from Veronica Amaku and Renu Jain of Houston Community College, with additional work by Shelli Carter. Primary sources for course materials include the OpenStax textbooks, Concepts of Biology and Biology, supplemented with relevant materials from Khan Academy and videos from multiple sources. Original practice activities were authored by Shelli Carter and Lumen Learning in the development of this course.

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COURSE CONTENTS AT A GLANCE

The following list shows a summary of the topics covered in this course. To see all of the course pages, visit the Table of Contents.

Module 1: Introduction to Biology

- Characteristics of Life
- Taxonomy
- The Branches of Biology
- The Process of Science

Module 2: Chemistry of Life

- Atoms and Elements
- Atomic Bonds
- The pH Scale

Module 3: Important Biological Macromolecules

- Carbon
- Carbohydrates
- Lipids
- Proteins
- Nucleic Acids
- Comparing Biological Macromolecules

Module 4: Cellular Structure

- Cell Theory
- Prokaryotes and Eukaryotes
- Organelles

Module 5: Cell Membranes

- Structure of the Membrane
- Kinds of Transport
- Endocytosis and Exocytosis

Module 6: Metabolic Pathways

- Energy and Metabolism
- ATP in Living Systems
- Cellular Respiration
- Fermentation
- Photosynthesis
- Connections to Other Metabolic Pathways

Module 7: Cell Division

- Chromosomes and DNA Packaging
- The Cell Cycle
- Cell Cycle Checkpoints
- Sexual Reproduction
- Meiosis
- Genetic Diversity
- Errors in Chromosome Number

Module 8: DNA Structure and Replication

- Storing Genetic Information
- DNA Replication
- Viruses

Module 9: DNA Transcription and Translation

- Transcription
- Translation
- The Central Dogma
- DNA Mutations

Module 10: Gene Expression

- Regulation of Gene Expression
- Prokaryotic Gene Regulation
- Eukaryotic Gene Regulation

Module 11: Trait Inheritance

- The Father of Genetics
- Beyond Dominance and Recessiveness
- Heredity and Disease
- Genetics and the Environment

Module 12: Theory of Evolution

- Charles Darwin
- Evidence for Evolution
- Mutations and Evolution
- Phylogenetic Trees

Module 13: Modern Biology

- Key Technologies
- Biotechnology Applications
- Risks and Benefits of Genomic Science

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LEARNING OUTCOMES

The content, assignments, and assessments for Biology for Non-Majors I are aligned to the following learning outcomes. A full list of course learning outcomes can be viewed here: Biology for Non-Majors I Learning Outcomes.

Module 1: Introduction to Biology

Define biology and apply its principles

- List the defining characteristics of biological life
- Describe classification and organizational tools biologists use, including modern taxonomy
- Identify the main branches of biology
- Describe biology as a science and identify the key components of scientific inquiry

Module 2: Chemistry of Life

Identify the principles of chemistry that are integral to biology

- Define atoms and elements
- Classify different types of atomic bonds
- Demonstrate familiarity with the pH scale

Module 3: Important Biological Macromolecules

Identify and describe the main features of the four main classes of important biological macromolecules

- Discuss why it is said that life is carbon-based and the bonding properties of carbon
- Summarize the roles carbohydrates play in biological systems
- Illustrate different types of lipids and relate their structure to their role in biological systems
- Describe the structure and function of proteins
- Discuss nucleic acids and the role they play in DNA and RNA
- Discuss macromolecules and the differences between the four classes

Module 4: Cellular Structure

Identify and explain a variety of cellular components

- State the basic principles of the unified cell theory
- Compare prokaryotes and eukaryotes
- Identify membrane-bound organelles found in eukaryotic cells

Module 5: Cell Membranes

Describe and explain the structure and function of membranes

- Describe the structure and function of membranes, especially the phospholipid bilayer
- Explain how substances are directly transported across a membrane
- Describe the primary mechanisms by which cells import and export macromolecules

Module 6: Metabolic Pathways

Explain the metabolic pathways involved in the capture and release of energy in cells

- Discuss energy and metabolism in living things
- Describe how cells store and transfer free energy using ATP
- Identify the reactants and products of cellular respiration and where these reactions occur in a cell
- Illustrate the basic components and steps of fermentation
- Identify the basic components and steps of photosynthesis
- Discuss the connections between metabolic pathways

Module 7: Cell Division

Describe and explain the various stages of cell division

- Understand chromosome structure and organization in eukaryotic cells
- Identify the stages of the cell cycle, by picture and by description of major milestones
- Identify and explain the important checkpoints that a cell passes through during the cell cycle
- Understand how sexual reproduction leads to different sexual life cycles
- Identify the stages of meiosis by picture and by description of major milestones; explain why meiosis involves two rounds of nuclear division

- Describe and explain a range of mechanisms for generating genetic diversity
- Examine karyotypes and identify the effects of significant changes in chromosome number

Module 8: DNA Structure and Replication

Relate DNA structure to the process of DNA replication

- Explain how DNA stores genetic information
- Explain the role of complementary base pairing in the precise replication process of DNA
- Identify different viruses and how they replicate

Module 9: DNA Transcription and Translation

Describe the conversion of DNA to RNA to proteins

- Outline the process of transcription
- Summarize the process of translation
- Identify the central dogma of life
- Recognize the impact of DNA mutations

Module 10: Gene Expression

Explain the regulation of gene expression

- Define the term regulation as it applies to genes
- Understand the basic steps in gene regulation in prokaryotic cells
- Discuss different components and types of epigenetic gene regulation

Module 11: Trait Inheritance

Complete monohybrid and dihybrid crosses and family pedigrees, and explain the inheritance of various traits

- Identify the impact of Gregor Mendel on the field of genetics and apply Mendel's two laws of genetics
- Explain complications to the phenotypic expression of genotype, including mutations
- Explain the conventions of a family pedigree and predict whether a disease will be passed through a family in one of three modes
- Discuss the role environment plays on phenotypes

Module 12: Theory of Evolution

Explain the theory of evolution, which documents the change in the genetic makeup of a biological population over time

- Describe the work of Charles Darwin in the Galapagos Islands, especially his discovery of natural selection in finch populations
- Describe how the theory of evolution by natural selection is supported by evidence
- Recognize that mutations are the basis of microevolution; and that adaptations enhance the survival and reproduction of individuals in a population

- Read and analyze a phylogenetic tree that documents evolutionary relationships

Module 13: Modern Biology

Describe and discuss techniques used in modern biology

- List key technologies enabling modern uses of biology
- Identify societal uses of biotechnology
- Discuss the risks and benefits involved in applications of genetic and genomic science

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MODULE 1: INTRODUCTION TO BIOLOGY

WHY IT MATTERS: INTRODUCTION TO BIOLOGY

Why learn about biology and its principles?

One night while she was scrolling through her social media feeds, Cristina saw that her brother had linked to an article about some of the world's weirdest animals. As she paged through the article, Cristina became increasingly interested in the different features these animals had: some were eyeless, some were colorless, and others had even stranger features.

Figure 1. The pangolin (also known as the scaly anteater) is a unique animal. It walks on just its hind legs and uses its front claws to tear open termite mounds.

Before Cristina could dig deeper into these animals, she got a message from her cousin Samuel. He'd sent a link to an article about genetically modified foods and the dangers they inherently contain. Cristina was only halfway through reading the first paragraph of the article when Samuel sent her another article: this one lauding the paleo diet and its benefits. Cristina started to read the article, but before she got too far, she remembered that she had a paper due the next day. She made a mental note to come back to the articles from her cousin, and she bookmarked the animals article.

Though Cristina might not realize it, she's just been presented with three different biological questions. How did these animals develop such unique characteristics? Are GMOs dangerous? Are extreme diets (like the paleo diet) beneficial?

Cristina still has to come to her own conclusions and make her own choices, but having an understanding of biology will help her make the best choices she can.

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INTRODUCTION TO CHARACTERISTICS OF LIFE

What you'll learn to do: List the defining characteristics of biological life

Biology is the science that studies life, but what exactly is life? This may sound like a silly question with an obvious response, but it is not always easy to define life. For example, a branch of biology called virology studies viruses, which exhibit some of the characteristics of living entities but lack others. It turns out that although viruses can attack living organisms, cause diseases, and even reproduce, they do not meet the criteria that biologists use to define life. Consequently, virologists are not biologists, strictly speaking. Similarly, some biologists study the early molecular evolution that gave rise to life; since the events that preceded life are not biological events, these scientists are also excluded from biology in the strict sense of the term.

From its earliest beginnings, biology has wrestled with these questions: What are the shared properties that make something "alive"? And once we know something is alive, how do we find meaningful levels of organization in its structure?

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PROPERTIES OF LIFE

Learning Outcomes

List the properties of life

All living organisms share several key characteristics or functions: order, sensitivity or response to the environment, reproduction, growth and development, regulation, homeostasis, and energy processing. When viewed together, these characteristics serve to define life.

Order

Organisms are highly organized, coordinated structures that consist of one or more cells. Even very simple, single-celled organisms are remarkably complex: inside each cell, atoms make up molecules; these in turn make up cell organelles and other cellular inclusions.

In multicellular organisms (Figure 1), similar cells form tissues. Tissues, in turn, collaborate to create organs (body structures with a distinct function). Organs work together to form organ systems.

Figure 1. This female monarch butterfly represents a highly organized structure consisting of cells, tissues, organs, and organ systems

Sensitivity or Response to Stimuli

Organisms respond to diverse stimuli. For example, plants can bend toward a source of light, climb on fences and walls, or respond to touch (Figure 2). Even tiny bacteria can move toward or away from chemicals (a process called *chemotaxis*) or light (*phototaxis*). Movement toward a stimulus is considered a positive response, while movement away from a stimulus is considered a negative response.

Watch this video to see how plants respond to a stimulus—from opening to light, to wrapping a tendril around a branch, to capturing prey.

Figure 2.The leaves of this sensitive plant (Mimosa pudica) will instantly droop and fold when touched. After a few minutes, the plant returns to normal.

Reproduction

Single-celled organisms reproduce by first duplicating their DNA, and then dividing it equally as the cell prepares to divide to form two new cells. Multicellular organisms often produce specialized reproductive germline cells that will form new individuals. When reproduction occurs, genes containing DNA are passed along to an organism's offspring. These genes ensure that the offspring will belong to the same species and will have similar characteristics, such as size and shape.

Growth and Development

Organisms grow and develop following specific instructions coded for by their genes. These genes provide instructions that will direct cellular growth and development, ensuring that a species' young (Figure 3) will grow up to exhibit many of the same characteristics as its parents.

Figure 3. Although no two look alike, these puppies have inherited genes from both parents and share many of the same characteristics.

Regulation

Even the smallest organisms are complex and require multiple regulatory mechanisms to coordinate internal functions, respond to stimuli, and cope with environmental stresses. Two examples of internal functions regulated in an organism are nutrient transport and blood flow. Organs (groups of tissues working together) perform specific functions, such as carrying oxygen throughout the body, removing wastes, delivering nutrients to every cell, and cooling the body.

Homeostasis

In order to function properly, cells need to have appropriate conditions such as proper temperature, pH, and appropriate concentration of diverse chemicals. These conditions may, however, change from one moment to the next. Organisms are able to maintain internal conditions within a narrow range almost constantly, despite environmental changes, through homeostasis (literally, "steady state")—the ability of an organism to maintain constant internal conditions. For example, an organism needs to regulate body temperature through a process known as thermoregulation. Organisms that live in cold climates, such as the polar bear (Figure 4), have body structures that help them withstand low temperatures and conserve body heat. Structures that aid in this type of insulation include fur, feathers, blubber, and fat. In hot climates, organisms have methods (such as perspiration in humans or panting in dogs) that help them to shed excess body heat.

Figure 4. Polar bears (Ursus maritimus) and other mammals living in ice-covered regions maintain their body temperature by generating heat and reducing heat loss through thick fur and a dense layer of fat under their skin.

Energy Processing

All organisms use a source of energy for their metabolic activities. Some organisms capture energy from the sun and convert it into chemical energy in food (photosynthesis); others use chemical energy in molecules they take in as food (cellular respiration).

Figure 5. The California condor (Gymnogyps californianus) uses chemical energy derived from food to power flight. California condors are an endangered species; this bird has a wing tag that helps biologists identify the individual.

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- Young California condor (Gymnogyps californianus) ready for flight. **Provided by**: US Fish and Wildlife Service. **Located at**: https://en.wikipedia.org/wiki/File:California-condor.jpg. **License**: *Public Domain: No Known Copyright*

LEVELS OF ORGANIZATION OF LIVING THINGS

Learning outcomes

Order the levels of organization of living things

Living things are highly organized and structured, following a hierarchy that can be examined on a scale from small to large. The **atom** is the smallest and most fundamental unit of matter. It consists of a nucleus surrounded by electrons. Two or more atoms are joined together by one or more chemical bonds to form **molecule.** Many molecules that are biologically important are **macromolecules**, large molecules that are typically formed by polymerization (a polymer is a large molecule that is made by combining smaller units called monomers, which are simpler than macromolecules). An example of a macromolecule is deoxyribonucleic acid (DNA) (Figure 1), which contains the instructions for the structure and functioning of all living organisms.

Some cells contain aggregates of macromolecules surrounded by membranes; these are called **organelles**. Organelles are small structures that exist within cells. Examples of organelles include mitochondria and chloroplasts, which carry out indispensable functions: mitochondria produce energy to power the cell, while chloroplasts enable green plants to utilize the energy in sunlight to make sugars. All living things are made of cells; the **cell** itself is the smallest fundamental unit of structure and function in living organisms. (This requirement is why viruses are not considered living: they are not made of cells. To make new viruses, they have to invade and hijack the reproductive mechanism of a living cell; only then can they obtain the materials they need to reproduce.) Some organisms consist of a single cell and others are multicellular. Cells are classified as prokaryotic or eukaryotic. **Prokaryotes** are single-celled or colonial organisms that do not have membrane-bound nuclei or organelles; in contrast, the cells of **eukaryotes** do have membrane-bound organelles and a membrane-bound nucleus.

In most multicellular organisms, cells combine to make **tissues**, which are groups of similar cells carrying out similar or related functions. **Organs** are collections of tissues grouped together performing a common function. Organs are present not only in animals but also in plants. An **organ system** is a higher level of organization that consists of functionally related organs. Mammals have many organ systems. For instance, the circulatory system transports blood through the body and to and from the lungs; it includes organs such as the heart and blood vessels. **Organisms** are individual living entities. For example, each tree in a forest is an organism. Single-celled prokaryotes and single-celled eukaryotes are also considered organisms and are typically referred to as microorganisms.

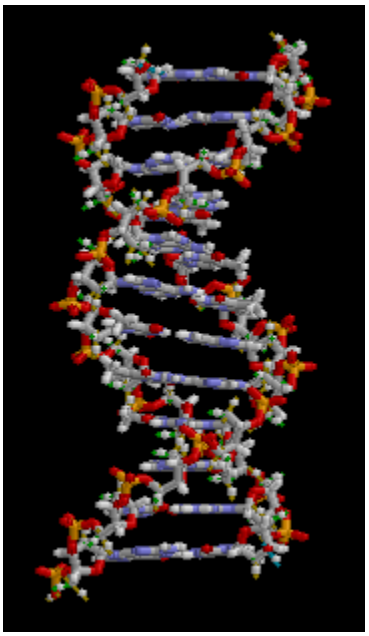

Figure 1. All molecules, including this DNA molecule, are composed of atoms. (credit: "brian0918"/Wikimedia Commons)

All the individuals of a species living within a specific area are collectively called a **population**. For example, a forest may include many pine trees. All of these pine trees represent the population of pine trees in this forest. Different populations may live in the same specific area. For example, the forest with the pine trees includes populations of flowering plants and also insects and microbial populations. A **community** is the sum of populations inhabiting a particular area. For instance, all of the trees, flowers, insects, and other populations in a forest form the forest's community. The forest itself is an ecosystem. An **ecosystem** consists of all the living things in a particular area together with the abiotic, non-living parts of that environment such as nitrogen in the soil or rain water. At the highest level of organization (Figure 2), the **biosphere** is the collection of all ecosystems, and it represents the zones of life on earth. It includes land, water, and even the atmosphere to a certain extent.

Practice Question

From a single organelle to the entire biosphere, living organisms are parts of a highly structured hierarchy.

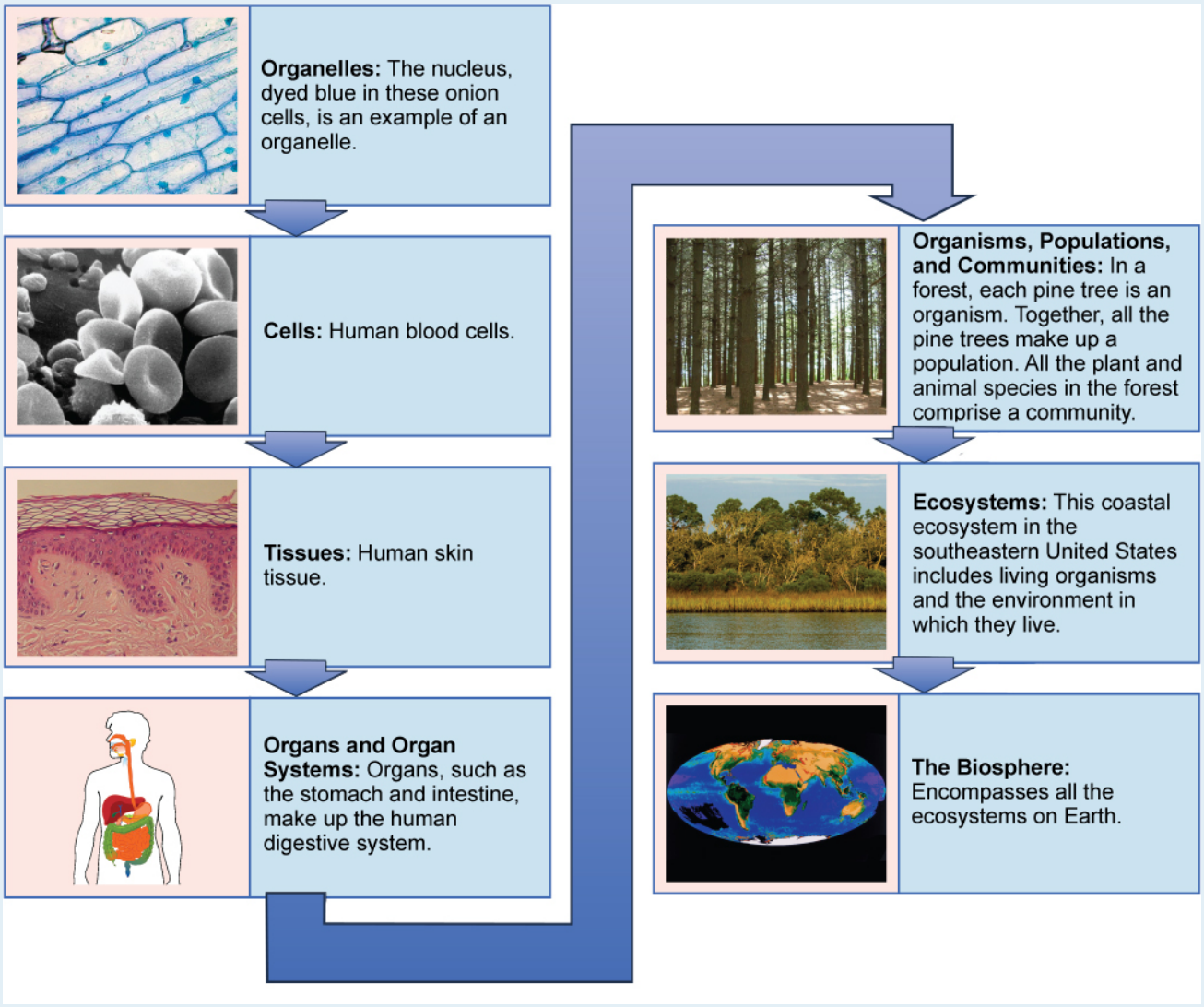

Figure 2. The biological levels of organization of living things are shown. From a single organelle to the entire biosphere, living organisms are parts of a highly structured hierarchy. (credit "organelles": modification of work by Umberto Salvagnin; credit "cells": modification of work by Bruce Wetzel, Harry Schaefer/ National Cancer Institute; credit "tissues": modification of work by Kilbad; Fama Clamosa; Mikael Häggström; credit "organs": modification of work by Mariana Ruiz Villareal; credit "organisms": modification of work by "Crystal"/Flickr; credit "ecosystems": modification of work by US Fish and Wildlife Service Headquarters; credit "biosphere": modification of work by NASA)

Which of the following statements is false?

a. Tissues exist within organs, which exist within organ systems.
b. Communities exist within populations, which exist within ecosystems.
c. Organelles exist within cells, which exist within tissues.
d. Communities exist within ecosystems, which exist in the biosphere.

Answer

Statement b is false: populations exist within communities.

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INTRODUCTION TO TAXONOMY

What you'll learn to do: Describe classification and organizational tools biologists use, including modern taxonomy

Viewed from space, Earth offers no clues about the diversity of life forms that reside there. The first forms of life on Earth are thought to have been microorganisms that existed for billions of years in the ocean before plants and animals appeared. The mammals, birds, and flowers so familiar to us are all relatively recent, originating 130 to 200 million years ago. Humans have inhabited this planet for only the last 2.5 million years, and only in the last 200,000 years have humans started looking like we do today.

When faced with the remarkable diversity of life, how do we organize the different kinds of organisms so that we can better understand them? As new organisms are discovered every day, biologists continue to seek answers to these and other questions. In this outcome, we will discuss *taxonomy*, which both demonstrates the vast diversity of life and tries to organize these organisms in a way we can understand.

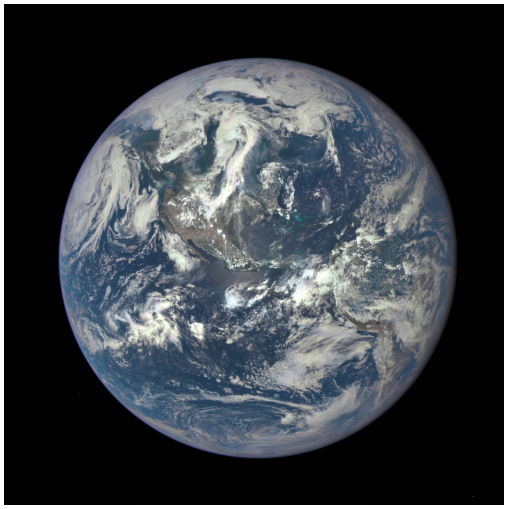

Figure 1. Our planet

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THE DIVERSITY OF LIFE

Learning Outcomes

Explain the diversity of life

Biological diversity is the variety of life on earth. This includes all the different plants, animals, and microorganisms; the genes they contain; and the ecosystems they form on land and in water. Biological diversity is constantly changing. It is increased by new genetic variation and reduced by extinction and habitat degradation.

Figure 1. Life on earth is incredibly diverse.

What Is Biodiversity?

Biodiversity refers to the variety of life and its processes, including the variety of living organisms, the genetic differences among them, and the communities and ecosystems in which they occur. Scientists have identified about 1.9 million species alive today. They are divided into the six kingdoms of life shown in Figure 2. Scientists are still discovering new species. Thus, they do not know for sure how many species really exist today. Most estimates range from 5 to 30 million species.

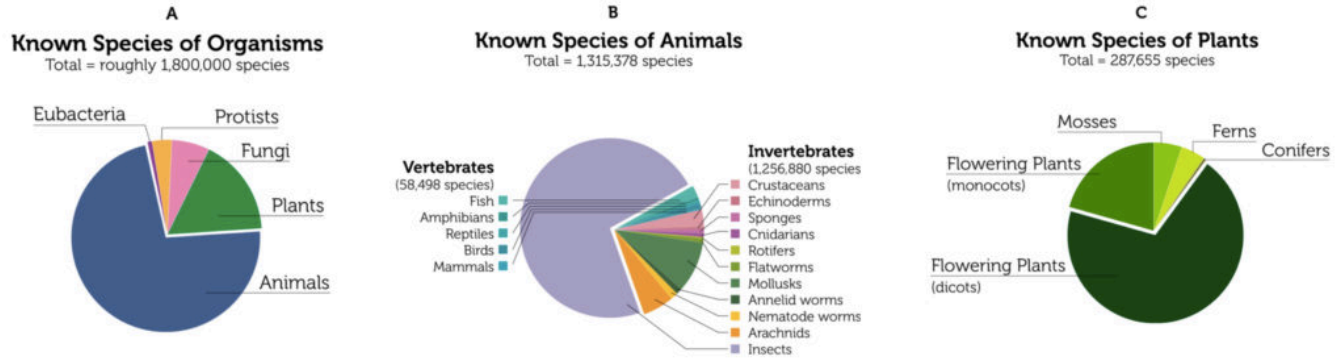

Figure 2. Known life on earth. Click for a larger image.

Video Review

Watch this discussion about biodiversity:

Watch this video online: https://youtu.be/vGxJArebKoc

Scale of Biodiversity

Diversity may be measured at different scales. These are three indices used by ecologists:

- **Alpha diversity** refers to diversity within a particular area, community or ecosystem, and is measured by counting the number of taxa within the ecosystem (usually species).
- **Beta diversity** is species diversity between ecosystems; this involves comparing the number of taxa that are unique to each of the ecosystems.
- **Gamma diversity** is a measurement of the overall diversity for different ecosystems within a region.

Benefits of Biodiversity

Biodiversity provides us with all of our food. It also provides for many medicines and industrial products, and it has great potential for developing new and improved products for the future. Perhaps most importantly, biological diversity provides and maintains a wide array of ecological "services." These include provision of clean air and water, soil, food and shelter. The quality—and the continuation— of our life and our economy is dependent on these "services."

Australia's Biological Diversity

The long isolation of Australia over much of the last 50 million years and its northward movement have led to the evolution of a distinct biota. Significant features of Australia's biological diversity include:

- A high percentage of endemic species (that is, they occur nowhere else):
  - over 80% of flowering plants
  - over 80% of land mammals
  - 88% of reptiles
  - 45% of birds
  - 92% of frogs
- Wildlife groups of great richness. Australia has an exceptional diversity of lizards in the arid zone, many ground orchids, and a total invertebrate fauna estimated at 200,000 species with more than 4,000 different species of ants alone. Marsupials and monotremes collectively account for about 56% of native terrestrial mammals in Australia.
- Wildlife of major evolutionary importance. For example, Australia has 12 of the 19 known families of primitive flowering plants, two of which occur nowhere else. Some species, such as the Queensland lungfish and peripatus, have remained relatively unchanged for hundreds of millions of years.

Figure 3. The short-beaked echidna is endemic to Australia. This animal—along with the platypus and three other species of echidnas—is one of the five surviving species of egg-laying mammals.

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PHYLOGENETIC TREES

Learning Outcomes

Explain the purpose of phylogenetic trees

In scientific terms, the evolutionary history and relationship of an organism or group of organisms is called phylogeny. **Phylogeny** describes the relationships of one organism to others—such as which organisms it is thought to have evolved from, which species it is most closely related to, and so forth. Phylogenetic relationships provide information on shared ancestry but not necessarily on how organisms are similar or different.

Phylogenetic Trees

Scientists use a tool called a phylogenetic tree to show the evolutionary pathways and connections among organisms. A **phylogenetic tree** is a diagram used to reflect evolutionary relationships among organisms or groups of organisms. Scientists consider phylogenetic trees to be a hypothesis of the evolutionary past since one cannot go back to confirm the proposed relationships. In other words, a "tree of life" can be constructed to illustrate when different organisms evolved and to show the relationships among different organisms (Figure 1).

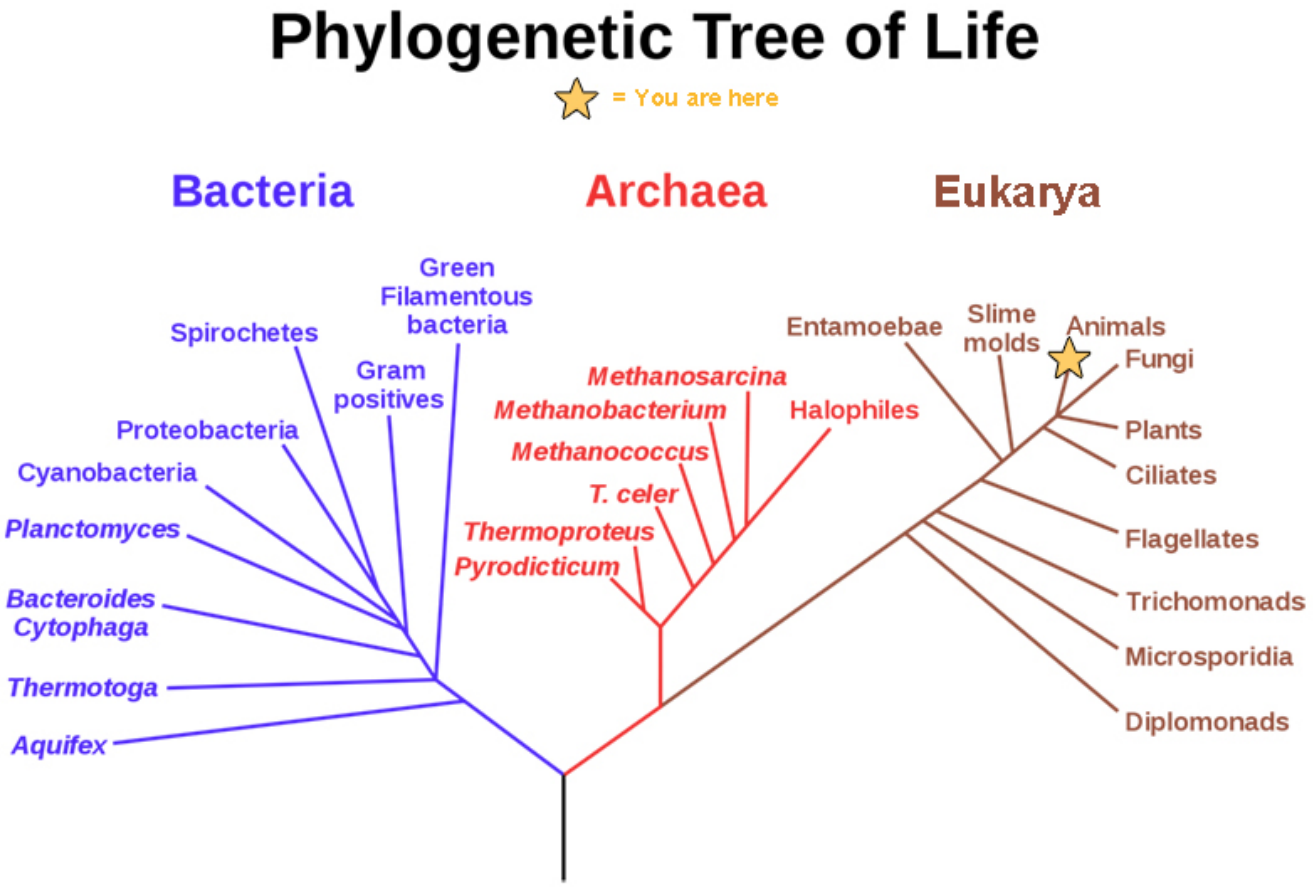


Figure 1. This phylogenetic tree was constructed by microbiologist Carl Woese (See inset below) using genetic relationships. The tree shows the separation of living organisms into three domains: Bacteria, Archaea, and Eukarya. Bacteria and Archaea are

organisms without a nucleus or other organelles surrounded by a membrane and, therefore, are prokaryotes. (credit: modification of work by Eric Gaba)

A phylogenetic tree can be read like a map of evolutionary history. Many phylogenetic trees have a single lineage at the base representing a common ancestor. Scientists call such trees rooted, which means there is a single ancestral lineage (typically drawn from the bottom or left) to which all organisms represented in the diagram relate. Notice in the rooted phylogenetic tree that the three domains—Bacteria, Archaea, and Eukarya—diverge from a single point and branch off. The small branch that plants and animals (including humans) occupy in this diagram shows how recent and minuscule these groups are compared with other organisms. Unrooted trees don't show a common ancestor but do show relationships among species (Figure 2).

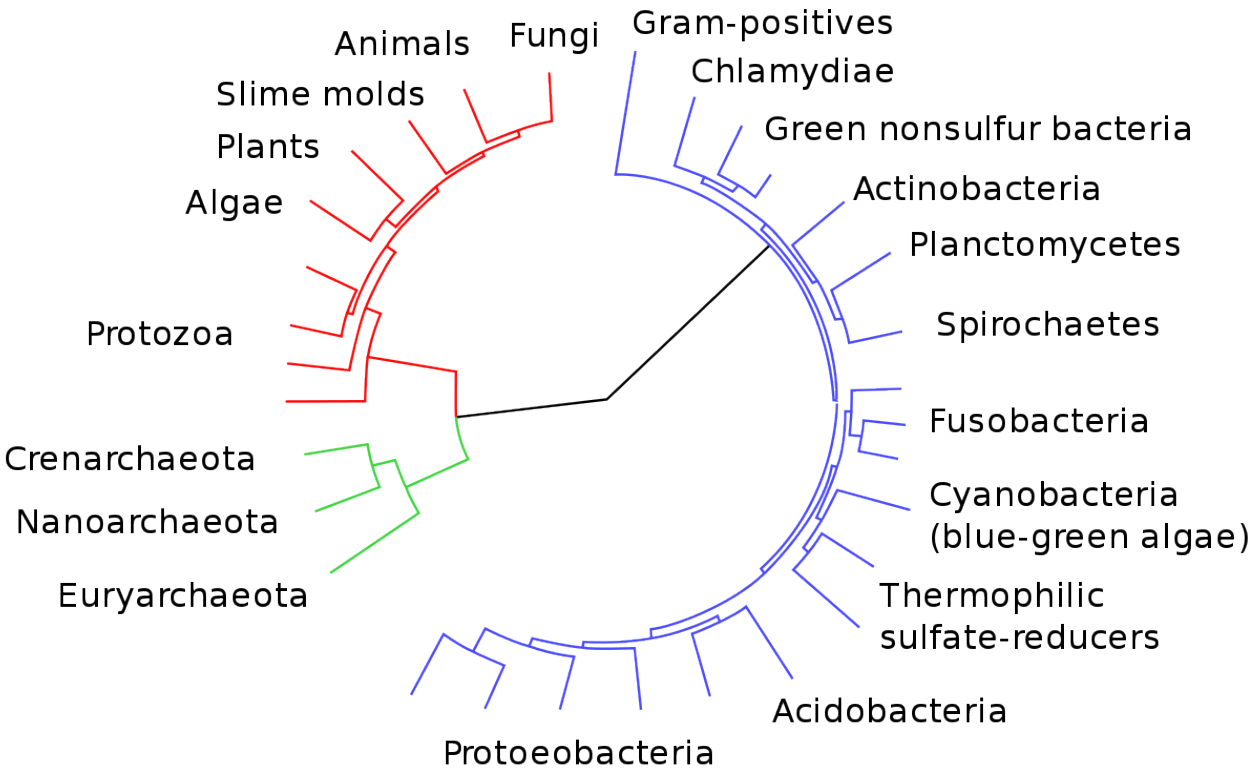
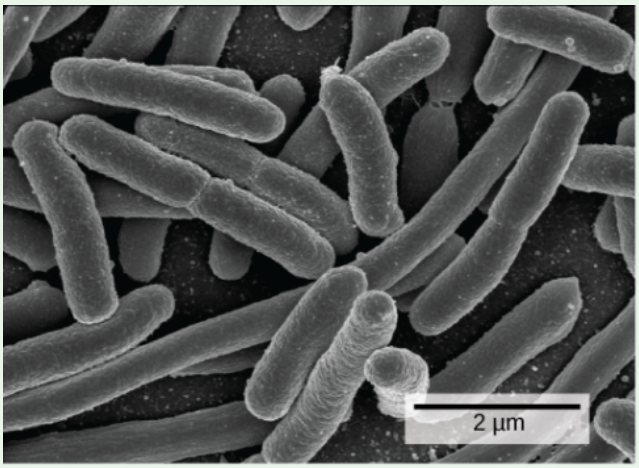


Figure 2. An unrooted phylogenetic tree

Carl Woese and the Phylogenetic Tree

In the past, biologists grouped living organisms into five kingdoms: animals, plants, fungi, protists, and bacteria. The organizational scheme was based mainly on physical features, as opposed to physiology, biochemistry, or molecular biology, all of which are used by modern systematics. The pioneering work of American microbiologist Carl Woese in the early 1970s has shown, however, that life on Earth has evolved along three lineages, now called domains—Bacteria, Archaea, and Eukarya. The first two are prokaryotic groups of microbes that lack membrane-enclosed nuclei and organelles. The third domain contains the eukaryotes and includes unicellular microorganisms together with the four original kingdoms (excluding bacteria). Woese defined Archaea as a new domain, and this resulted in a new taxonomic tree (Figure 1). Many organisms belonging to the Archaea domain live under extreme conditions and are called extremophiles. To construct his tree, Woese used genetic relationships rather than similarities based on morphology (shape).

Woese's tree was constructed from comparative sequencing of the genes that are universally distributed, present in every organism, and conserved (meaning that these genes have remained essentially unchanged throughout evolution). Woese's approach was revolutionary because comparisons of physical features are insufficient to differentiate between the prokaryotes that appear fairly similar in spite of their tremendous biochemical diversity and genetic variability (Figure 3). The comparison of homologous DNA and RNA sequences provided Woese with a sensitive device that revealed the extensive variability of prokaryotes, and which justified the separation of the prokaryotes into two domains: bacteria and archaea.



(a)

(b)

(c)

(d)

Figure 3. These organisms represent different domains. The (a) bacteria in this micrograph belong to Domain Bacteria, while the (b) extremophiles (not visible) living in this hot vent belong to Domain Archaea. Both the (c) sunflower and (d) lion are part of Domain Eukarya. (credit a: modification of work by Drew March; credit b: modification of work by Steve Jurvetson; credit c: modification of work by Michael Arrighi; credit d: modification of work by Leszek Leszcynski)

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TAXONOMY

Learning Outcomes

Explain how relationships are indicated by the binomial naming system

Taxonomy (which literally means “arrangement law”) is the science of classifying organisms to construct internationally shared classification systems with each organism placed into more and more inclusive groupings. Think about how a grocery store is organized. One large space is divided into departments, such as produce, dairy, and meats. Then each department further divides into aisles, then each aisle into categories and brands, and then finally a single product. This organization from larger to smaller, more specific categories is called a hierarchical system.

In the eighteenth century, a scientist named Carl Linnaeus first proposed organizing the known **species** of organisms into a hierarchical taxonomy. In this system, species that are most similar to each other are put together within a grouping known as a **genus**. Furthermore, similar genera (the plural of genus) are put together within a **family**. This grouping continues until all organisms are collected together into groups at the highest level. The current taxonomic system now has eight levels in its hierarchy, from lowest to highest, they are: **species, genus, family, order, class, phylum, kingdom, domain**. Thus species are grouped within genera, genera are grouped within families, families are grouped within orders, and so on (Figure 1).

The kingdom Animalia stems from the Eukarya domain. For the common dog, the classification levels would be as shown in Figure 1. Therefore, the full name of an organism technically has eight terms. For the dog, it is: Eukarya, Animalia, Chordata, Mammalia, Carnivora, Canidae, *Canis,* and *lupus*. Notice that each name is capitalized except for species, and the genus and species names are italicized. Scientists generally refer to an organism only by its genus and species, which is its two-word scientific name, in what is called **binomial nomenclature**. Therefore, the scientific name of the dog is *Canis lupus*. The name at each level is also called a **taxon**. In other words, dogs are in order Carnivora. Carnivora is the name of the taxon at the order level; Canidae is the taxon at the family level, and so forth. Organisms also have a common name that people typically use, in this case, dog. Note that the dog is additionally a subspecies: the “*familiaris*” in *Canis lupus familiaris.* Subspecies are members of the same species that are capable of mating and reproducing viable offspring, but they are considered separate subspecies due to geographic or behavioral isolation or other factors.

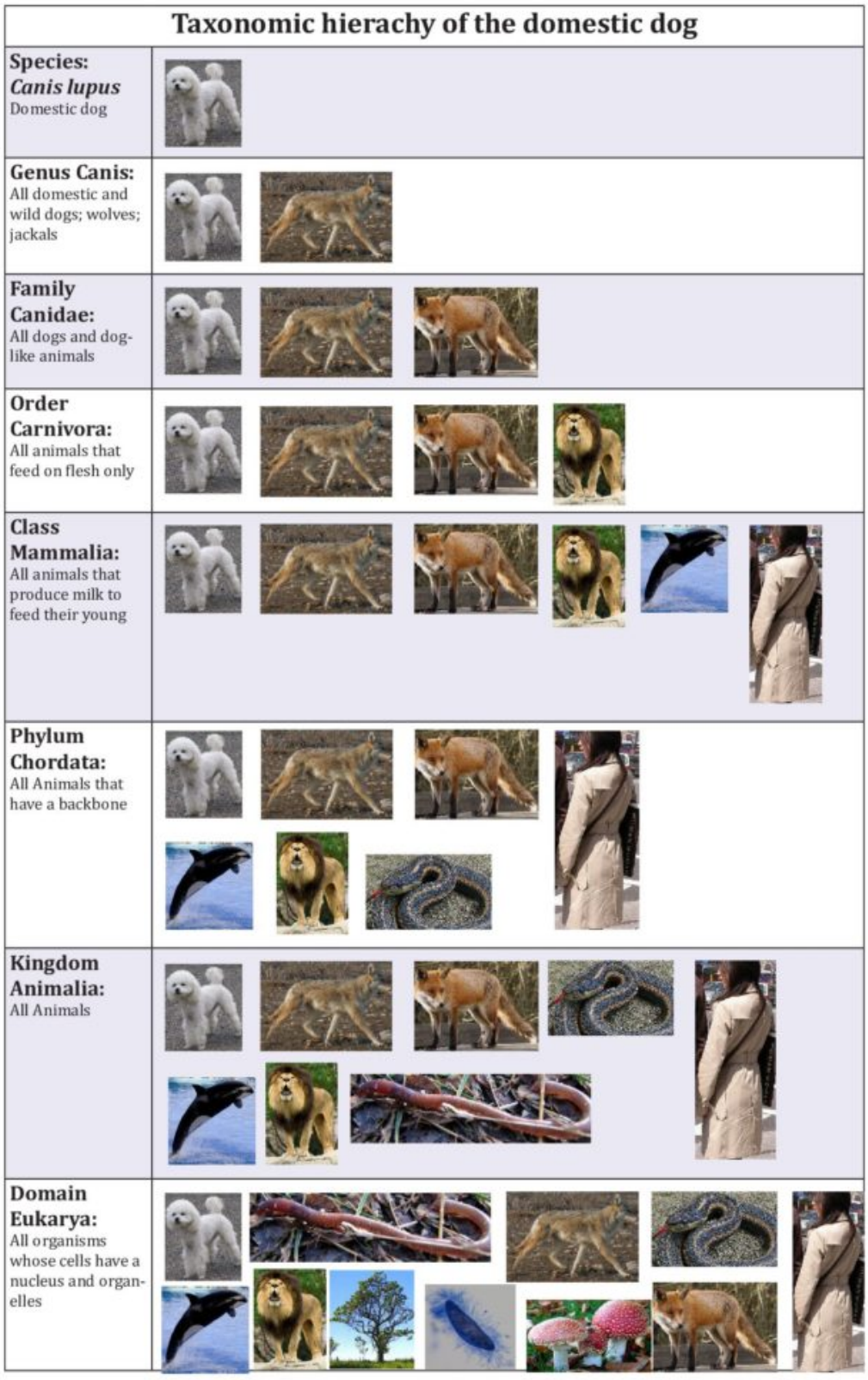

Figure 1. This diagram shows the levels of taxonomic hierarchy for a dog, from the broadest category—domain—to the most specific—species. Click for a larger image.

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INTRODUCTION TO THE BRANCHES OF BIOLOGY

What you'll learn to do: Identify the main branches of biology

While this course provides a broad introduction to the science of biology, higher level study of the subject quickly breaks down into a vast number of sub-disciplines (e.g., microbiology, immunology, neurobiology, anatomy and physiology). Specialists in these different fields of biology include doctors, nutritionists, pharmacologists, botanists, astrobiologists, and many many more.

In this section, we'll learn about the different paths you can take as you study the science of life.

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THE BRANCHES OF BIOLOGY

Learning Outcomes

Identify the main branches of biology

The scope of biology is broad and therefore contains many branches and sub-disciplines. Biologists may pursue one of those sub-disciplines and work in a more focused field. For instance, molecular biology and biochemistry study biological processes at the molecular and chemical level, including interactions among molecules such as DNA, RNA, and proteins, as well as the way they are regulated. Microbiology, the study of microorganisms, is the

study of the structure and function of single-celled organisms. It is quite a broad branch itself, and depending on the subject of study, there are also microbial physiologists, ecologists, and geneticists, among others.

Forensic Science

Forensic science is the application of science to answer questions related to the law. Biologists as well as chemists and biochemists can be forensic scientists. Forensic scientists provide scientific evidence for use in courts, and their job involves examining trace materials associated with crimes. Interest in forensic science has increased in the last few years, possibly because of popular television shows that feature forensic scientists on the job. Also, the development of molecular techniques and the establishment of DNA databases have expanded the types of work that forensic scientists can do.

Figure 1. This forensic scientist works in a DNA extraction room at the U.S. Army Criminal Investigation Laboratory at Fort Gillem, GA. (credit: United States Army CID Command Public Affairs)

Their job activities are primarily related to crimes against people such as murder, rape, and assault. Their work involves analyzing samples such as hair, blood, and other body fluids and also processing DNA (Figure 1) found in many different environments and materials.

Forensic scientists also analyze other biological evidence left at crime scenes, such as insect larvae or pollen grains. Students who want to pursue careers in forensic science will most likely be required to take chemistry and biology courses as well as some intensive math courses.

Another field of biological study, neurobiology, studies the biology of the nervous system, and although it is considered a branch of biology, it is also recognized as an interdisciplinary field of study known as neuroscience. Because of its interdisciplinary nature, this sub-discipline studies different functions of the nervous system using molecular, cellular, developmental, medical, and computational approaches.

Paleontology, another branch of biology, uses fossils to study life's history (Figure 2). Zoology and botany are the study of animals and plants, respectively. Biologists can also specialize as biotechnologists, ecologists, or physiologists, to name just a few areas. Biotechnologists apply the knowledge of biology to create useful products. Ecologists study the interactions of organisms in their environments. Physiologists study the workings of cells, tissues and organs. This is just a small sample of the many fields that biologists can pursue. From our own bodies to the world we live in, discoveries in biology can affect us in very direct and important ways. We depend on these discoveries for our health, our food sources, and the benefits provided by our ecosystem. Because of this, knowledge of biology can benefit us in making decisions in our day-to-day lives.

Figure 2. Researchers work on excavating dinosaur fossils at a site in Castellón, Spain. (credit: Mario Modesto)

The development of technology in the twentieth century that continues today, particularly the technology to describe and manipulate the genetic material, DNA, has transformed biology. This transformation will allow biologists to continue to understand the history of life in greater detail, how the human body works, our human origins, and how humans can survive as a species on this planet despite the stresses caused by our increasing numbers. Biologists continue to decipher huge mysteries about life suggesting that we have only

begun to understand life on the planet, its history, and our relationship to it. For this and other reasons, the knowledge of biology gained through this textbook and other printed and electronic media should be a benefit in whichever field you enter.

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INTRODUCTION TO THE PROCESS OF SCIENCE

What you'll learn to do: Describe biology as a science and identify the key components of scientific inquiry

Like geology, physics, and chemistry, biology is a science that gathers knowledge about the natural world. Specifically, biology is the study of life. The discoveries of biology are made by a community of researchers who work individually and together using agreed-on methods. In this sense, biology, like all sciences, is a social enterprise like politics or the arts. The methods of science include careful observation, record keeping, logical and mathematical reasoning, experimentation, and submitting conclusions to the scrutiny of others. Science also requires considerable imagination and creativity; a well-designed experiment is commonly described as elegant, or beautiful. Like politics, science has considerable practical implications and some science is dedicated to practical applications, such as the prevention of disease (see Figure 1). Other science proceeds largely motivated by curiosity. Whatever its goal, there is no doubt that science, including biology, has transformed human existence and will continue to do so.

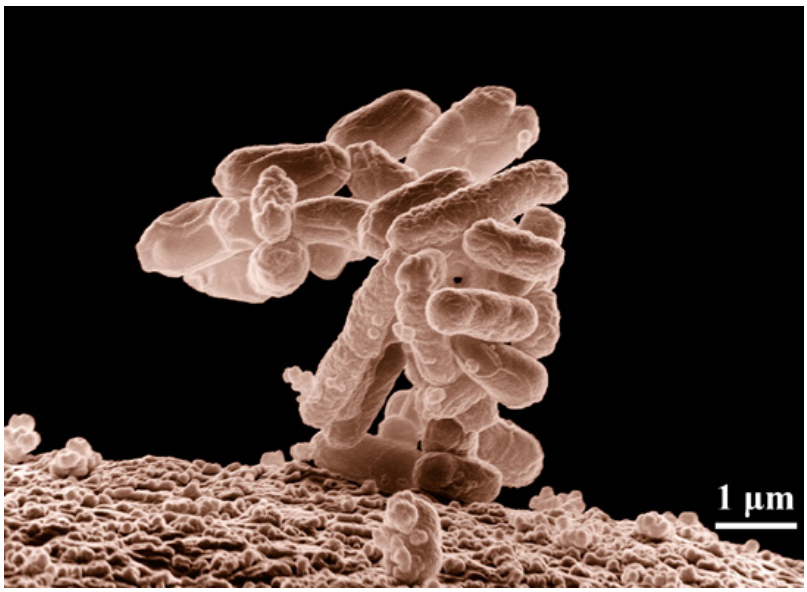

Figure 1. Biologists may choose to study Escherichia coli (E. coli), a bacterium that is a normal resident of our digestive tracts but which is also sometimes responsible for disease outbreaks. In this micrograph, the bacterium is visualized using a scanning electron microscope and digital colorization. (credit: Eric Erbe; digital colorization by Christopher Pooley, USDA-ARS)

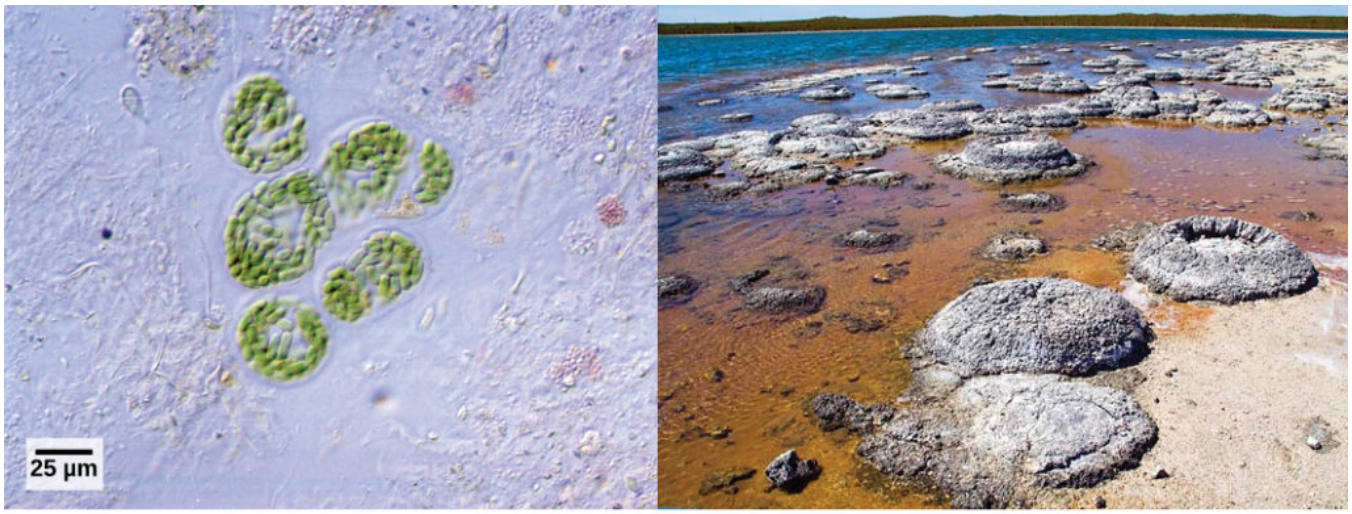

Figure 2. Formerly called blue-green algae, the (a) cyanobacteria seen through a light microscope are some of Earth's oldest life forms. These (b) stromatolites along the shores of Lake Thetis in Western Australia are ancient structures formed by the layering of cyanobacteria in shallow waters. (credit a: modification of work by NASA; scale-bar data from Matt Russell; credit b: modification of work by Ruth Ellison)

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SCIENTIFIC INQUIRY

Learning Outcomes

- Compare inductive reasoning with deductive reasoning
- Describe the process of scientific inquiry

One thing is common to all forms of science: an ultimate goal "to know." Curiosity and inquiry are the driving forces for the development of science. Scientists seek to understand the world and the way it operates. Two methods of logical thinking are used: inductive reasoning and deductive reasoning.

Inductive reasoning is a form of logical thinking that uses related observations to arrive at a general conclusion. This type of reasoning is common in descriptive science. A life scientist such as a biologist makes observations and records them. These data can be qualitative (descriptive) or quantitative (consisting of numbers), and the raw data can be supplemented with drawings, pictures, photos, or videos. From many observations, the scientist can infer conclusions (inductions) based on evidence. Inductive reasoning involves formulating generalizations inferred from careful observation and the analysis of a large amount of data. Brain studies often work this way. Many brains are observed while people are doing a task. The part of the brain that lights up, indicating activity, is then demonstrated to be the part controlling the response to that task.

Deductive reasoning or deduction is the type of logic used in hypothesis-based science. In deductive reasoning, the pattern of thinking moves in the opposite direction as compared to inductive reasoning. **Deductive reasoning** is a form of logical thinking that uses a general principle or law to forecast specific results. From those general principles, a scientist can extrapolate and predict the specific results that would be valid as long as the general

principles are valid. For example, a prediction would be that if the climate is becoming warmer in a region, the distribution of plants and animals should change. Comparisons have been made between distributions in the past and the present, and the many changes that have been found are consistent with a warming climate. Finding the change in distribution is evidence that the climate change conclusion is a valid one.

Both types of logical thinking are related to the two main pathways of scientific study: descriptive science and hypothesis-based science. **Descriptive** (or discovery) **science** aims to observe, explore, and discover, while **hypothesis-based science** begins with a specific question or problem and a potential answer or solution that can be tested. The boundary between these two forms of study is often blurred, because most scientific endeavors combine both approaches. Observations lead to questions, questions lead to forming a hypothesis as a possible answer to those questions, and then the hypothesis is tested. Thus, descriptive science and hypothesis-based science are in continuous dialogue.

Hypothesis Testing

Biologists study the living world by posing questions about it and seeking science-based responses. This approach is common to other sciences as well and is often referred to as the scientific method. The scientific method was used even in ancient times, but it was first documented by England's Sir Francis Bacon (1561–1626) (Figure 1), who set up inductive methods for scientific inquiry. The scientific method is not exclusively used by biologists but can be applied to almost anything as a logical problem-solving method.

Figure 1. Sir Francis Bacon is credited with being the first to document the scientific method.

The scientific process typically starts with an observation (often a problem to be solved) that leads to a question. Let's think about a simple problem that starts with an observation and apply the scientific method to solve the problem. One Monday morning, a student arrives at class and quickly discovers that the classroom is too warm. That is an observation that also describes a problem: the classroom is too warm. The student then asks a question: "Why is the classroom so warm?"

Recall that a hypothesis is a suggested explanation that can be tested. To solve a problem, several hypotheses may be proposed. For example, one hypothesis might be, "The classroom is warm because no one turned on the air conditioning." But there could be other responses to the question, and therefore other hypotheses may be proposed. A second hypothesis might be, "The classroom is warm because there is a power failure, and so the air conditioning doesn't work."

Once a hypothesis has been selected, a prediction may be made. A prediction is similar to a hypothesis but it typically has the format "If . . . then . . . ." For example, the prediction for the first hypothesis might be, "*If* the student turns on the air conditioning, *then* the classroom will no longer be too warm."

A hypothesis must be testable to ensure that it is valid. For example, a hypothesis that depends on what a bear thinks is not testable, because it can never be known what a bear thinks. It should also be **falsifiable**, meaning that it can be disproven by experimental results. An example of an unfalsifiable hypothesis is "Botticelli's *Birth of Venus* is beautiful." There is no experiment that might show this statement to be false. To test a hypothesis, a researcher will conduct one or more experiments designed to eliminate one or more of the hypotheses. This is important. A hypothesis can be disproven, or eliminated, but it can never be proven. Science does not deal in proofs like mathematics. If an experiment fails to disprove a hypothesis, then we find support for that explanation, but this is not to say that down the road a better explanation will not be found, or a more carefully designed experiment will be found to falsify the hypothesis.

Scientific inquiry has not displaced faith, intuition, and dreams. These traditions and ways of knowing have emotional value and provide moral guidance to many people. But hunches, feelings, deep convictions, old traditions, or dreams cannot be accepted directly as scientifically valid. Instead, science limits itself to ideas that can be tested through verifiable observations. Supernatural claims that events are caused by ghosts, devils, God, or other spiritual entities cannot be tested in this way.

Practice Question

Your friend sees this image of a circle of mushrooms and excitedly tells you it was caused by fairies dancing in a circle on the grass the night before. Can your friend's explanation be studied using the process of science?

Answer

In theory, you might try to observe the fairies. But fairies are magical or supernatural beings. We have never observed them using any verifiable method, so scientists agree that they cannot be studied using scientific tools. Instead, science has an explanation supported by strong evidence: "fairy rings" result when a single colony of fungus spreads out into good habitat over a period of many years. The core area is clear of mushrooms because the soil nutrients have been partly depleted there. This idea can be evaluated with repeated observations over time using chemical soil tests and other verifiable measurements.

Each experiment will have one or more variables and one or more controls. A **variable** is any part of the experiment that can vary or change during the experiment. A **control** is a part of the experiment that does not change. Look for the variables and controls in the example that follows. As a simple example, an experiment might be conducted to test the hypothesis that phosphate limits the growth of algae in freshwater ponds. A series of artificial ponds are filled with water and half of them are treated by adding phosphate each week, while the other half are treated by adding a salt that is known not to be used by algae. The variable here is the phosphate (or lack of phosphate), the experimental or treatment cases are the ponds with added phosphate and the control ponds are those with something inert added, such as the salt. Just adding something is also a control against the possibility that adding extra matter to the pond has an effect. If the treated ponds show lesser growth of algae, then we have found support for our hypothesis. If they do not, then we reject our hypothesis. Be aware that rejecting one hypothesis does not determine whether or not the other hypotheses can be accepted; it simply eliminates one hypothesis that is not valid (Figure 2). Using the scientific method, the hypotheses that are inconsistent with experimental data are rejected.

Practice Question

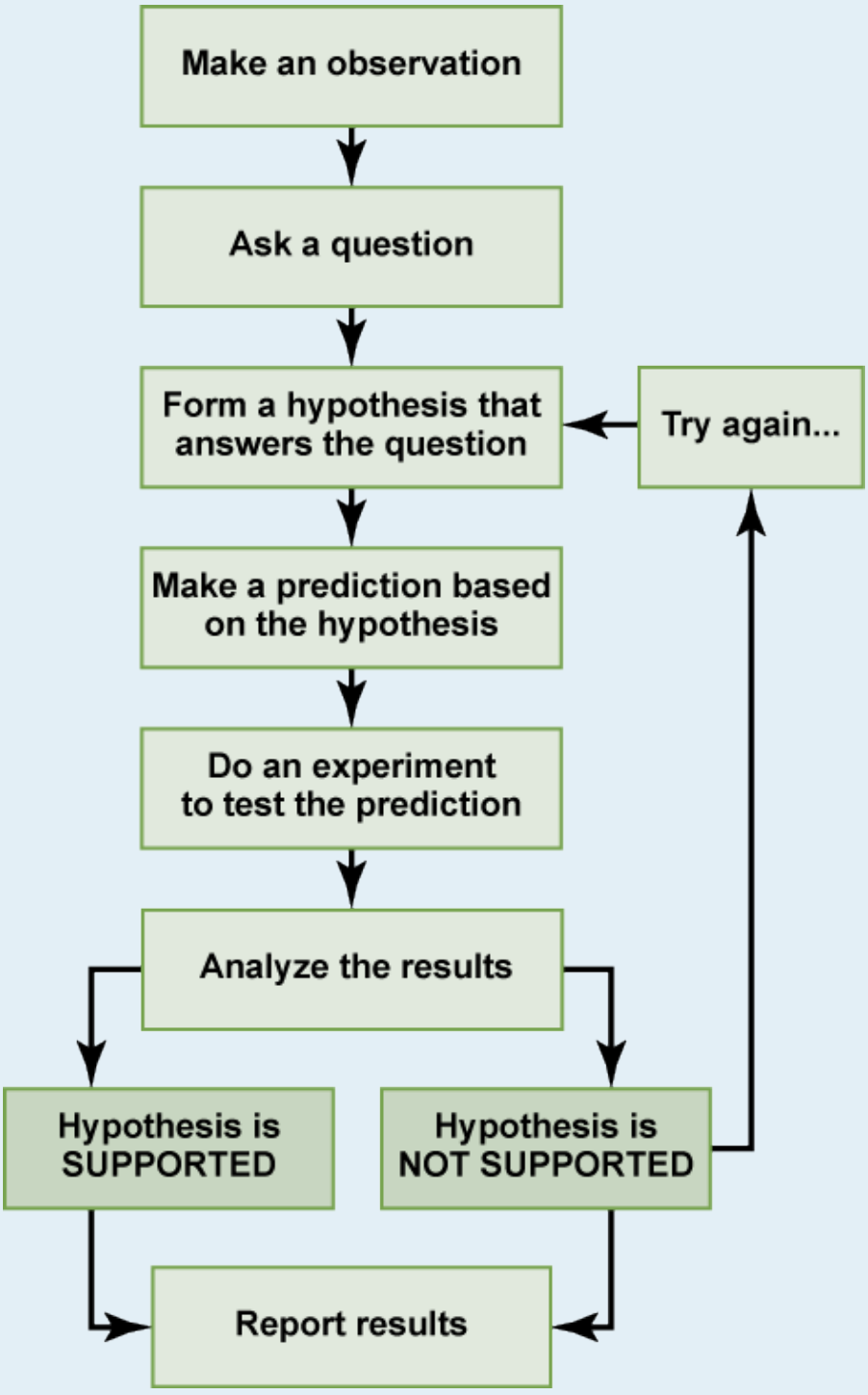

Figure 2. The scientific method is a series of defined steps that include experiments and careful observation. If a hypothesis is not supported by data, a new hypothesis can be proposed.

In the example below, the scientific method is used to solve an everyday problem. Which part in the example below is the hypothesis? Which is the prediction? Based on the results of the experiment, is the hypothesis supported? If it is not supported, propose some alternative hypotheses.

1. My toaster doesn't toast my bread.
2. Why doesn't my toaster work?
3. There is something wrong with the electrical outlet.
4. If something is wrong with the outlet, my coffeemaker also won't work when plugged into it.
5. I plug my coffeemaker into the outlet.
6. My coffeemaker works.

Answer

The hypothesis is #3 (there is something wrong with the electrical outlet), and the prediction is #4 (if something is wrong with the outlet, then the coffeemaker also won't work when plugged into the outlet). The original hypothesis is not supported, as the coffee maker works when plugged into the outlet. Alternative hypotheses may include (1) the toaster might be broken or (2) the toaster wasn't turned on.

In practice, the scientific method is not as rigid and structured as it might at first appear. Sometimes an experiment leads to conclusions that favor a change in approach; often, an experiment brings entirely new scientific questions to the puzzle. Many times, science does not operate in a linear fashion; instead, scientists continually draw inferences and make generalizations, finding patterns as their research proceeds. Scientific reasoning is more complex than the scientific method alone suggests.

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BASIC AND APPLIED SCIENCE

Learning Outcomes

Describe the goals of basic science and applied science

The scientific community has been debating for the last few decades about the value of different types of science. Is it valuable to pursue science for the sake of simply gaining knowledge, or does scientific knowledge only have worth if we can apply it to solving a specific problem or bettering our lives? This question focuses on the differences between two types of science: basic science and applied science.

Basic science or "pure" science seeks to expand knowledge regardless of the short-term application of that knowledge. It is not focused on developing a product or a service of immediate public or commercial value. The immediate goal of basic science is knowledge for knowledge's sake, though this does not mean that in the end it may not result in an application.

In contrast, **applied science** or "technology," aims to use science to solve real-world problems, making it possible, for example, to improve a crop yield, find a cure for a particular disease, or save animals threatened by a natural disaster. In applied science, the problem is usually defined for the researcher.

Some individuals may perceive applied science as "useful" and basic science as "useless." A question these people might pose to a scientist advocating knowledge acquisition would be, "What for?" A careful look at the history of science, however, reveals that basic knowledge has resulted in many remarkable applications of great

value. Many scientists think that a basic understanding of science is necessary before an application is developed; therefore, applied science relies on the results generated through basic science. Other scientists think that it is time to move on from basic science and instead to find solutions to actual problems. Both approaches are valid. It is true that there are problems that demand immediate attention; however, few solutions would be found without the help of the knowledge generated through basic science.

One example of how basic and applied science can work together to solve practical problems occurred after the discovery of DNA structure led to an understanding of the molecular mechanisms governing DNA replication. Strands of DNA, unique in every human, are found in our cells, where they provide the instructions necessary for life. During DNA replication, new copies of DNA are made, shortly before a cell divides to form new cells. Understanding the mechanisms of DNA replication enabled scientists to develop laboratory techniques that are now used to identify genetic diseases, pinpoint individuals who were at a crime scene, and determine paternity. Without basic science, it is unlikely that applied science would exist.

Another example of the link between basic and applied research is the Human Genome Project, a study in which each human chromosome was analyzed and mapped to determine the precise sequence of DNA subunits and the exact location of each gene. (The gene is the basic unit of heredity; an individual's complete collection of genes is his or her genome.) Other organisms have also been studied as part of this project to gain a better understanding of human chromosomes. The Human Genome Project (Figure 1) relied on basic research carried out with non-human organisms and, later, with the human genome. An important end goal eventually became using the data for applied research seeking cures for genetically related diseases.

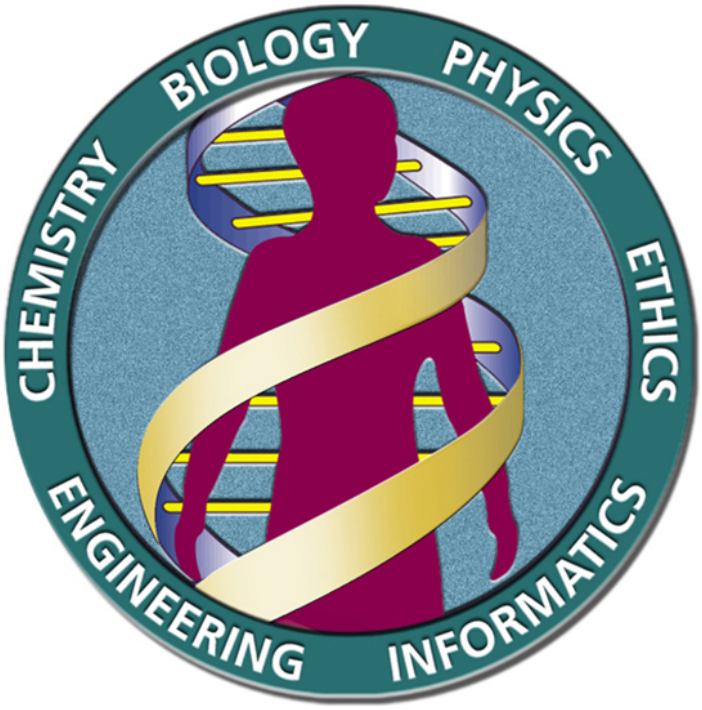

Figure 1. The Human Genome Project was a 13-year collaborative effort among researchers working in several different fields of science. The project was completed in 2003. (credit: the U.S. Department of Energy Genome Programs)

While research efforts in both basic science and applied science are usually carefully planned, it is important to note that some discoveries are made by serendipity, that is, by means of a fortunate accident or a lucky surprise. Penicillin was discovered when biologist Alexander Fleming accidentally left a petri dish of *Staphylococcus* bacteria open. An unwanted mold grew, killing the bacteria. The mold turned out to be *Penicillium*, and a new antibiotic was discovered. Even in the highly organized world of science, luck—when combined with an observant, curious mind—can lead to unexpected breakthroughs.

Reporting Scientific Work

Whether scientific research is basic science or applied science, scientists must share their findings for other researchers to expand and build upon their discoveries. Communication and collaboration within and between sub disciplines of science are key to the advancement of knowledge in science. For this reason, an important aspect of a scientist's work is disseminating results and communicating with peers. Scientists can share results by presenting them at a scientific meeting or conference, but this approach can reach only the limited few who are present. Instead, most scientists present their results in peer-reviewed articles that are published in scientific journals. **Peer-reviewed articles** are scientific papers that are reviewed by a scientist's colleagues, or peers. These colleagues are qualified individuals, often experts in the same research area, who judge whether or not the scientist's work is suitable for publication. The process of peer review helps to ensure that the research described in a scientific paper or grant proposal is original, significant, logical, and thorough. Grant proposals, which are requests for research funding, are also subject to peer review. Scientists publish their work so other scientists can

reproduce their experiments under similar or different conditions to expand on the findings. The experimental results must be consistent with the findings of other scientists.

There are many journals and the popular press that do not use a peer-review system. A large number of online open-access journals, journals with articles available without cost, are now available many of which use rigorous peer-review systems, but some of which do not. Results of any studies published in these forums without peer review are not reliable and should not form the basis for other scientific work. In one exception, journals may allow a researcher to cite a personal communication from another researcher about unpublished results with the cited author's permission.

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SUMMARY: THE PROCESS OF SCIENCE

Learning Outcomes

- Compare inductive reasoning with deductive reasoning
- Describe the process of scientific inquiry
- Describe the goals of basic science and applied science

Biology is the science that studies living organisms and their interactions with one another and their environments. Science attempts to describe and understand the nature of the universe in whole or in part. Science has many fields; those fields related to the physical world and its phenomena are considered natural sciences.

A hypothesis is a tentative explanation for an observation. A scientific theory is a well-tested and consistently verified explanation for a set of observations or phenomena. A scientific law is a description, often in the form of a mathematical formula, of the behavior of an aspect of nature under certain circumstances. Two types of logical reasoning are used in science. Inductive reasoning uses results to produce general scientific principles. Deductive reasoning is a form of logical thinking that predicts results by applying general principles. The common thread throughout scientific research is the use of the scientific method. Scientists present their results in peer-reviewed scientific papers published in scientific journals.

Science can be basic or applied. The main goal of basic science is to expand knowledge without any expectation of short-term practical application of that knowledge. The primary goal of applied research, however, is to solve practical problems.

Practice Question

A suggested and testable explanation for an event is called a _____.

a. hypothesis
b. variable
c. theory
d. control

Answer

A suggested and testable explanation for an event is called a **hypothesis**.

Practice Question

Give an example of how applied science has had a direct effect on your daily life.

Answer

Answers will vary. One example of how applied science has had a direct effect on daily life is the presence of vaccines. Vaccines to prevent diseases such polio, measles, tetanus, and even the influenza affect daily life by contributing to individual and societal health.

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PUTTING IT TOGETHER: INTRODUCTION TO BIOLOGY

Biology is the study of life. As we've learned, this field covers a broad scope of subjects. As you progress through this course, you'll gain the knowledge you need to make informed decisions. Let's think back to the articles Cristina encountered at the beginning of this module: how could a knowledge of biological principle help with her understanding of each article?

Think Back

Cristina read an article about some of the world's weirdest animals. By learning about evolution and natural selection, Cristina could begin to see the different evolutionary reasons behind the extreme features some animals have. For example, the aye-aye, a mammal native to Madagascar, has evolved to have an extra long middle finger, which it can use to dig grubs out of trees. We'll learn more about how these types of traits are selected for in Module 12: Theory of Evolution.

The next article Cristina read talked about GMOs (genetically modified organisms) and the risks they have. In order to truly understand potential risks of genetically modified foods, Cristina will need to first understand the science behind these foods. How are GMOs created? We'll learn more about this in the Module 13: Modern Biology.

Cristina then looked at an article about the paleo diet. In order to survive, humans require specific nutrients. While we won't get into too much depth about nutrition in this course, we will learn about different biological macromolecules in Module 3: Important Biological Macromolecules. These macromolecules include categories you may recognize, like proteins, lipids (more commonly called fats), and carbohydrates. In that module, we'll learn about the roles these molecules play in our bodies—we'll learn the essential functions they perform. With this knowledge, Cristina could better decide what she should or shouldn't remove from her diet.

As you can see in Cristina's example, biology is all around us—after all, you're a living human being! In this course, we'll learn about key biological principles that can help you live your life the best you can.

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MODULE 2: CHEMISTRY OF LIFE

WHY IT MATTERS: THE CHEMISTRY OF LIFE

Why learn about chemistry?

You may have heard the phrase “carbon-based life” when people discuss life on earth. Carbon is an element—one of the basic substances everything is made up of—but just what it mean to say life is “carbon-based”?

In order to understand this concept, we’ll need to understand more about elements, which means learning about chemistry. While chemistry is a separate field, its principles form the basis of biology. As you learned in the last chapter, all living things demonstrate hierarchical organization. You cannot truly understand the higher levels of organization (such as organisms or ecosystems) without understanding their component parts (like cells, molecules, and atoms).

Watch this video online: https://youtu.be/QnQe0xW_JY4

Professionals who use this chemistry in their daily work include nutritionists, healthcare workers (especially when prescribing and administering medications), geneticists, and pharmacologists.

Occupation Spotlight: Nutritionists

Nutritionists are often responsible for planning healthy menus and meal plans, for example in schools or daycares. They may advise individuals on changes in diet in order to achieve a particular health goal. Why do you think they might need to know about chemistry?

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INTRODUCTION TO ATOMS AND ELEMENTS

What you'll learn to do: Define atoms and elements

You're probably familiar with the concept of atoms. They are the fundamental unit of matter—everything is made up of atoms, which come together in unique ways to form different things. Before you can understand chemical reactions, you must first understand the way that atoms work.

Over the years, scientists have used different models to visualize atoms as our understanding has changed. You may be familiar with a few of the models in Figure 1. In this course, we we largely use the Bohr model.

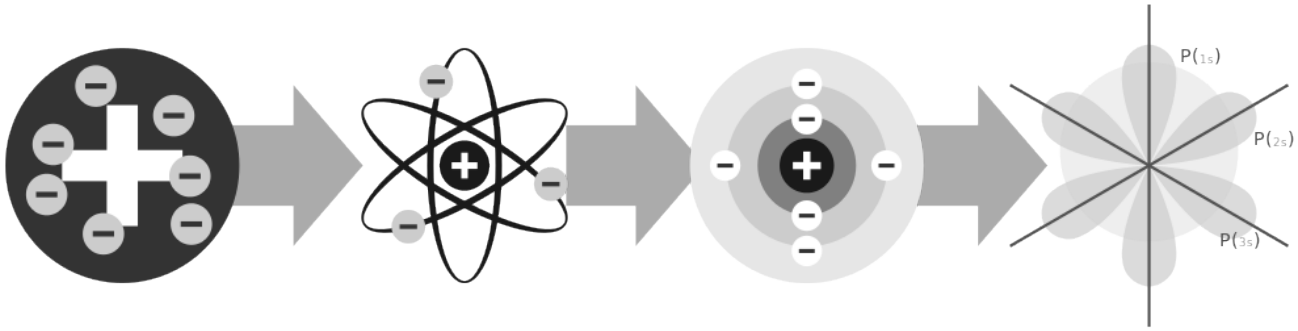

Figure 1. The evolution of atomic models over the years. As our understanding of atoms has evolved, the models we use to depict them have changed. The first model is the Thomson model, followed by the Rutherford model, the Bohr model, and the Heisenberg/Schrödinger model.

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ELEMENTS IN BIOLOGICAL MATTER

Learning Outcomes

Identify the elements common in biological matter

At its most fundamental level, life is made up of matter. **Matter** is any substance that occupies space and has mass. **Elements** are unique forms of matter with specific chemical and physical properties that cannot be broken down into smaller substances by ordinary chemical reactions. There are 118 elements, but only 92 occur naturally. The remaining elements are synthesized in laboratories and are unstable.

Each element is designated by its chemical symbol, which is a single capital letter or, when the first letter is already "taken" by another element, a combination of two letters. Some elements follow the English term for the

element, such as C for carbon and Ca for calcium. Other elements' chemical symbols derive from their Latin names; for example, the symbol for sodium is Na, referring to *natrium*, the Latin word for sodium.

The four elements common to all living organisms are oxygen (O), carbon (C), hydrogen (H), and nitrogen (N). In the non-living world, elements are found in different proportions, and some elements common to living organisms are relatively rare on the earth as a whole, as shown in Table 1. For example, the atmosphere is rich in nitrogen and oxygen but contains little carbon and hydrogen, while the earth's crust, although it contains oxygen and a small amount of hydrogen, has little nitrogen and carbon. In spite of their differences in abundance, *all elements and the chemical reactions between them obey the same chemical and physical laws regardless of whether they are a part of the living or non-living world.*

Table 1. Approximate Percentage of Elements in Living Organisms (Humans) Compared to the Non-living World

Element	Life (Humans)	Atmosphere	Earth's Crust
Oxygen (O)	65%	21%	46%
Carbon (C)	18%	trace	trace
Hydrogen (H)	10%	trace	0.1%
Nitrogen (N)	3%	78%	trace

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ATOMS

Learning Outcomes

Explain the structure and components of an atom

To understand how elements come together, we must first discuss the smallest component or building block of an element, the atom. An **atom** is the smallest unit of matter that retains all of the chemical properties of an element. For example, one gold atom has all of the properties of gold in that it is a solid metal at room temperature. A gold coin is simply a very large number of gold atoms molded into the shape of a coin and containing small amounts of other elements known as impurities. Gold atoms cannot be broken down into anything smaller while still retaining the properties of gold.

All atoms contain protons, electrons, and neutrons (Figure 1). The only exception is hydrogen (H), which is made of one proton and one electron.

A **proton** is a positively charged particle that resides in the **nucleus** (the core of the atom) of an atom and has a mass of 1 and a charge of +1.

Neutrons, like protons, reside in the nucleus of an atom. They have a mass of 1 and no charge. The positive (protons) and negative (electrons) charges balance each other in a neutral atom, which has a net zero charge.

An **electron** is a negatively charged particle that travels in the space around the nucleus. In other words, it resides outside of the nucleus. It has a negligible mass and has a charge of –1.

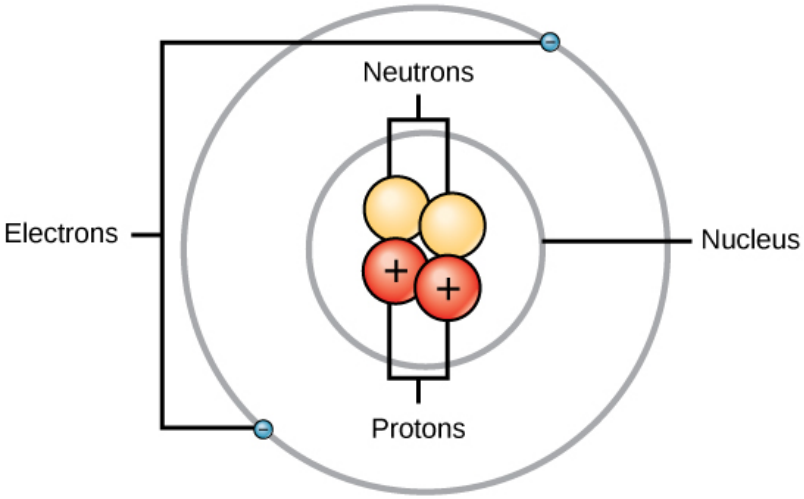

Figure 1. Elements, such as helium, depicted here, are made up of atoms. Atoms are made up of protons and neutrons located within the nucleus, with electrons in orbitals surrounding the nucleus.

Because protons and neutrons each have a mass of 1, the mass of an atom is equal to the number of protons and neutrons of that atom. The number of electrons does not factor into the overall mass, because their mass is so small.

Build An Atom

Build an atom out of protons, neutrons, and electrons, and see how the element, charge, and mass change. Then play a game to test your ideas!

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PROPERTIES OF ELEMENTS

Learning Outcomes

Identify the properties of elements given a periodic table

Atomic Number and Mass

Each element has its own unique properties. Each contains a different number of protons and neutrons, giving it its own atomic number and mass number. The **atomic number** of an element is equal to the number of protons that element contains. The **mass number** is the number of protons plus the number of neutrons of that element.

Therefore, it is possible to determine the number of neutrons by subtracting the atomic number from the mass number.

These numbers provide information about the elements and how they will react when combined. Different elements have different melting and boiling points, and are in different states (liquid, solid, or gas) at room temperature. They also combine in different ways. Some form specific types of bonds, whereas others do not. How they combine is based on the number of electrons present. Because of these characteristics, the elements are arranged into the **periodic table of elements**, a chart of the elements that includes the atomic number and relative atomic mass of each element. The periodic table also provides key information about the properties of elements (Figure 1)—often indicated by color-coding. The arrangement of the table also shows how the electrons in each element are organized and provides important details about how atoms will react with each other to form molecules.

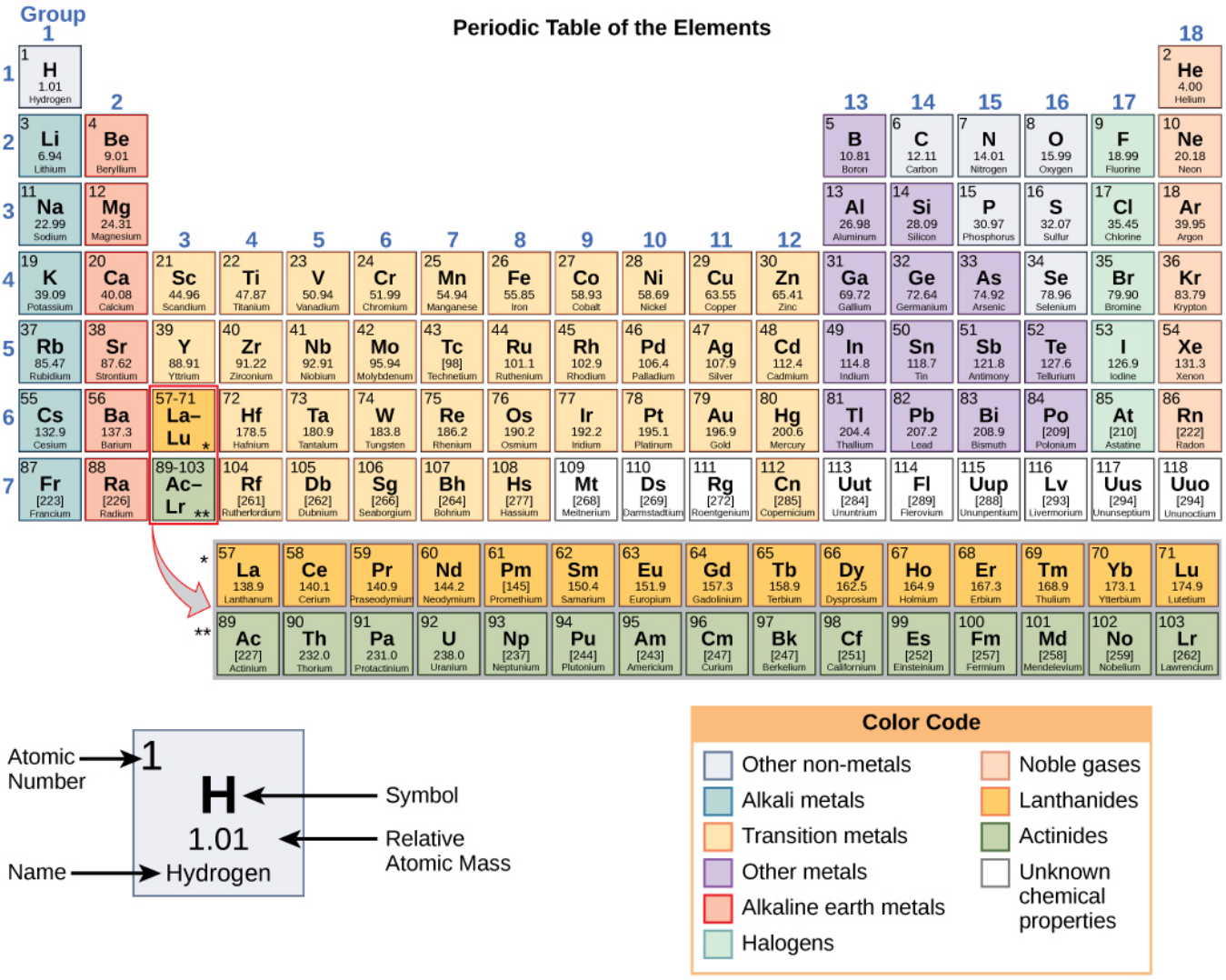

Figure 1. Arranged in columns and rows based on the characteristics of the elements, the periodic table provides key information about the elements and how they might interact with each other to form molecules. Most periodic tables provide a key or legend to the information they contain.

Practice Question

Complete the following table with information from the periodic table

Name of Element	Symbol	Atomic Number	Atomic Mass (Round to the nearest whole number)
Beryllium			
		8	
	C		
			32
	Na		

Show Completed Table

Name of Element	Symbol	Atomic Number	Atomic Mass (Round to the nearest whole number)
Beryllium	Be	4	9
Oxygen	O	8	16
Carbon	C	6	12
Sulfur	S	16	32
Sodium	Na	11	22

Element Interactions

How elements interact with one another depends on how their electrons are arranged and how many openings for electrons exist at the outermost region where electrons are present in an atom. Electrons exist at energy levels that form shells around the nucleus. The closest shell can hold up to two electrons. The closest shell to the nucleus is always filled first, before any other shell can be filled. Hydrogen has one electron; therefore, it has only one spot occupied within the lowest shell. Helium has two electrons; therefore, it can completely fill the lowest shell with its two electrons. If you look at the periodic table, you will see that hydrogen and helium are the only two elements in the first row. This is because they only have electrons in their first shell. Hydrogen and helium are the only two elements that have the lowest shell and no other shells.

The second and third energy levels can hold up to eight electrons. The eight electrons are arranged in four pairs and one position in each pair is filled with an electron before any pairs are completed.

Looking at the periodic table again (Figure 1), you will notice that there are seven rows. These rows correspond to the number of shells that the elements within that row have. The elements within a particular row have increasing numbers of electrons as the columns proceed from left to right. Although each element has the same number of shells, not all of the shells are completely filled with electrons. If you look at the second row of the periodic table, you will find lithium (Li), beryllium (Be), boron (B), carbon (C), nitrogen (N), oxygen (O), fluorine (F), and neon (Ne). These all have electrons that occupy only the first and second shells. Lithium has only one electron in its outermost shell, beryllium has two electrons, boron has three, and so on, until the entire shell is filled with eight electrons, as is the case with neon.

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ISOTOPES

Learning Outcomes

Define the term isotope

Isotopes are different forms of the same element that have the same number of protons, but a different number of neutrons. Some elements, such as carbon, potassium, and uranium, have naturally occurring isotopes. Carbon-12, the most common isotope of carbon, contains six protons and six neutrons. Therefore, it has a mass number of 12 (six protons and six neutrons) and an atomic number of 6 (which makes it carbon). Carbon-14 contains six protons and eight neutrons. Therefore, it has a mass number of 14 (six protons and eight neutrons) and an atomic number of 6, meaning it is still the element carbon. These two alternate forms of carbon are isotopes. Some isotopes are unstable and will lose protons, other subatomic particles, or energy to form more stable elements. These are called **radioactive isotopes** or **radioisotopes**.

Evolution in Action: Carbon Dating

Carbon-14 (^{14}C) is a naturally occurring radioisotope that is created in the atmosphere by cosmic rays. This is a continuous process, so more ^{14}C is always being created. As a living organism develops, the relative level of ^{14}C in its body is equal to the concentration of ^{14}C in the atmosphere. When an organism dies, it is no longer ingesting ^{14}C, so the ratio will decline. ^{14}C decays to ^{14}N by a process called beta decay; it gives off energy in this slow process.

After approximately 5,730 years, only one-half of the starting concentration of ^{14}C will have been converted to ^{14}N. The time it takes for half of the original concentration of an isotope to decay to its more stable form is called its half-life. Because the half-life of ^{14}C is long, it is used to age formerly living objects, such as fossils. Using the ratio of the ^{14}C concentration found in an object to the amount of ^{14}C detected in the atmosphere, the amount of the isotope that has not yet decayed can be determined. Based on this amount, the age of the fossil can be calculated to about 50,000 years (Figure 1). Isotopes with longer half-lives, such as potassium-40, are used to calculate the ages of older fossils. Through the use of

Figure 1. The age of remains that contain carbon and are less than about 50,000 years old, such as this pygmy mammoth, can be determined using carbon dating.

carbon dating, scientists can reconstruct the ecology and biogeography of organisms living within the past 50,000 years.

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INTRODUCTION TO ATOMIC BONDS

What you'll learn to do: Classify different types of atomic bonds

When atoms bond together, they create molecules: a sodium atom bonds with a chlorine atom to create salt (sodium chloride), two hydrogen atoms bond with an oxygen atom to create water (hydrogen dioxide). However, not all atomic bonds are the same; in fact salt and water are created with two very different types of bonds (ionic and polar covalent bonds respectively).

The different types of bonds (ionic, polar covalent, and non-polar covalent bonds) behave differently, and these differences have an impact on the molecules they create. Understanding the types of bonds that create living things can help us understand those living things themselves.

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CHEMICAL BONDING

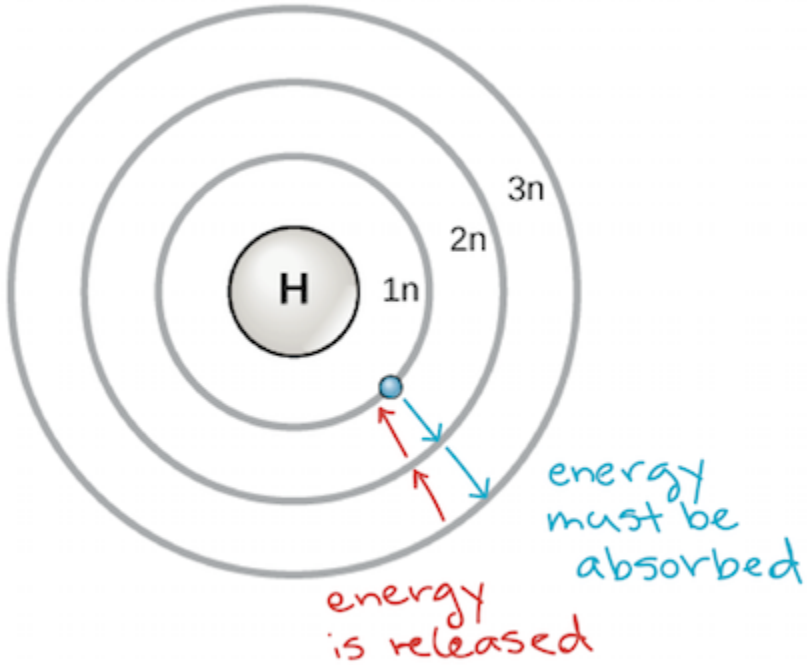
Learning Outcomes

Define the octet rule and its role in chemical bonds

Not all elements have enough electrons to fill their outermost shells, but *an atom is at its most stable when all of the electron positions in the outermost shell are filled*. Because of these vacancies in the outermost shells, we see the formation of chemical bonds, or interactions between two or more of the same or different elements that result in the formation of molecules. To achieve greater stability, atoms will tend to completely fill their outer shells and will bond with other elements to accomplish this goal by sharing electrons, accepting electrons from another atom, or donating electrons to another atom. Because the outermost shells of the elements with low atomic numbers (up to calcium, with atomic number 20) can hold eight electrons, this is referred to as the **octet rule**. An element can donate, accept, or share electrons with other elements to fill its outer shell and satisfy the octet rule.

An early model of the atom was developed in 1913 by the Danish scientist Niels Bohr (1885–1962). The Bohr model shows the atom as a central nucleus containing protons and neutrons, with the electrons in circular electron shells at specific distances from the nucleus, similar to planets orbiting around the sun. Each electron shell has a different energy level, with those shells closest to the nucleus being lower in energy than those farther

from the nucleus. By convention, each shell is assigned a number and the symbol n—for example, the electron shell closest to the nucleus is called 1n. In order to move between shells, an electron must absorb or release an amount of energy corresponding exactly to the difference in energy between the shells. For instance, if an electron absorbs energy from a photon, it may become excited and move to a higher-energy shell; conversely, when an excited electron drops back down to a lower-energy shell, it will release energy, often in the form of heat.

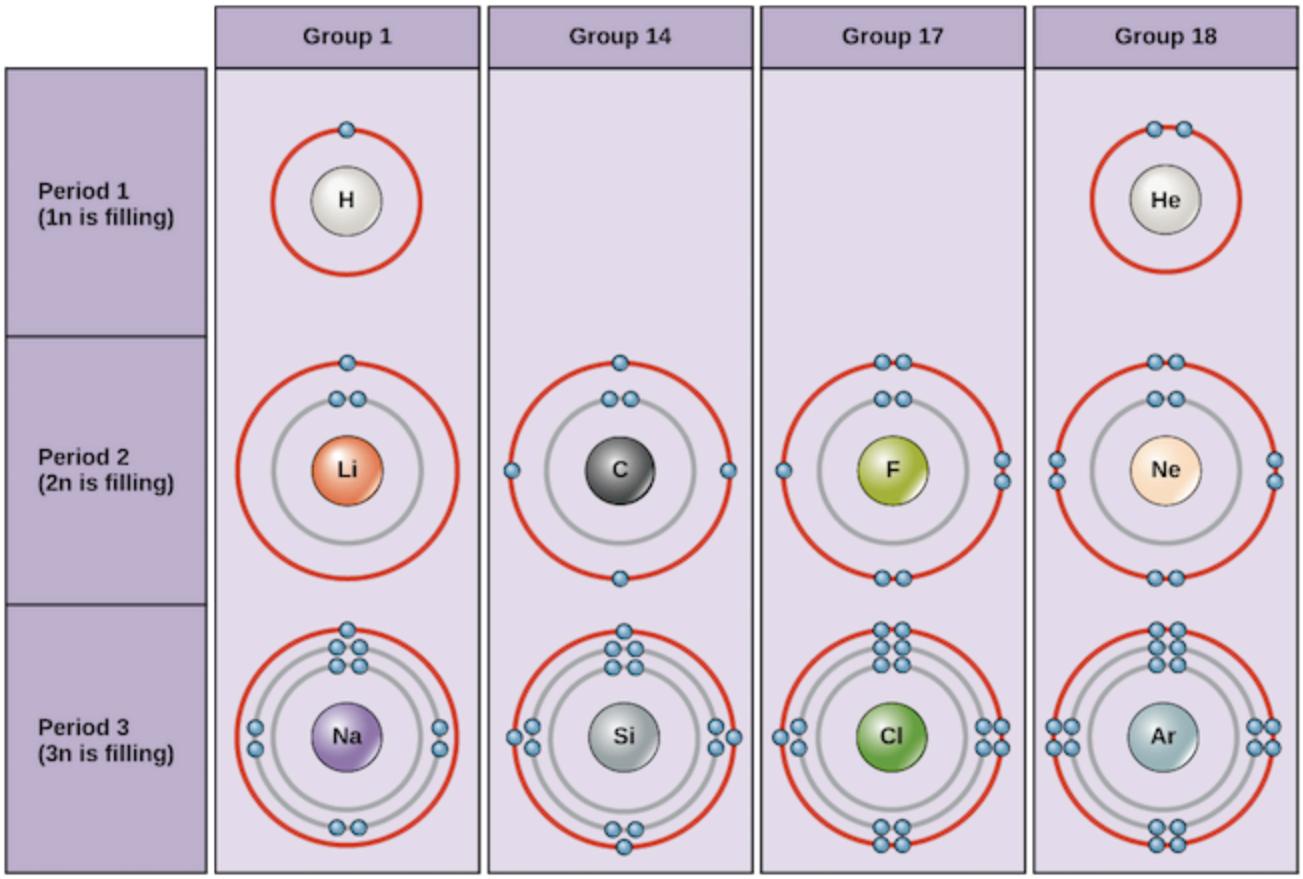


Bohr model of an atom, showing energy levels as concentric circles surrounding the nucleus. Energy must be added to move an electron outward to a higher energy level, and energy is released when an electron falls down from a higher energy level to a closer-in one. Image credit: modified from OpenStax Biology

Atoms, like other things governed by the laws of physics, tend to take on the lowest-energy, most stable configuration they can. Thus, the electron shells of an atom are populated from the inside out, with electrons filling up the low-energy shells closer to the nucleus before they move into the higher-energy shells further out. The shell closest to the nucleus, 1n, can hold two electrons, while the next shell, 2n, can hold eight, and the third shell, 3n, can hold up to eighteen.

The number of electrons in the outermost shell of a particular atom determines its reactivity, or tendency to form chemical bonds with other atoms. This outermost shell is known as the **valence shell**, and the electrons found in it are called **valence electrons**. In general, atoms are most stable, least reactive, when their outermost electron shell is full. Most of the elements important in biology need eight electrons in their outermost shell in order to be stable, and this rule of thumb is known as the **octet rule**. Some atoms can be stable with an octet even though their valence shell is the 3n shell, which can hold up to 18 electrons. We will explore the reason for this when we discuss electron orbitals below.

Examples of some neutral atoms and their electron configurations are shown below. In this table, you can see that helium has a full valence shell, with two electrons in its first and only, 1n, shell. Similarly, neon has a complete outer 2n shell containing eight electrons. These electron configurations make helium and neon very stable. Although argon does not technically have a full outer shell, since the 3n shell can hold up to eighteen electrons, it is stable like neon and helium because it has eight electrons in the 3n shell and thus satisfies the octet rule. In contrast, chlorine has only seven electrons in its outermost shell, while sodium has just one. These patterns do not fill the outermost shell or satisfy the octet rule, making chlorine and sodium reactive, eager to gain or lose electrons to reach a more stable configuration.



Bohr diagrams of various elements Image credit: OpenStax Biology

Electron configurations and the periodic table

Elements are placed in order on the periodic table based on their atomic number, how many protons they have. In a neutral atom, the number of electrons will equal the number of protons, so we can easily determine electron number from atomic number. In addition, the position of an element in the periodic table—its column, or group, and row, or period—provides useful information about how those electrons are arranged.

If we consider just the first three rows of the table, which include the major elements important to life, each row corresponds to the filling of a different electron shell: helium and hydrogen place their electrons in the 1n shell, while second-row elements like Li start filling the 2n shell, and third-row elements like Na continue with the 3n shell. Similarly, an element's column number gives information about its number of valence electrons and reactivity. In general, the number of valence electrons is the same within a column and increases from left to right within a row. Group 1 elements have just one valence electron and group 18 elements have eight, except for helium, which has only two electrons total. Thus, group number is a good predictor of how reactive each element will be:

- Helium (He), neon (Ne), and argon (Ar), as group 18 elements, have outer electron shells that are full or satisfy the octet rule. This makes them highly stable as single atoms. Because of their non-reactivity, they are called the **inert gases** or **noble gases**.
- Hydrogen (H), lithium (Li), and sodium (Na), as group 1 elements, have just one electron in their outermost shells. They are unstable as single atoms, but can become stable by losing or sharing their one valence electron. If these elements fully lose an electron—as Li and Na typically do—they become positively charged ions: Li^+, Na^+.
- Fluorine (F) and chlorine (Cl), as group 17 elements, have seven electrons in their outermost shells. They tend to achieve a stable octet by taking an electron from other atoms, becoming negatively charged ions: F^- and Cl^-.

- Carbon (C), as a group 14 element, has four electrons in its outer shell. Carbon typically shares electrons to achieve a complete valence shell, forming bonds with multiple other atoms.

Thus, the columns of the periodic table reflect the number of electrons found in each element's valence shell, which in turn determines how the element will react.

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IONIC BONDS

Learning Outcomes

Describe the characteristics of ionic bonds and identify common ions

Some atoms are more stable when they gain or lose an electron (or possibly two) and form ions. This fills their outermost electron shell and makes them energetically more stable. Because the number of electrons does not equal the number of protons, each ion has a net charge. **Cations** are positive ions that are formed by losing electrons. Negative ions are formed by gaining electrons and are called anions. **Anions** are designated by their elemental name being altered to end in "-ide": the anion of chlorine is called chloride, and the anion of sulfur is called sulfide, for example.

This movement of electrons from one element to another is referred to as **electron transfer**. As Figure 1 illustrates, sodium (Na) only has one electron in its outer electron shell. It takes less energy for sodium to donate that one electron than it does to accept seven more electrons to fill the outer shell. If sodium loses an electron, it now has 11 protons, 11 neutrons, and only 10 electrons, leaving it with an overall charge of +1. It is now referred to as a sodium ion. Chlorine (Cl) in its lowest energy state (called the ground state) has seven electrons in its outer shell. Again, it is more energy-efficient for chlorine to gain one electron than to lose seven. Therefore, it tends to gain an electron to create an ion with 17 protons, 17 neutrons, and 18 electrons, giving it a net negative (–1) charge. It is now referred to as a chloride ion. In this example, sodium will donate its one electron to empty its shell, and chlorine will accept that electron to fill its shell. Both ions now satisfy the octet rule and have complete outermost shells. Because the number of electrons is no longer equal to the number of protons, each is now an ion and has a +1 (sodium cation) or –1 (chloride anion) charge. Note that these transactions can normally only take place simultaneously: in order for a sodium atom to lose an electron, it must be in the presence of a suitable recipient like a chlorine atom.

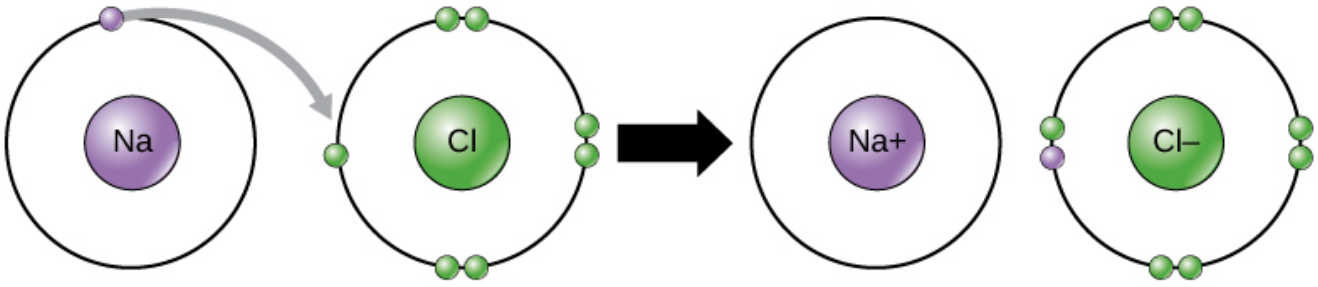

Figure 1. In the formation of an ionic compound, metals lose electrons and nonmetals gain electrons to achieve an octet. Ionic bonds are formed between ions with opposite charges. For instance, positively charged sodium ions and negatively charged chloride ions bond together to make crystals of sodium chloride, or table salt, creating a crystalline molecule with zero net charge.

Certain salts are referred to in physiology as **electrolytes** (including sodium, potassium, and calcium), ions necessary for nerve impulse conduction, muscle contractions and water balance. Many sports drinks and dietary supplements provide these ions to replace those lost from the body via sweating during exercise.

Video Review

This video shows how ionic compounds form from anions and cations.

Watch this video online: https://youtu.be/hiyTfhjeF_U

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COVALENT BONDS

Learning Outcomes

Describe the characteristics of covalent bonds and differentiate between polar and nonpolar bonds

Another way the octet rule can be satisfied is by the sharing of electrons between atoms to form **covalent bonds**. These bonds are stronger and much more common than ionic bonds in the molecules of living organisms. Covalent bonds are commonly found in carbon-based organic molecules, such as our DNA and proteins. Covalent bonds are also found in inorganic molecules like H_2O, CO_2, and O_2. One, two, or three pairs of electrons may be shared, making single, double, and triple bonds, respectively. The more covalent bonds between two atoms, the stronger their connection. Thus, triple bonds are the strongest.

The strength of different levels of covalent bonding is one of the main reasons living organisms have a difficult time in acquiring nitrogen for use in constructing their molecules, even though molecular nitrogen, N_2, is the most abundant gas in the atmosphere. Molecular nitrogen consists of two nitrogen atoms triple bonded to each other and, as with all molecules, the sharing of these three pairs of electrons between the two nitrogen atoms allows for the filling of their outer electron shells, making the molecule more stable than the individual nitrogen atoms. This strong triple bond makes it difficult for living systems to break apart this nitrogen in order to use it as constituents of proteins and DNA.

The formation of water molecules provides an example of covalent bonding. The hydrogen and oxygen atoms that combine to form water molecules are bound together by covalent bonds. The electron from the hydrogen splits its time between the incomplete outer shell of the hydrogen atoms and the incomplete outer shell of the oxygen atoms. To completely fill the outer shell of oxygen, which has six electrons in its outer shell but which would be more stable with eight, two electrons (one from each hydrogen atom) are needed: hence the well-known formula H_2O. The electrons are shared between the two elements to fill the outer shell of each, making both elements more stable.

View this short video to see an animation of ionic and covalent bonding.

Watch this video online: https://youtu.be/QqjcCvzWwww

Polar and Nonpolar Covalent Bonds

There are two types of covalent bonds: polar and nonpolar. **Nonpolar** covalent bonds form between two atoms of the same element or between different elements that share the electrons equally. For example, an oxygen atom can bond with another oxygen atom to fill their outer shells. This association is nonpolar because the electrons will be equally distributed between each oxygen atom. Two covalent bonds form between the two oxygen atoms because oxygen requires two shared electrons to fill its outermost shell. Nitrogen atoms will form three covalent bonds (also called triple covalent) between two atoms of nitrogen because each nitrogen atom needs three electrons to fill its outermost shell. Another example of a nonpolar covalent bond is found in the methane (CH_4) molecule. The carbon atom has four electrons in its outermost shell and needs four more to fill it. It gets these four from four hydrogen atoms, each atom providing one. These elements all share the electrons equally, creating four nonpolar covalent bonds.

In a **polar** covalent bond, the electrons shared by the atoms spend more time closer to one nucleus than to the other nucleus. Because of the unequal distribution of electrons between the different nuclei, a slightly positive ($\delta+$) or slightly negative ($\delta-$) charge develops. The covalent bonds between hydrogen and oxygen atoms in water are polar covalent bonds. The shared electrons spend more time near the oxygen nucleus, giving it a small negative charge, than they spend near the hydrogen nuclei, giving these molecules a small positive charge. Polar covalent bonds form more often when atoms that differ greatly in size share electrons.

Examples of Covalent Bonding

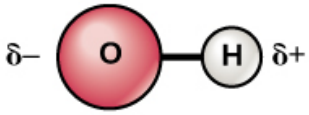
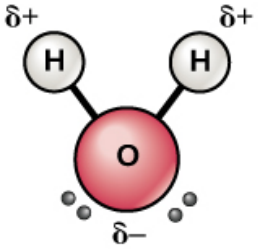
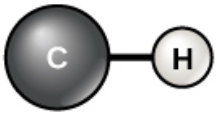
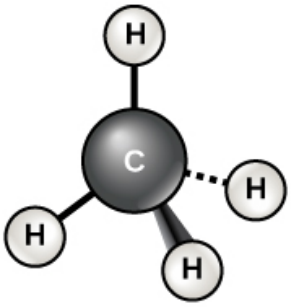
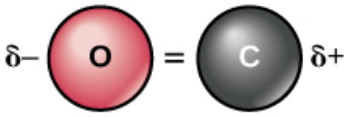
	Bond type	Molecular shape	Molecular type
Water	 Polar covalent	 Bent	Polar
Methane	 Nonpolar covalent	 Tetrahedral	Nonpolar
Carbon dioxide	 Polar covalent	Linear	Nonpolar

Figure 1. Whether a molecule is polar or nonpolar depends both on bond type and molecular shape. Both water and carbon dioxide have polar covalent bonds, but carbon dioxide is linear, so the partial charges on the molecule cancel each other out.

Video Review

Watch this video for another explanation of covalent bonds and how they form:
Watch this video online: https://youtu.be/Mo4Vfqt5v2A

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WEAKER BONDS IN BIOLOGY

Learning Outcomes

- Model a Hydrogen bond and identify its unique qualities
- Model van der Waals interactions identify their unique qualities

Hydrogen Bonds

Ionic and covalent bonds between elements require energy to break. Iconic bonds are not as strong as covalent, which determines their behavior in biological systems. However, not all bonds are ionic or covalent bonds. Weaker bonds can also form between molecules. Two weak bonds that occur frequently are hydrogen bonds and van der Waals interactions. Without these two types of bonds, life as we know it would not exist. Hydrogen bonds provide many of the critical, life-sustaining properties of water and also stabilize the structures of proteins and DNA, the building block of cells.

When polar covalent bonds containing hydrogen form, the hydrogen in that bond has a slightly positive charge because hydrogen's electron is pulled more strongly toward the other element and away from the hydrogen. Because the hydrogen is slightly positive, it will be attracted to neighboring negative charges. When this happens, a weak interaction occurs between the $\delta+$ of the hydrogen from one molecule and the $\delta-$ charge on the more electronegative atoms of another molecule, usually oxygen or nitrogen, or within the same molecule.

Hydrogen Bonding between water molecules

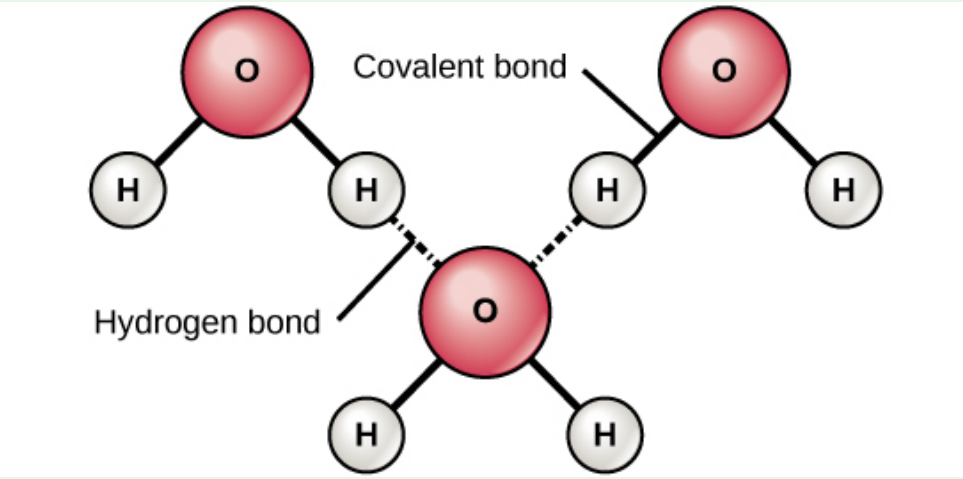

Figure 1. Hydrogen bonds form between slightly positive (δ+) and slightly negative (δ–) charges of polar covalent molecules, such as water.

This interaction is called a **hydrogen bond**. This type of bond is common and occurs regularly between water molecules. Individual hydrogen bonds are weak and easily broken; however, they occur in very large numbers in water and in organic polymers, creating a major force in combination. Hydrogen bonds are also responsible for zipping together the DNA double helix.

Video Review

Watch this video online: https://youtu.be/iOOvX0jmhJ4

van der Waals Interactions

Like hydrogen bonds, van der Waals interactions are weak attractions or interactions between molecules. They are also called inter-molecular forces. They occur between polar, covalently bound atoms in different molecules. Some of these weak attractions are caused by temporary partial charges formed when electrons move around a nucleus. These weak interactions between molecules are important in biological systems and occur based on physical proximity.

Radiology Technician

Have you or anyone you know ever had a magnetic resonance imaging (MRI) scan, a mammogram, or an X-ray? These tests produce images of your soft tissues and organs (as with an MRI or mammogram) or your bones (as happens in an X-ray) by using either radiowaves or special isotopes (radiolabeled or fluorescently labeled) that are ingested or injected into the body. These tests provide data for disease diagnoses by creating images of your organs or skeletal system.

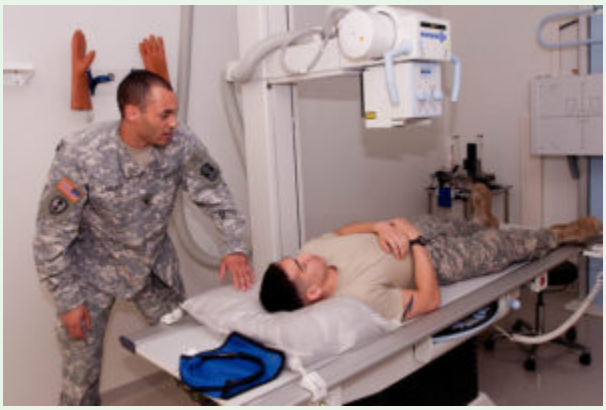

Figure 2. Spc. Arbor L. LaClave practices his spinal X-ray positions utilizing Spc. Justin J. Reichelt, a radiology technician, as his mock patient to practice his skills in the health clinic at Grafenwoehr Training Area.

MRI imaging works by subjecting hydrogen nuclei, which are abundant in the water in soft tissues, to fluctuating magnetic fields, which cause them to emit their own magnetic field. This signal is then read by sensors in the machine and interpreted by a computer to form a detailed image.

Some radiography technologists and technicians specialize in computed tomography, MRI, and mammography. They produce films or images of the body that help medical professionals examine and diagnose. Radiologists work directly with patients, explaining machinery, preparing them for exams, and ensuring that their body or body parts are positioned correctly to produce the needed images. Physicians or radiologists then analyze the test results.

Radiography technicians can work in hospitals, doctors' offices, or specialized imaging centers. Training to become a radiography technician happens at hospitals, colleges, and universities that offer certificates, associate's degrees, or bachelor's degrees in radiography.

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WHY LIFE DEPENDS ON WATER

Learning Outcomes

Describe the properties of water that are critical to maintaining life

Do you ever wonder why scientists spend time looking for water on other planets? It is because water is essential to life; even minute traces of it on another planet can indicate that life could or did exist on that planet. Water is one of the more abundant molecules in living cells and the one most critical to life as we know it. Approximately 60–70 percent of your body is made up of water. Without it, life simply would not exist.

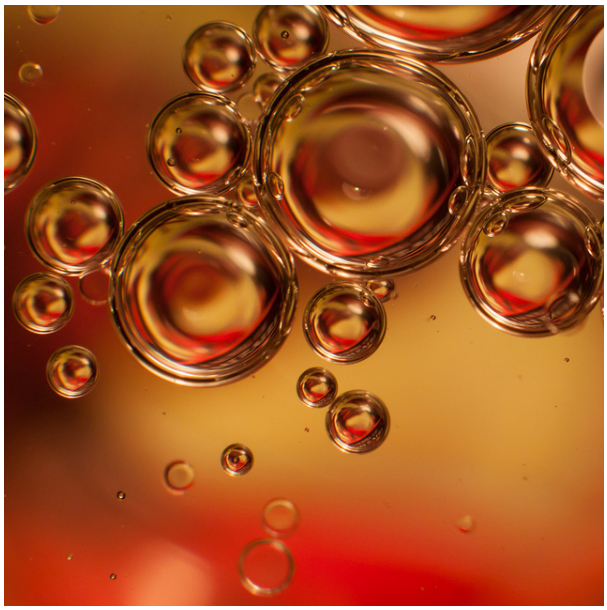

Figure 1. As this image of oil and water shows, oil is a nonpolar compound and, hence, will not dissolve in water. Oil and water do not mix.

Water Is Polar

The hydrogen and oxygen atoms within water molecules form polar covalent bonds. The shared electrons spend more time associated with the oxygen atom than they do with hydrogen atoms. There is no overall charge to a water molecule, but there is a slight positive charge on each hydrogen atom and a slight negative charge on the oxygen atom. Because of these charges, the slightly positive hydrogen atoms repel each other and form the unique shape seen in Figure 2. Each water molecule attracts other water molecules because of the positive and negative charges in the different parts of the molecule. Water also attracts other polar molecules (such as sugars), forming hydrogen bonds. When a substance readily forms hydrogen bonds with water, it can dissolve in water and is referred to as hydrophilic (“water-loving”). Hydrogen bonds are not readily formed with nonpolar substances like oils and fats (Figure 1). These nonpolar compounds are hydrophobic (“water-fearing”) and will not dissolve in water.

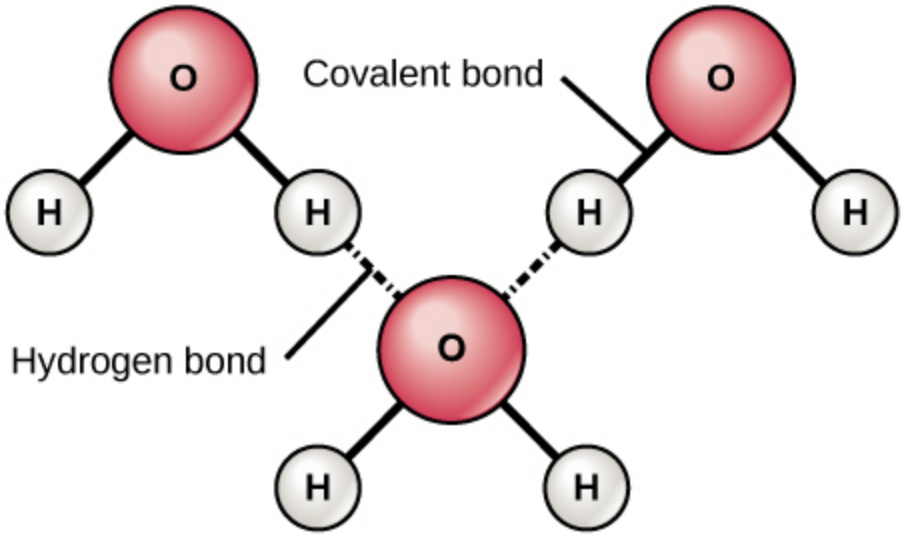

Figure 2. Hydrogen bonds form between slightly positive (δ+) and slightly negative (δ–) charges of polar covalent molecules, such as water.

Water Stabilizes Temperature

The hydrogen bonds in water allow it to absorb and release heat energy more slowly than many other substances. Temperature is a measure of the motion (kinetic energy) of molecules. As the motion increases, energy is higher and thus temperature is higher. Water absorbs a great deal of energy before its temperature rises. Increased energy disrupts the hydrogen bonds between water molecules. Because these bonds can be created and disrupted rapidly, water absorbs an increase in energy and temperature changes only minimally. This means that water moderates temperature changes within organisms and in their environments. As energy input continues, the balance between hydrogen-bond formation and destruction swings toward the destruction side. More bonds are broken than are formed. This process results in the release of individual water molecules at the surface of the liquid (such as a body of water, the leaves of a plant, or the skin of an organism) in a process called evaporation. Evaporation of sweat, which is 90 percent water, allows for cooling of an organism, because breaking hydrogen bonds requires an input of energy and takes heat away from the body.

Conversely, as molecular motion decreases and temperatures drop, less energy is present to break the hydrogen bonds between water molecules. These bonds remain intact and begin to form a rigid, lattice-like structure (e.g., ice) (Figure 3a). When frozen, ice is less dense than liquid water (the molecules are farther apart). This means that ice floats on the surface of a body of water (Figure 3b). In lakes, ponds, and oceans, ice will form on the surface of the water, creating an insulating barrier to protect the animal and plant life beneath from freezing in the water. If this did not happen, plants and animals living in water would freeze in a block of ice and could not move freely, making life in cold temperatures difficult or impossible.

Water Is an Excellent Solvent

Because water is polar, with slight positive and negative charges, ionic compounds and polar molecules can readily dissolve in it. Water is, therefore, what is referred to as a solvent—a substance capable of dissolving another substance. The charged particles will form hydrogen bonds with a surrounding layer of water molecules. This is referred to as a sphere of hydration and serves to keep the particles separated or dispersed in the water. In the case of table salt (NaCl) mixed in water (Figure 4), the sodium and chloride ions separate, or dissociate, in the water, and spheres of hydration are formed around the ions.

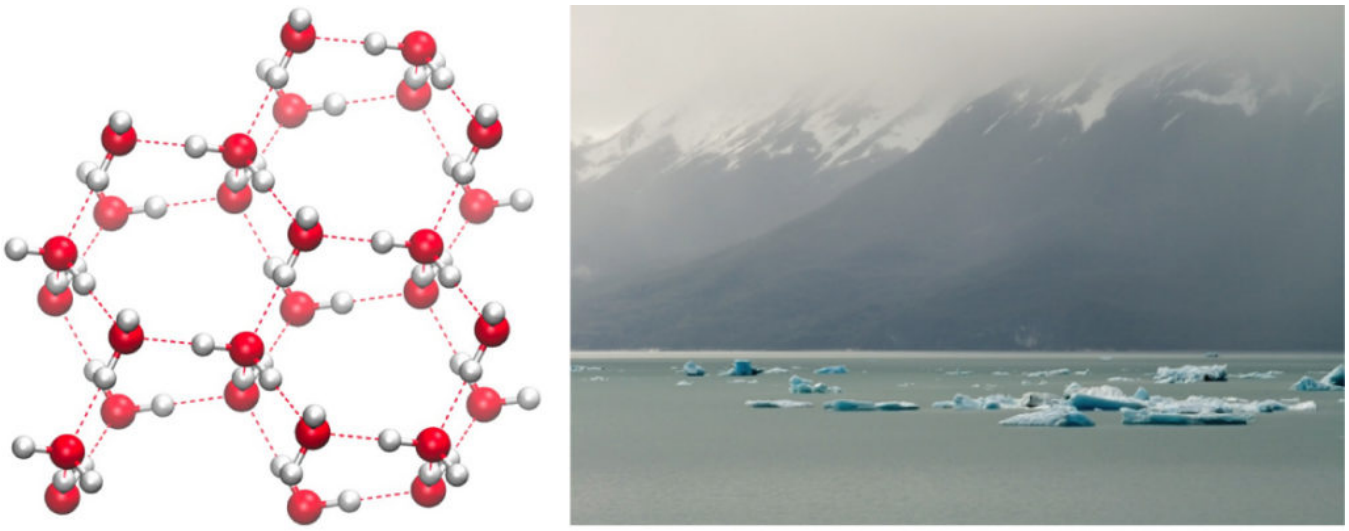

Figure 3. (a) The lattice structure of ice makes it less dense than the freely flowing molecules of liquid water. Ice's lower density enables it to (b) float on water. (credit a: modification of work by Jane Whitney; credit b: modification of work by Carlos Ponte)

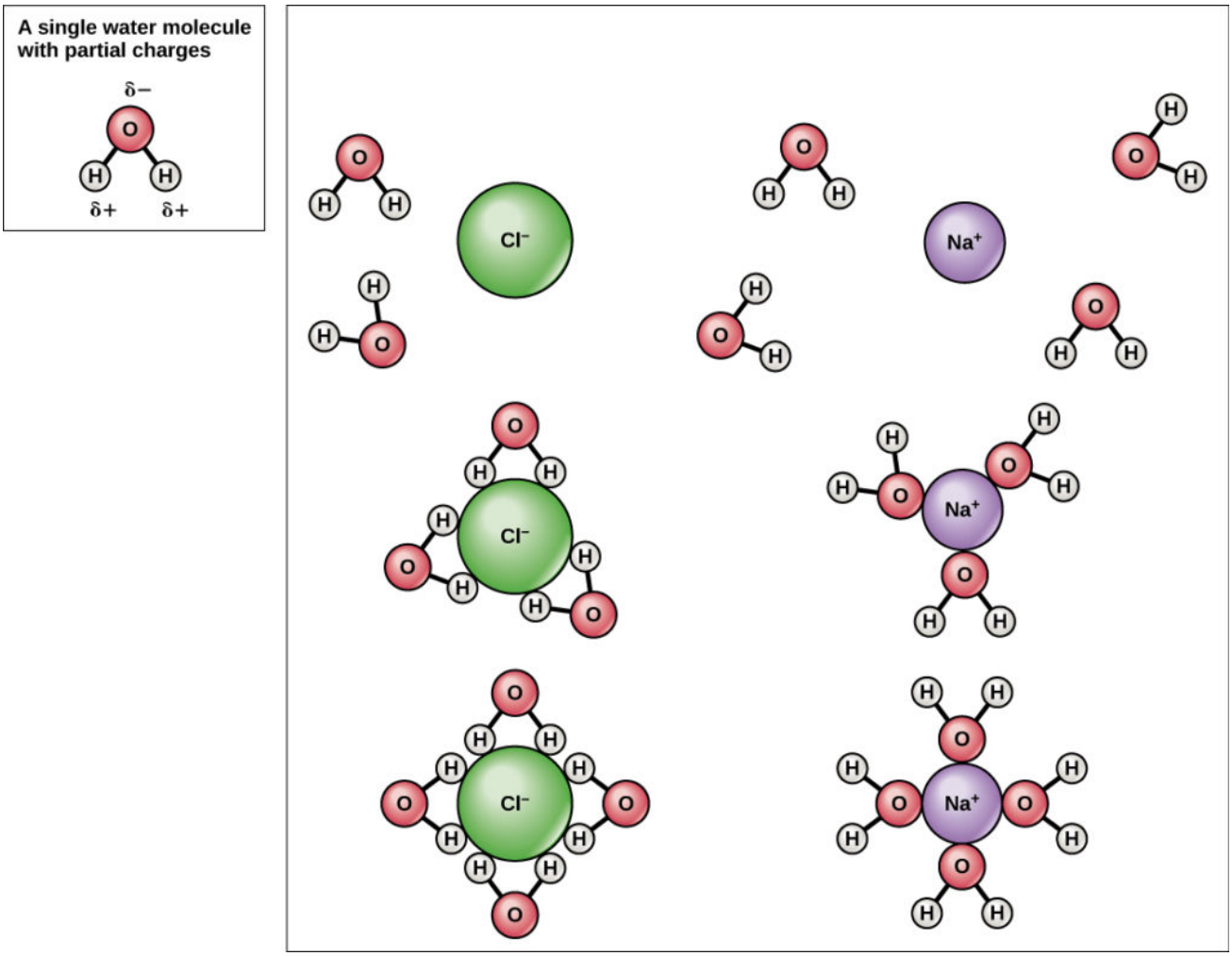

Figure 4. When table salt (NaCl) is mixed in water, spheres of hydration form around the ions.

A positively charged sodium ion is surrounded by the partially negative charges of oxygen atoms in water molecules. A negatively charged chloride ion is surrounded by the partially positive charges of hydrogen atoms in

water molecules. These spheres of hydration are also referred to as hydration shells. The polarity of the water molecule makes it an effective solvent and is important in its many roles in living systems.

Water Is Cohesive

Have you ever filled up a glass of water to the very top and then slowly added a few more drops? Before it overflows, the water actually forms a dome-like shape above the rim of the glass. This water can stay above the glass because of the property of cohesion. In cohesion, water molecules are attracted to each other (because of hydrogen bonding), keeping the molecules together at the liquid-air (gas) interface, although there is no more room in the glass. Cohesion gives rise to surface tension, the capacity of a substance to withstand rupture when placed under tension or stress. When you drop a small scrap of paper onto a droplet of water, the paper floats on top of the water droplet, although the object is denser (heavier) than the water. This occurs because of the surface tension that is created by the water molecules. Cohesion and surface tension keep the water molecules intact and the item floating on the top. It is even possible to "float" a steel needle on top of a glass of water if you place it gently, without breaking the surface tension (Figure 5).

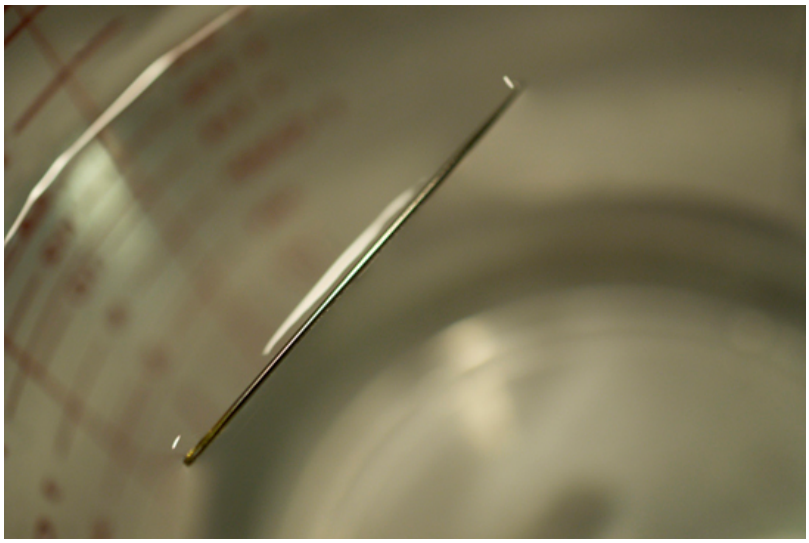

Figure 5. The weight of a needle on top of water pulls the surface tension downward; at the same time, the surface tension of the water is pulling it up, suspending the needle on the surface of the water and keeping it from sinking. Notice the indentation in the water around the needle. (credit: Cory Zanker)

These cohesive forces are also related to the water's property of adhesion, or the attraction between water molecules and other molecules. This is observed when water "climbs" up a straw placed in a glass of water. You will notice that the water appears to be higher on the sides of the straw than in the middle. This is because the water molecules are attracted to the straw and therefore adhere to it.

Cohesive and adhesive forces are important for sustaining life. For example, because of these forces, water can flow up from the roots to the tops of plants to feed the plant.

Video Review

Watch this video online: https://youtu.be/HVT3Y3_gHGg

Practice Question

Which of the following statements is **not** true?

a. Water is polar.
b. Water stabilizes temperature.
c. Water is essential for life.
d. Water is the most abundant atom in Earth's atmosphere.

Answer

Statement d is not true. Water is not the most abundant atom in Earth's atmosphere—nitrogen is.

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INTRODUCTION TO THE PH SCALE

What you'll learn to do: Demonstrate familiarity with the pH scale

Most people are familiar with the words *acid* and *acidic*—whether it's because of acid rain or acidic foods like lemon juice. However, fewer people are aware of acid's opposite: *base* (also called *alkaline*). Basic substances include things like baking soda, soap, and bleach. Distilled water is a neutral substance. The pH scale, which measures from 0 to 14, provides an indication of just how acidic or basic a substance is.

Most parts of our body (excluding things like stomach acid) measure around 7.2 and 7.6 on the pH scale (a 7 is neutral on the scale). If foreign strong substances dramatically change this pH, our bodies can no longer function properly.

In this outcome, we'll learn about acids and bases, and what impact they can have on living systems.

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BUFFERS, PH, ACIDS, AND BASES

Learning Outcomes

- Identify the characteristics of acids
- Identify the characteristics of bases
- Define buffers and discuss the role they play in human biology

The pH scale ranges from 0 to 14. The pH of a solution is a measure of its acidity or alkalinity (base). You have probably used litmus paper, paper that has been treated with a natural water-soluble dye so it can be used as a pH indicator, to test how much acid or base (alkalinity) exists in a solution. You might have even used some to make sure the water in an outdoor swimming pool is properly treated.

This pH test measures the amount of hydrogen ions that exists in a given solution. High concentrations of hydrogen ions yield a low pH (acidic substances), whereas low levels of hydrogen ions result in a high pH (basic substances). The overall concentration of hydrogen ions is inversely related to its pH and can be measured on the pH scale (Figure 1). Therefore, the more hydrogen ions present, the lower the pH; conversely, the fewer hydrogen ions, the higher the pH. A change of one unit on the pH scale represents a change in the concentration of hydrogen ions by a factor of 10, a change in two units represents a change in the concentration of hydrogen ions by a factor of 100. Thus, small changes in pH represent large changes in the concentrations of hydrogen ions. Pure water is neutral. It is neither acidic nor basic, and has a pH of 7.0. Anything below 7.0 (ranging from 0.0 to 6.9) is acidic, and anything above 7.0 (from 7.1 to 14.0) is alkaline. The blood in your veins is slightly alkaline (pH = 7.4). The environment in your stomach is highly acidic (pH = 1 to 2). Orange juice is mildly acidic (pH = approximately 3.5), whereas baking soda is basic (pH = 9.0).

Acids are substances that provide hydrogen ions (H^+) and lower pH, whereas bases provide hydroxide ions (OH^-) and raise pH. The stronger the acid, the more readily it donates H^+. For example, hydrochloric acid and lemon juice are very acidic and readily give up H^+ when added to water. Conversely, bases are those substances that readily donate OH^-. The OH^- ions combine with H^+ to produce water, which raises a substance's pH. Sodium hydroxide and many household cleaners are very alkaline and give up OH^- rapidly when placed in water, thereby raising the pH.

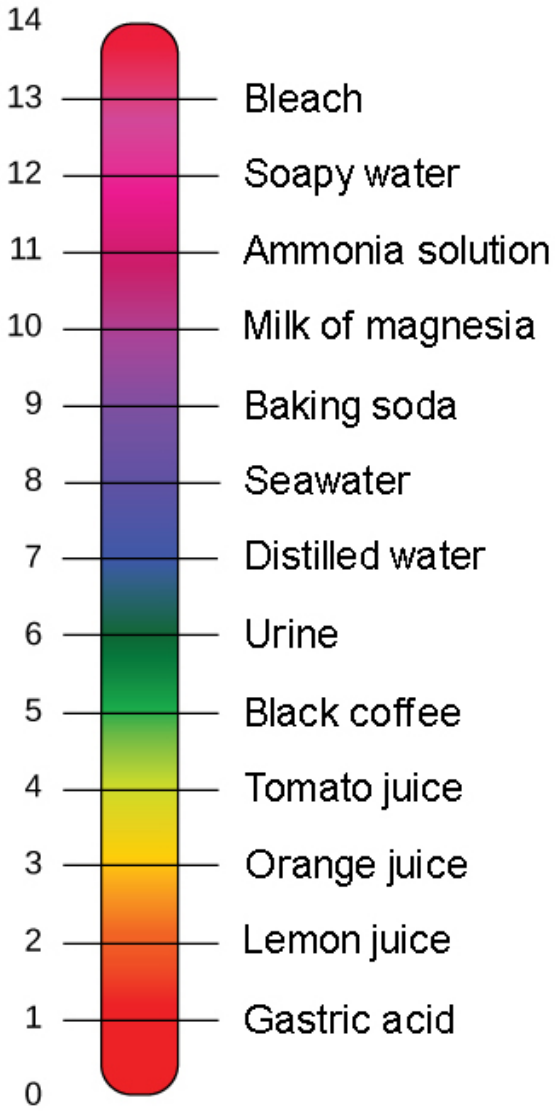

Figure 1. The pH scale measures the amount of hydrogen ions (H^+) in a substance. (credit: modification of work by Edward Stevens)

Buffers

Most cells in our bodies operate within a very narrow window of the pH scale, typically ranging only from 7.2 to 7.6. If the pH of the body is outside of this range, the respiratory system malfunctions, as do other organs in the body. Cells no longer function properly, and proteins will break down. Deviation outside of the pH range can induce coma or even cause death.

So how is it that we can ingest or inhale acidic or basic substances and not die? Buffers are the key. Buffers readily absorb excess H^+ or OH^-, keeping the pH of the body carefully maintained in the aforementioned narrow range. Carbon dioxide is part of a prominent buffer system in the human body; it keeps the pH within the proper range. This buffer system involves carbonic acid (H_2CO_3) and bicarbonate (HCO_3^-) anion. If too much H^+ enters the body, bicarbonate will combine with the H^+ to create carbonic acid and limit the decrease in pH.

Likewise, if too much OH^- is introduced into the system, carbonic acid will rapidly dissociate into bicarbonate and H^+ ions. The H^+ ions can combine with the OH^- ions, limiting the increase in pH. While carbonic acid is an important product in this reaction, its presence is fleeting because the carbonic acid is released from the body as carbon dioxide gas each time we breathe. Without this buffer system, the pH in our bodies would fluctuate too much and we would fail to survive.

In Summary: Buffers, pH, Acids, and Bases

The pH of a solution is a measure of the concentration of hydrogen ions in the solution. A solution with a high number of hydrogen ions is acidic and has a low pH value. A solution with a high number of hydroxide ions is basic and has a high pH value. The pH scale ranges from 0 to 14, with a pH of 7 being neutral. Buffers are solutions that moderate pH changes when an acid or base is added to the buffer system. Buffers are important in biological systems because of their ability to maintain constant pH conditions.

Practice Question

Using a pH meter, you find the pH of an unknown solution to be 8.0. How would you describe this solution?

a. weakly acidic
b. strongly acidic
c. weakly basic
d. strongly basic

Answer

This solution is weakly basic. Remember, a pH of 7.0 is neutral. Anything above that (7–14) is acidic, and anything below that (0–6) is basic.

Practice Question

The pH of lemon juice is about 2.0, whereas tomato juice's pH is about 4.0. Approximately how much of an increase in hydrogen ion concentration is there between tomato juice and lemon juice?

a. 2 times
b. 10 times
c. 100 times
d. 1000 times

Answer

Lemon juice is 100 times as acidic as tomato juice. Remember, each step in the pH scale represents a change in concentration by a factor of 10. Since tomato juice has a pH of 4.0, and lemon juice has a pH of 2.0, the concentration would change by 10 times 10.

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PUTTING IT TOGETHER: CHEMISTRY OF LIFE

As we've just learned, chemistry is essential to life: we are all made of compounds and molecules. Think back to the beginning of this chapter, where we briefly discussed the term *carbon-based life*. In the video below, we'll learn about why carbon is so essential to life:

Watch this video online: https://youtu.be/QnQe0xW_JY4

Occupation Spotlight: Nutritionists

Let's look back to our earlier occupation spotlight on the nutritionist. You should now be able see how an understanding of chemistry is essential to this job: a nutritionist needs to understand how the body builds complex molecules from the food a person ingests. A nutritionist also should understand how energy is used and moved about in the body. All this requires a basic understanding of elements, their structures, and how they interact.

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MODULE 3: IMPORTANT BIOLOGICAL MACROMOLECULES

WHY IT MATTERS: IMPORTANT BIOLOGICAL MACROMOLECULES

Why learn about the four main classes of important biological macromolecules?

Fad diets: we've all heard about them and maybe followed one or two in our lives. These diets have strict rules, and often have restrictions on eating a certain thing like fats or carbs (carbohydrates).

There are several things to critically consider about this type of diet. First off, is it even possible for a person to cut all carbs out of his or her diet? More importantly, is it actually healthy to remove an entire class of molecules from the diet? Fats aren't really important right? Certainly cholesterol is bad—right?

Before you decide to swear off carbs or fats, you should know that these types of foods are named after the kind of molecules that build them. Then you should learn what they actually do in cells.

Biological macromolecules are large molecules, necessary for life, that are built from smaller organic molecules. There are four major classes of biological macromolecules: carbohydrates, lipids, proteins, and nucleic acids (found in DNA and RNA). Each is an important component of the cell and performs a wide array of functions. Combined, these molecules make up the majority of a cell's mass. Biological macromolecules are organic, meaning that they contain carbon. In addition, they may contain hydrogen, oxygen, nitrogen, phosphorus, sulfur, and additional minor elements. We'll discuss each class and how they compare to each other.

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INTRODUCTION TO CARBON

What you'll learn to do: Discuss why it is said that life is carbon-based and the bonding properties of carbon

You've probably heard the term *carbon-based life* thrown about in scientific conversations before. But what exactly does the term mean? Possibly the quickest answer to this question is simply that all living things are reliant on molecules that include carbon. There are no living things on our planet that do not have carbon (however, there are nonliving things made up of carbon as well: e.g, diamonds and, well, carbon itself).

In this outcome we'll learn about the importance of carbon to every living thing on earth and its unique properties that make it particularly suited for its role in living things.

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CARBON AND CARBON BONDING

Learning Outcomes

Discuss why it is said that life is carbon-based and the bonding properties of carbon.

Carbon

Living things are carbon-based because carbon plays such a prominent role in the chemistry of living things. This means that carbon atoms, bonded to other carbon atoms or other elements, form the fundamental components of many, if not most, of the molecules found uniquely in living things. Other elements play important roles in biological molecules, but carbon certainly qualifies as the "foundation" element for molecules in living things. It is the bonding properties of carbon atoms that are responsible for its important role.

Carbon Bonding

The four covalent bonding positions of the carbon atom can give rise to a wide diversity of compounds with many functions, accounting for the importance of carbon in living things.

Carbon contains four electrons in its outer shell. Therefore, it can form four covalent bonds with other atoms or molecules. The simplest organic carbon molecule is methane (CH_4), in which four hydrogen atoms bind to a carbon atom (Figure 1).

However, structures that are more complex are made using carbon. Any of the hydrogen atoms can be replaced with another carbon atom covalently bonded to the first carbon atom. In this way, long and branching chains of carbon compounds can be made (Figure 2**a**). The carbon atoms may bond with atoms of other elements, such as nitrogen, oxygen, and phosphorus (Figure 2**b**). The molecules may also form rings, which themselves can link with other rings (Figure 2**c**). This diversity of molecular forms accounts for the diversity of functions of the biological macromolecules and is based to a large degree on the ability of carbon to form multiple bonds with itself and other atoms.

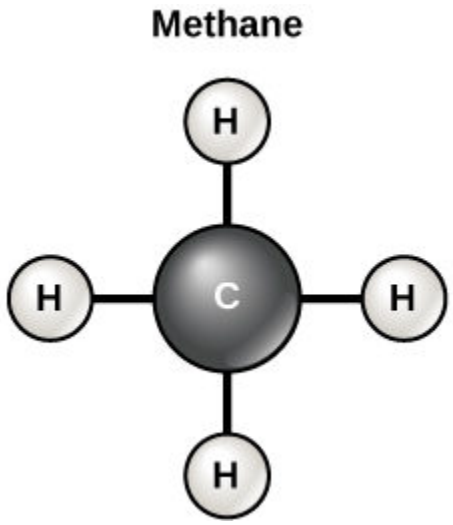

Figure 1. Carbon can form four covalent bonds to create an organic molecule. The simplest carbon molecule is methane (CH_4), depicted here.

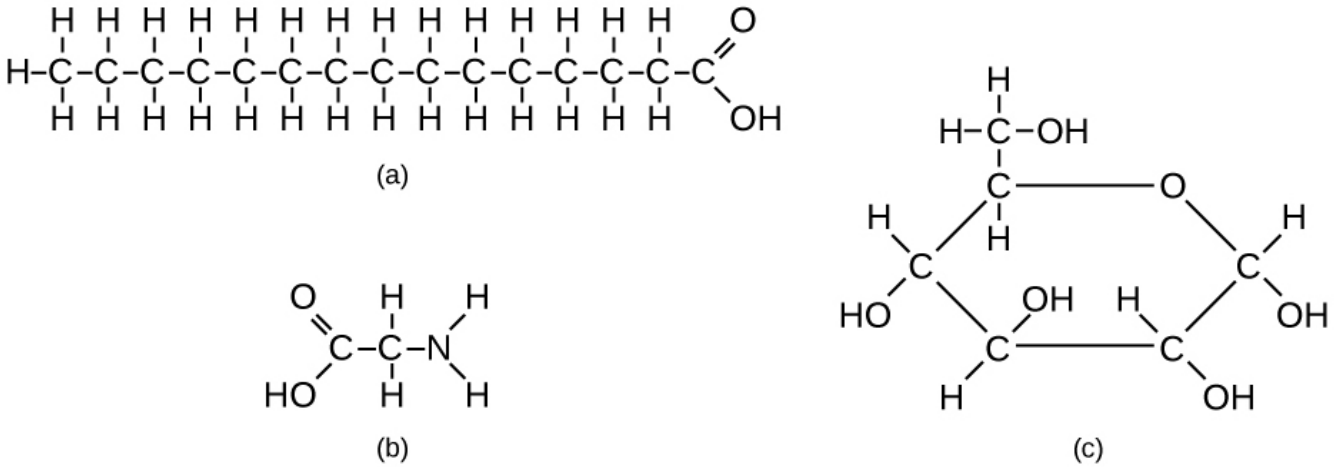

Figure 2. These examples show three molecules (found in living organisms) that contain carbon atoms bonded in various ways to other carbon atoms and the atoms of other elements. (a) This molecule of stearic acid has a long chain of carbon atoms. (b) Glycine, a component of proteins, contains carbon, nitrogen, oxygen, and hydrogen atoms. (c) Glucose, a sugar, has a ring of carbon atoms and one oxygen atom.

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INTRODUCTION TO CARBOHYDRATES

What you'll learn to do: Summarize the roles carbohydrates play in biological systems

Are carbohydrates good for you? People who wish to lose weight are often told that carbohydrates are bad for them and should be avoided. Some diets completely forbid carbohydrate consumption, claiming that a low-carbohydrate diet helps people to lose weight faster. However, carbohydrates have been an important part of the human diet for thousands of years; artifacts from ancient civilizations show the presence of wheat, rice, and corn in our ancestors' storage areas.

Carbohydrates should be supplemented with proteins, vitamins, and fats to be parts of a well-balanced diet. Calorie-wise, a gram of carbohydrate provides 4.3 Kcal. For comparison, fats provide 9 Kcal/g, a less desirable ratio. Carbohydrates contain soluble and insoluble elements; the insoluble part is known as fiber, which is mostly cellulose. Fiber has many uses; it promotes regular bowel movement by adding bulk, and it regulates the rate of consumption of blood glucose. Fiber also helps to remove excess cholesterol from the body. In addition, a meal containing whole grains and vegetables gives a feeling of fullness. As an immediate source of energy, glucose is broken down during the process of cellular respiration, which produces ATP, the energy currency of the cell. Without the consumption of carbohydrates, the availability of "instant energy" would be reduced.

Eliminating carbohydrates from the diet is not the best way to lose weight. A low-calorie diet that is rich in whole grains, fruits, vegetables, and lean meat, together with plenty of exercise and plenty of water, is the more sensible way to lose weight.

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STRUCTURE AND FUNCTION OF CARBOHYDRATES

Learning Outcomes

- Distinguish between monosaccharides, disaccharides, and polysaccharides
- Identify several major functions of carbohydrates

Most people are familiar with carbohydrates, one type of macromolecule, especially when it comes to what we eat. To lose weight, some individuals adhere to "low-carb" diets. Athletes, in contrast, often "carb-load" before important competitions to ensure that they have enough energy to compete at a high level. Carbohydrates are, in fact, an essential part of our diet; grains, fruits, and vegetables are all natural sources of carbohydrates. Carbohydrates provide energy to the body, particularly through glucose, a simple sugar that is a component of starch and an ingredient in many staple foods. Carbohydrates also have other important functions in humans, animals, and plants.

Molecular Structures

Carbohydrates can be represented by the formula $(CH_2O)_n$, where n is the number of carbons in the molecule. In other words, the ratio of carbon to hydrogen to oxygen is 1:2:1 in carbohydrate molecules. This formula also explains the origin of the term "carbohydrate": the components are carbon ("carbo") and the components of water (hence, "hydrate"). Carbohydrates are classified into three subtypes: monosaccharides, disaccharides, and polysaccharides.

Monosaccharides

Monosaccharides (*mono–* = "one"; *sacchar–* = "sweet") are simple sugars, the most common of which is glucose. In monosaccharides, the number of carbons usually ranges from three to seven. Most monosaccharide names end with the suffix *–ose*. If the sugar has an aldehyde group (the functional group with the structure R-CHO), it is known as an aldose, and if it has a ketone group (the functional group with the structure RC(=O)R′), it is known as a ketose. Depending on the number of carbons in the sugar, they also may be known as trioses (three carbons), pentoses (five carbons), and or hexoses (six carbons). See Figure 1 for an illustration of the monosaccharides.

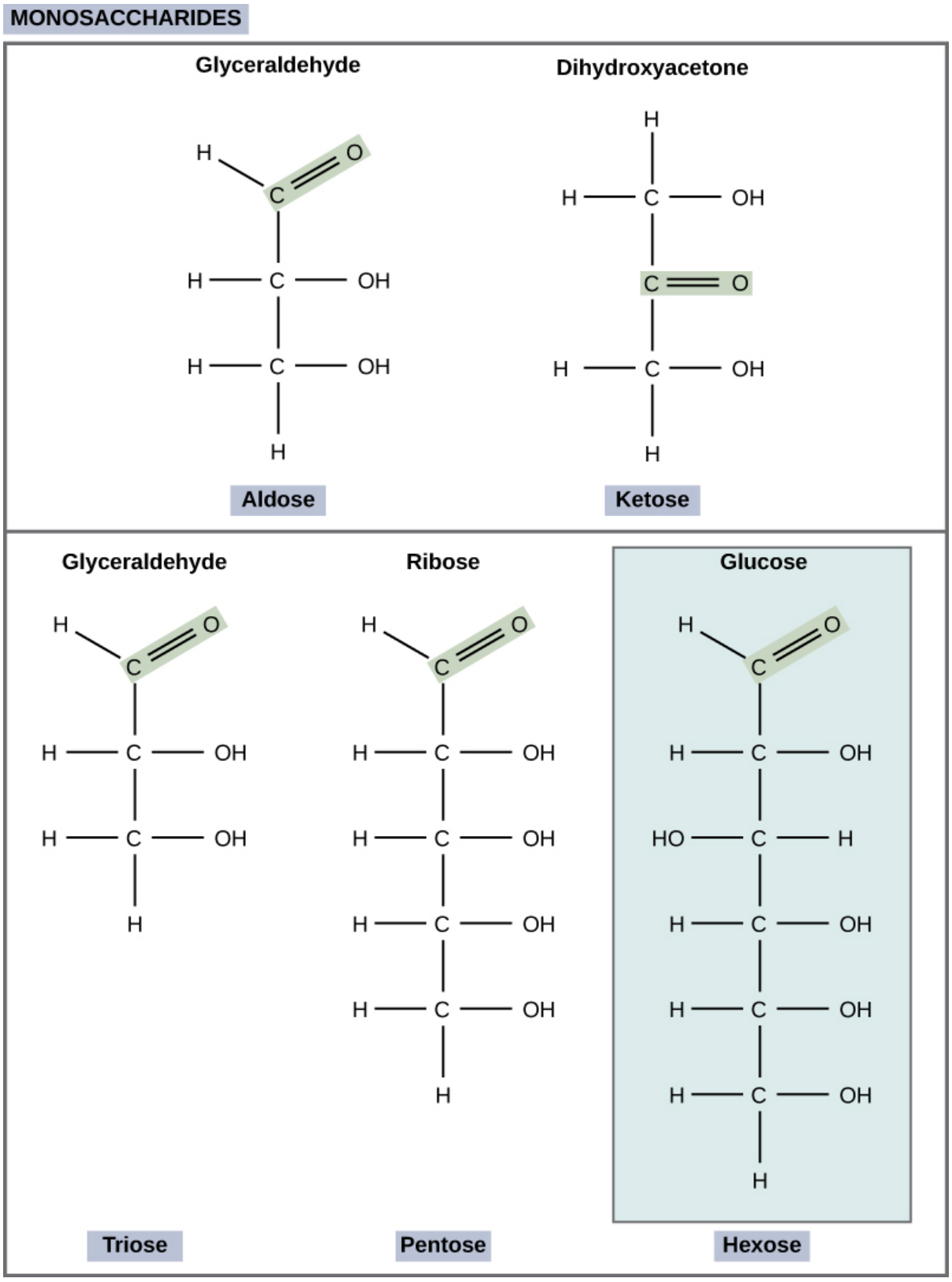


Figure 1. Monosaccharides are classified based on the position of their carbonyl group and the number of carbons in the backbone. Aldoses have a carbonyl group (indicated in green) at the end of the carbon chain, and ketoses have a carbonyl group in the middle of the carbon chain. Trioses, pentoses, and hexoses have three, five, and six carbon backbones, respectively.

The chemical formula for glucose is $C_6H_{12}O_6$. In humans, glucose is an important source of energy. During cellular respiration, energy is released from glucose, and that energy is used to help make adenosine triphosphate (ATP). Plants synthesize glucose using carbon dioxide and water, and glucose in turn is used for energy requirements for the plant. Excess glucose is often stored as starch that is catabolized (the breakdown of larger molecules by cells) by humans and other animals that feed on plants.

Galactose (part of lactose, or milk sugar) and fructose (part of sucrose, or fruit sugar) are other common monosaccharides. Although glucose, galactose, and fructose all have the same chemical formula ($C_6H_{12}O_6$), they differ structurally and chemically (and are known as isomers) because of the different arrangement of functional groups around the asymmetric carbon; all of these monosaccharides have more than one asymmetric carbon.

Monosaccharides can exist as a linear chain or as ring-shaped molecules; in aqueous solutions they are usually found in ring forms.

Disaccharides

Disaccharides (*di–* = "two") form when two monosaccharides undergo a dehydration reaction (also known as a condensation reaction or dehydration synthesis). During this process, the hydroxyl group of one monosaccharide combines with the hydrogen of another monosaccharide, releasing a molecule of water and forming a covalent bond (Figure 2).

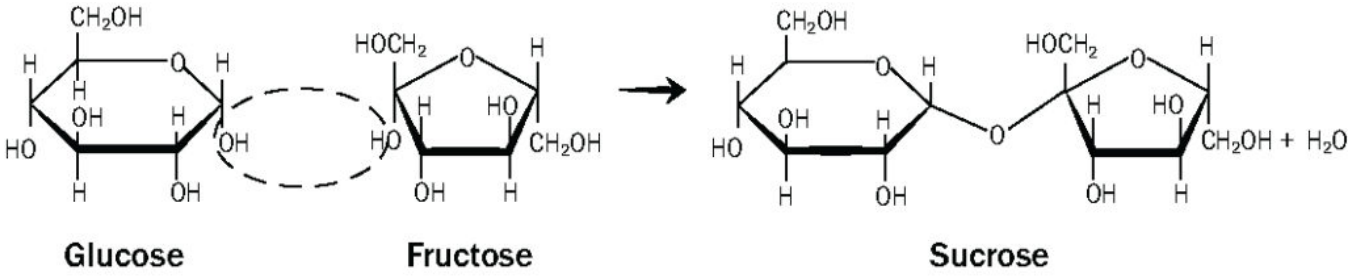


Figure 2. Sucrose is produced from the chemical reaction between two simple sugars called glucose and fructose.

Common disaccharides include lactose, maltose, and sucrose. Lactose is a disaccharide consisting of the monomers glucose and galactose. It is found naturally in milk. Maltose, or malt sugar, is a disaccharide formed by a dehydration reaction between two glucose molecules. The most common disaccharide is sucrose, or table sugar, which is composed of the monomers glucose and fructose.

Polysaccharides

A long chain of monosaccharides linked by covalent bonds is known as a polysaccharide (*poly–* = "many"). The chain may be branched or unbranched, and it may contain different types of monosaccharides. Polysaccharides may be very large molecules. Starch, glycogen, cellulose, and chitin are examples of polysaccharides.

Starch is the stored form of sugars in plants and is made up of amylose and amylopectin (both polymers of glucose). Plants are able to synthesize glucose, and the excess glucose is stored as starch in different plant parts, including roots and seeds. The starch that is consumed by animals is broken down into smaller molecules, such as glucose. The cells can then absorb the glucose.

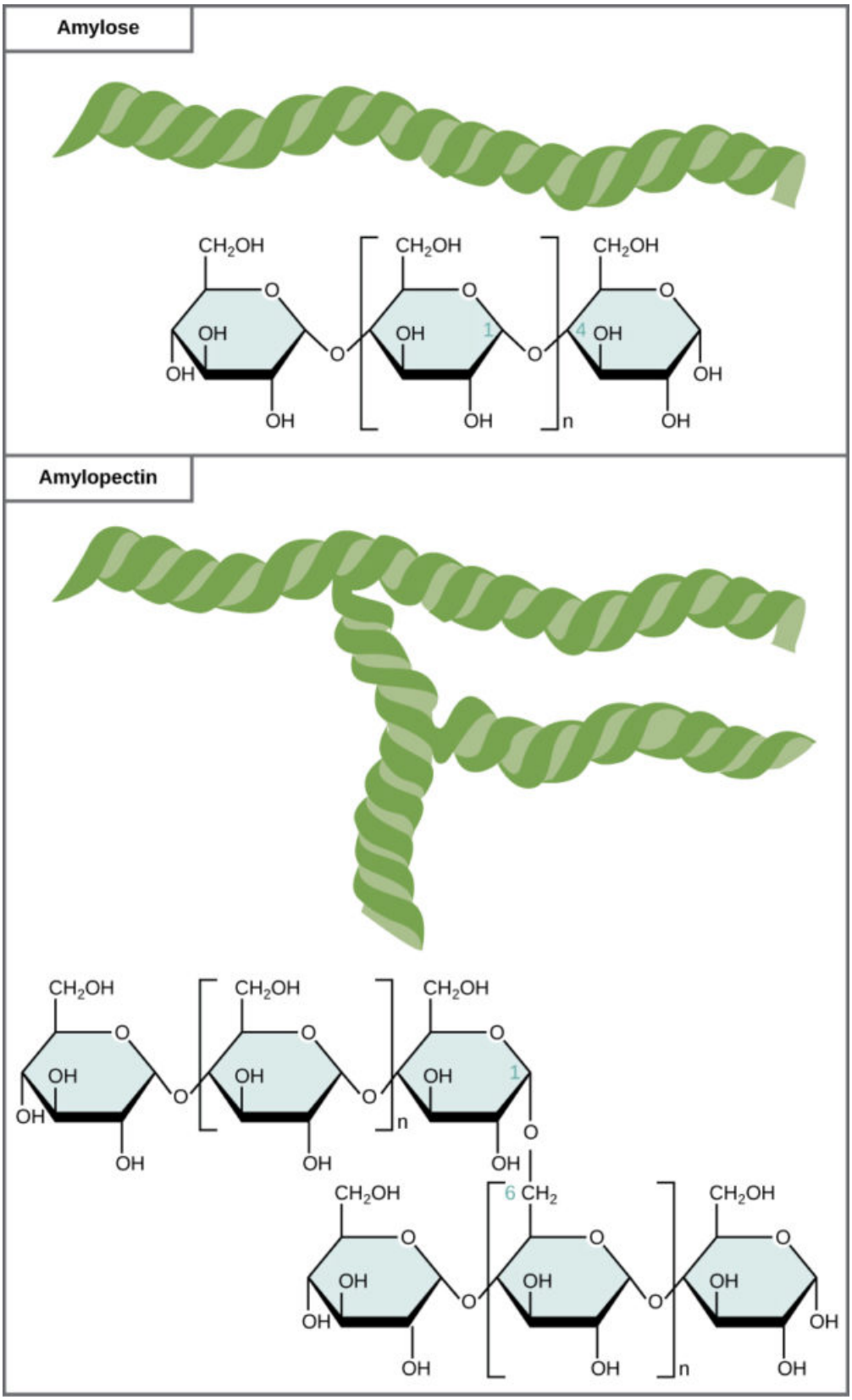


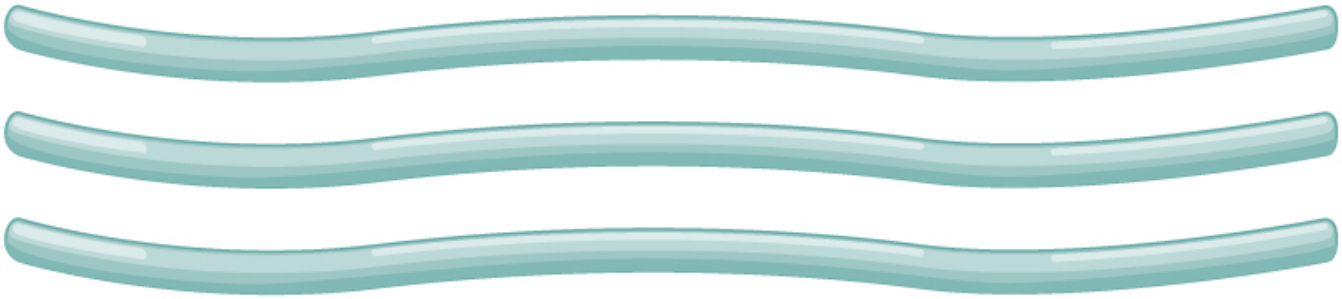
Figure 3. Amylose and amylopectin are two different forms of starch. Glycogen is the storage form of glucose in humans and other vertebrates and is made up of monomers of glucose.

Glycogen is the storage form of glucose in humans and other vertebrates, and is made up of monomers of glucose. Glycogen is the animal equivalent of starch and is a highly branched molecule usually stored in liver and muscle cells. Whenever glucose levels decrease, glycogen is broken down to release glucose.

Cellulose is one of the most abundant natural biopolymers. The cell walls of plants are mostly made of cellulose, which provides structural support to the cell. Wood and paper are mostly cellulosic in nature. Cellulose is made up of glucose monomers that are linked by bonds between particular carbon atoms in the glucose molecule.

Every other glucose monomer in cellulose is flipped over and packed tightly as extended long chains. This gives cellulose its rigidity and high tensile strength—which is so important to plant cells. Cellulose passing through our digestive system is called dietary fiber. While the glucose-glucose bonds in cellulose cannot be broken down by human digestive enzymes, herbivores such as cows, buffalos, and horses are able to digest grass that is rich in cellulose and use it as a food source. In these animals, certain species of bacteria reside in the rumen (part of the digestive system of herbivores) and secrete the enzyme cellulase. The appendix also contains bacteria that break down cellulose, giving it an important role in the digestive systems of ruminants. Cellulases can break down cellulose into glucose monomers that can be used as an energy source by the animal.

Cellulose fibers



Cellulose structure

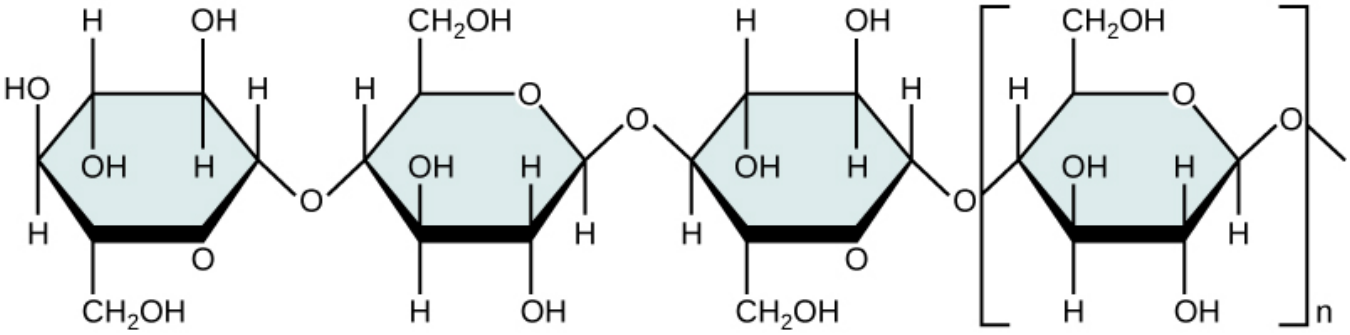


Figure 4. In cellulose, glucose monomers are linked in unbranched chains by β 1-4 glycosidic linkages. Because of the way the glucose subunits are joined, every glucose monomer is flipped relative to the next one resulting in a linear, fibrous structure.

As shown in Figure 4, every other glucose monomer in cellulose is flipped over, and the monomers are packed tightly as extended long chains. This gives cellulose its rigidity and high tensile strength—which is so important to plant cells.

Figure 5. Insects have a hard outer exoskeleton made of chitin, a type of polysaccharide.

Carbohydrates serve other functions in different animals. Arthropods, such as insects, spiders, and crabs, have an outer skeleton, called the exoskeleton, which protects their internal body parts. This exoskeleton is made of the biological macromolecule chitin, which is a nitrogenous carbohydrate. It is made of repeating units of a modified sugar containing nitrogen.

Registered Dietitian

Obesity is a worldwide health concern, and many diseases, such as diabetes and heart disease, are becoming more prevalent because of obesity. This is one of the reasons why registered dietitians are increasingly sought after for advice. Registered dietitians help plan food andnutrition programs for individuals in various settings. They often work with patients in health-care facilities, designing nutrition plans to prevent and treat diseases. For example, dietitians may teach a patient with diabetes how to manage blood-sugar levels by eating the correct types and amounts of carbohydrates. Dietitians may also work in nursing homes, schools, and private practices.

To become a registered dietitian, one needs to earn at least a bachelor's degree in dietetics, nutrition, food technology, or a related field. In addition, registered dietitians must complete a supervised internship program and pass a national exam. Those who pursue careers in dietetics take courses in nutrition, chemistry, biochemistry, biology, microbiology, and human physiology. Dietitians must become experts in the chemistry and functions of food (proteins, carbohydrates, and fats).

Figure 6. Registered Dietitian Nutritionist (RDN)Chef Brenda Thompson works with foodservice staff to assemble her breakfast burrito recipe during the chef designed school taste testing in Idaho. Thanks to a U.S. Department of Agriculture (USDA) Team Nutrition grant RDN Chef Brenda Thompson, developed recipes for the Chef Designed School Lunch cookbook.

In Summary: Structure and Function of Carbohydrates

Carbohydrates are a group of macromolecules that are a vital energy source for the cell and provide structural support to plant cells, fungi, and all of the arthropods that include lobsters, crabs, shrimp, insects, and spiders. Carbohydrates are classified as monosaccharides, disaccharides, and polysaccharides depending on the number of monomers in the molecule. Monosaccharides are linked by glycosidic bonds that are formed as a result of dehydration reactions, forming disaccharides and polysaccharides with the elimination of a water molecule for each bond formed. Glucose, galactose, and fructose are common monosaccharides, whereas common disaccharides include lactose, maltose, and sucrose. Starch and glycogen, examples of polysaccharides, are the storage forms of glucose in plants and animals, respectively. The long polysaccharide chains may be branched or unbranched. Cellulose is an example of an unbranched polysaccharide, whereas amylopectin, a constituent of starch, is a highly branched molecule. Storage of

glucose, in the form of polymers like starch of glycogen, makes it slightly less accessible for metabolism; however, this prevents it from leaking out of the cell or creating a high osmotic pressure that could cause excessive water uptake by the cell.

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- Formation of sucrose. **Authored by**: Anthony Fernandez. **Provided by**: American Chemical Society . **Located at**: https://www.acs.org/content/acs/en/education/resources/highschool/chemmatters/past-issues/archive-2014-2015/candymaking.html. **License**: *CC BY: Attribution*

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INTRODUCTION TO LIPIDS

What you'll learn to do: Illustrate different types of lipids and relate their structure to their role in biological systems

Fats and oils are probably the type of lipid that you're most familiar with in your everyday life. The word *fat* typically brings up a negative picture in our minds. In diets, we're advised to stay away from fatty foods. However, our bodies require some fat in order to survive. There are also other lipids essential to human life, including phospholipids, steroids, and waxes.

While an excess of any substance can be a problem, all of these lipids play essential roles in living things.

In this outcome, we will discuss lipids and the role they plan in our bodies.

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LIPIDS

Learning Outcomes

- Distinguish between the different kinds of lipids
- Identify several major functions of lipids

Lipids include a diverse group of compounds that are largely nonpolar in nature. This is because they are hydrocarbons that include mostly nonpolar carbon–carbon or carbon–hydrogen bonds. Non-polar molecules are hydrophobic (“water fearing”), or insoluble in water. Lipids perform many different functions in a cell. Cells store energy for long-term use in the form of fats. Lipids also provide insulation from the environment for plants and animals (Figure 1). For example, they help keep aquatic birds and mammals dry when forming a protective layer over fur or feathers because of their water-repellant hydrophobic nature. Lipids are also the building blocks of many hormones and are an important constituent of all cellular membranes. Lipids include fats, oils, waxes, phospholipids, and steroids.

Figure 1. Hydrophobic lipids in the fur of aquatic mammals, such as this river otter, protect them from the elements. (credit: Ken Bosma)

Fats and Oils

A fat molecule, such as a triglyceride, consists of two main components—glycerol and fatty acids. Glycerol is an organic compound with three carbon atoms, five hydrogen atoms, and three hydroxyl (–OH) groups. Fatty acids have a long chain of hydrocarbons to which an acidic carboxyl group is attached, hence the name “fatty acid.” The number of carbons in the fatty acid may range from 4 to 36; most common are those containing 12–18 carbons. In a fat molecule, a fatty acid is attached to each of the three oxygen atoms in the –OH groups of the glycerol molecule with a covalent bond (Figure 2).

During this covalent bond formation, three water molecules are released. The three fatty acids in the fat may be similar or dissimilar. These fats are also called triglycerides because they have three fatty acids. Some fatty acids have common names that specify their origin. For example, palmitic acid, a saturated fatty acid, is derived from the palm tree. Arachidic acid is derived from *Arachis hypogaea*, the scientific name for peanuts.

Fatty acids may be saturated or unsaturated. In a fatty acid chain, if there are only single bonds between neighboring carbons in the hydrocarbon chain, the fatty acid is saturated. Saturated fatty acids are saturated with hydrogen; in other words, the number of hydrogen atoms attached to the carbon skeleton is maximized.

When the hydrocarbon chain contains a double bond, the fatty acid is an unsaturated fatty acid.

Most unsaturated fats are liquid at room temperature and are called oils. If there is one double bond in the molecule, then it is known as a monounsaturated fat (e.g., olive oil), and if there is more than one double bond, then it is known as a polyunsaturated fat (e.g., canola oil).

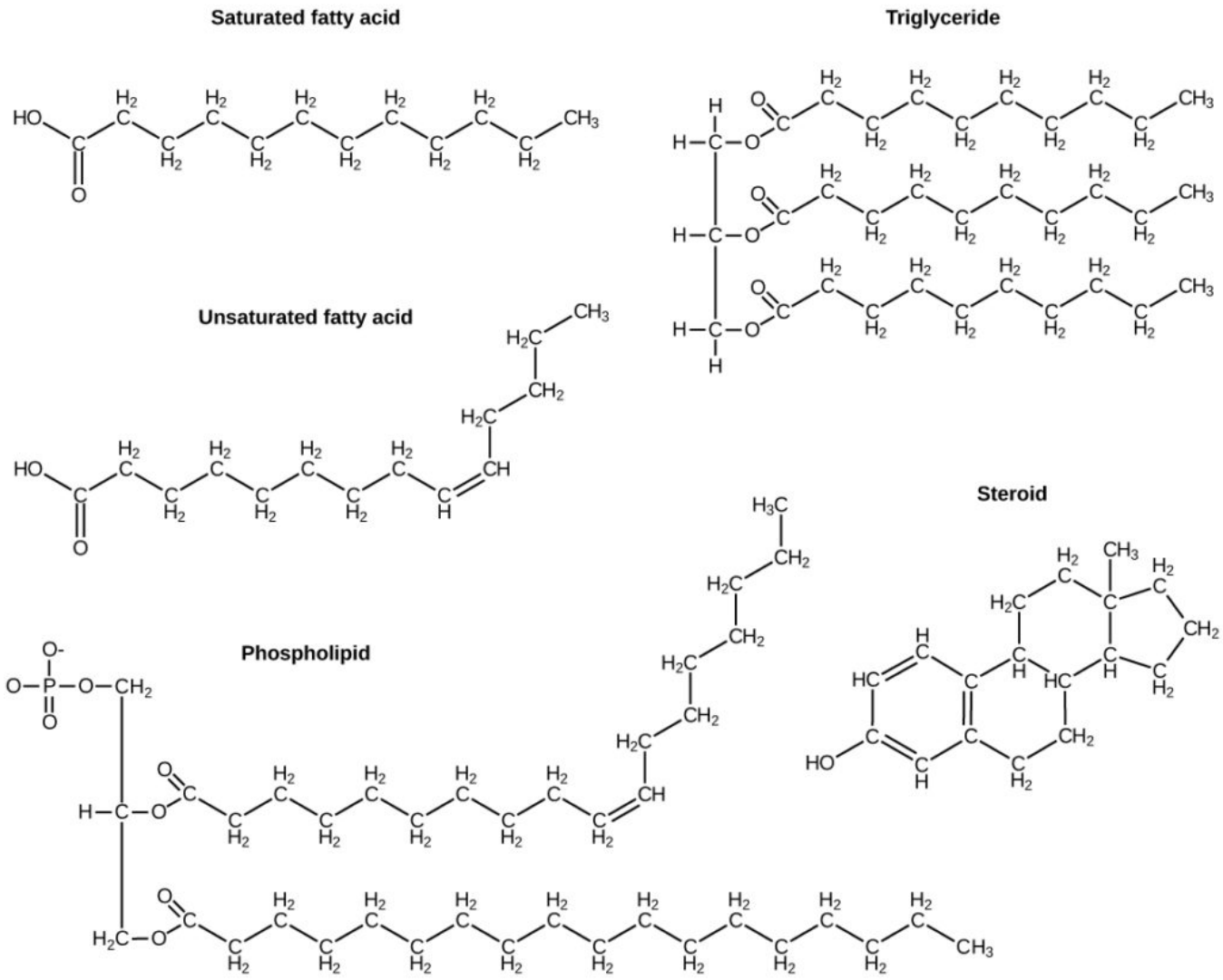


Figure 2. Lipids include fats, such as triglycerides, which are made up of fatty acids and glycerol, phospholipids, and steroids.

Saturated fats tend to get packed tightly and are solid at room temperature. Animal fats with stearic acid and palmitic acid contained in meat, and the fat with butyric acid contained in butter, are examples of saturated fats. Mammals store fats in specialized cells called adipocytes, where globules of fat occupy most of the cell. In plants, fat or oil is stored in seeds and is used as a source of energy during embryonic development.

Unsaturated fats or oils are usually of plant origin and contain unsaturated fatty acids. The double bond causes a bend or a “kink” that prevents the fatty acids from packing tightly, keeping them liquid at room temperature. Olive oil, corn oil, canola oil, and cod liver oil are examples of unsaturated fats. Unsaturated fats help to improve blood cholesterol levels, whereas saturated fats contribute to plaque formation in the arteries, which increases the risk of a heart attack.

Margarine, some types of peanut butter, and shortening are examples of artificially hydrogenated *trans*-fats. Recent studies have shown that an increase in *trans*-fats in the human diet may lead to an increase in levels of low-density lipoprotein (LDL), or “bad” cholesterol, which, in turn, may lead to plaque deposition in the arteries, resulting in heart disease. Many fast food restaurants have recently eliminated the use of *trans*-fats, and U.S. food labels are now required to list their *trans*-fat content.In the food industry, oils are artificially hydrogenated to make them semi-solid, leading to less spoilage and increased shelf life. Simply speaking, hydrogen gas is bubbled through oils to solidify them. During this hydrogenation process, double bonds of the *cis*-conformation in the hydrocarbon chain may be converted to double bonds in the *trans*-conformation. This forms a *trans*-fat from a *cis*-fat. The orientation of the double bonds affects the chemical properties of the fat (Figure 3).

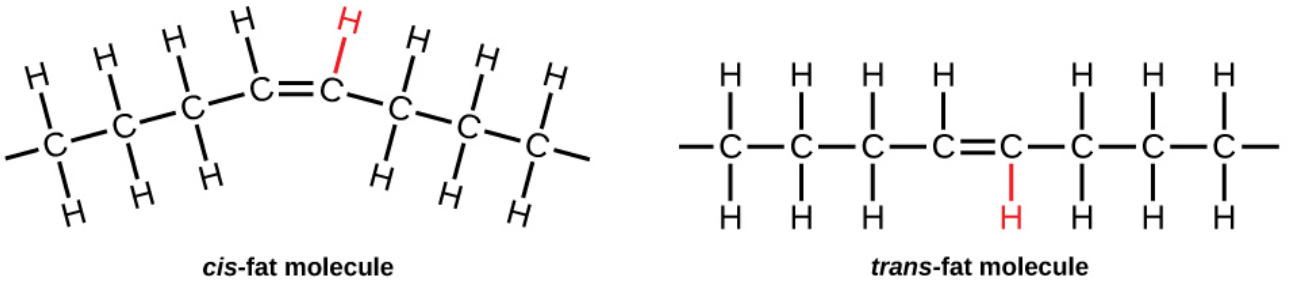


Figure 3. During the hydrogenation process, the orientation around the double bonds is changed, making a trans-fat from a cis-fat. This changes the chemical properties of the molecule.

Essential fatty acids are fatty acids that are required but not synthesized by the human body. Consequently, they must be supplemented through the diet. Omega-3 fatty acids fall into this category and are one of only two known essential fatty acids for humans (the other being omega-6 fatty acids). They are a type of polyunsaturated fat and are called omega-3 fatty acids because the third carbon from the end of the fatty acid participates in a double bond.

Salmon, trout, and tuna are good sources of omega-3 fatty acids. Omega-3 fatty acids are important in brain function and normal growth and development. They may also prevent heart disease and reduce the risk of cancer.

Like carbohydrates, fats have received a lot of bad publicity. It is true that eating an excess of fried foods and other “fatty” foods leads to weight gain. However, fats do have important functions. Fats serve as long-term energy storage. They also provide insulation for the body. Therefore, “healthy” unsaturated fats in moderate amounts should be consumed on a regular basis.

Phospholipids

Phospholipids are the major constituent of the plasma membrane. Like fats, they are composed of fatty acid chains attached to a glycerol or similar backbone. Instead of three fatty acids attached, however, there are two fatty acids and the third carbon of the glycerol backbone is bound to a phosphate group. The phosphate group is modified by the addition of an alcohol.

A phospholipid has both hydrophobic and hydrophilic regions. The fatty acid chains are hydrophobic and exclude themselves from water, whereas the phosphate is hydrophilic and interacts with water.

Cells are surrounded by a membrane, which has a bilayer of phospholipids. The fatty acids of phospholipids face inside, away from water, whereas the phosphate group can face either the outside environment or the inside of the cell, which are both aqueous.

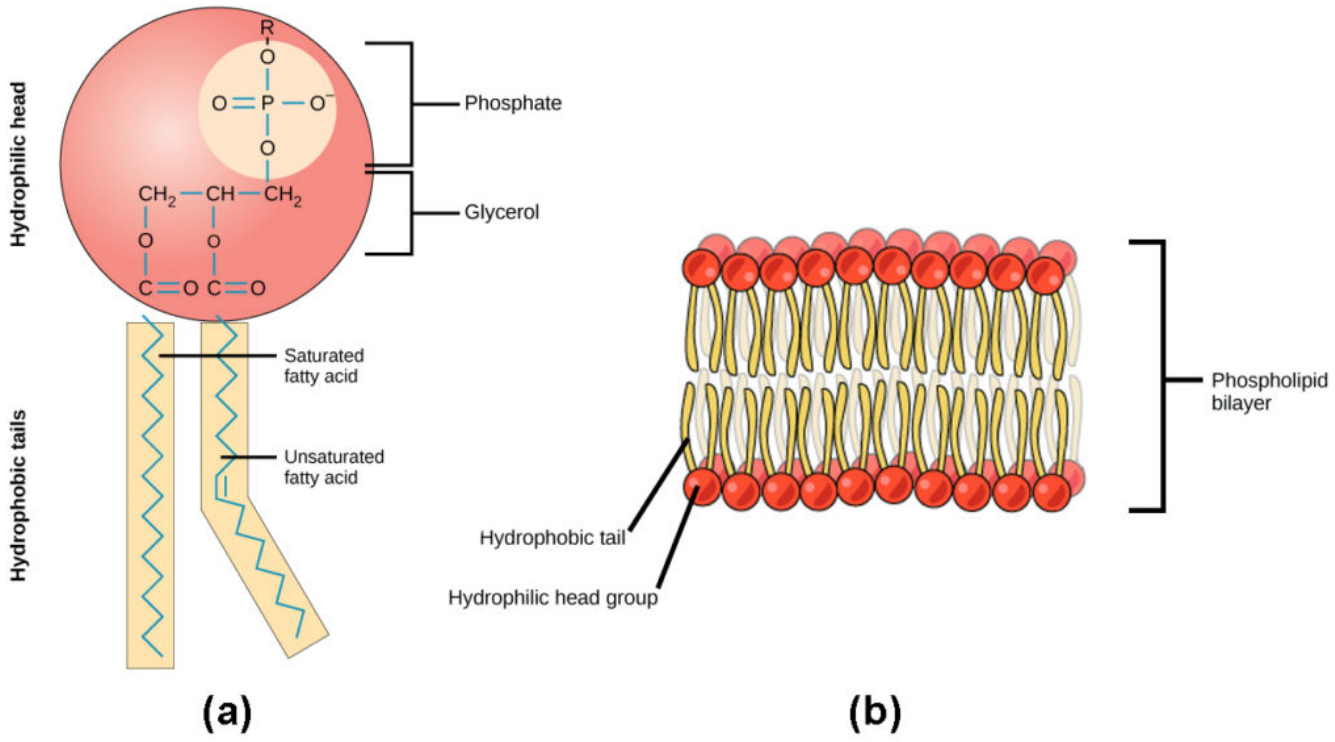


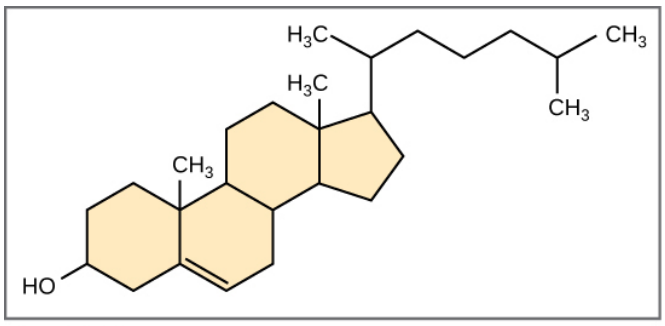
Figure 4. (a) A phospholipid is a molecule with two fatty acids and a modified phosphate group attached to a glycerol backbone. The phosphate may be modified by the addition of charged or polar chemical groups. Two chemical groups that may modify the phosphate, choline and serine, are shown here. Both choline and serine attach to the phosphate group at the position labeled R. (b) The phospholipid bilayer is the major component of all cellular membranes. The hydrophilic head groups of the phospholipids face the aqueous solution. The hydrophobic tails are sequestered in the middle of the bilayer.

Steroids and Waxes

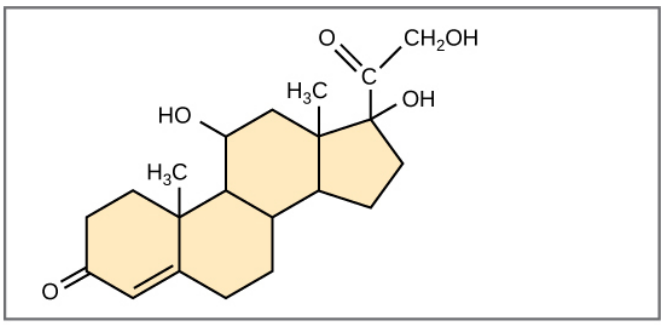
Unlike the phospholipids and fats discussed earlier, steroids have a ring structure. Although they do not resemble other lipids, they are grouped with them because they are also hydrophobic. All steroids have four, linked carbon rings and several of them, like cholesterol, have a short tail.

Cholesterol is a steroid. Cholesterol is mainly synthesized in the liver and is the precursor of many steroid hormones, such as testosterone and estradiol. It is also the precursor of vitamins E and K. Cholesterol is the precursor of bile salts, which help in the breakdown of fats and their subsequent absorption by cells. Although cholesterol is often spoken of in negative terms, it is necessary for the proper functioning of the body. It is a key component of the plasma membranes of animal cells.

Waxes are made up of a hydrocarbon chain with an alcohol (–OH) group and a fatty acid. Examples of animal waxes include beeswax and lanolin. Plants also have waxes, such as the coating on their leaves, that helps prevent them from drying out.



Cholesterol



Cortisol

Figure 5. Steroids such as cholesterol and cortisol are composed of four fused hydrocarbon rings

For an additional perspective on lipids, explore this interactive animation.

In Summary: Lipids

Lipids are a class of macromolecules that are nonpolar and hydrophobic in nature. Major types include fats and oils, waxes, phospholipids, and steroids. Fats are a stored form of energy and are also known as triacylglycerols or triglycerides. Fats are made up of fatty acids and either glycerol or sphingosine. Fatty acids may be unsaturated or saturated, depending on the presence or absence of double bonds in the hydrocarbon chain. If only single bonds are present, they are known as saturated fatty acids. Unsaturated fatty acids may have one or more double bonds in the hydrocarbon chain. Phospholipids make up the matrix of membranes. They have a glycerol or sphingosine backbone to which two fatty acid chains and a phosphate-containing group are attached. Steroids are another class of lipids. Their basic structure has four fused carbon rings. Cholesterol is a type of steroid and is an important constituent of the plasma membrane, where it helps to maintain the fluid nature of the membrane. It is also the precursor of steroid hormones such as testosterone.

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INTRODUCTION TO PROTEINS

What you'll learn to do: Describe the structure and function proteins

Proteins are polymers of amino acids. Each amino acid contains a central carbon, a hydrogen, a carboxyl group, an amino group, and a variable R group. The R group specifies which class of amino acids it belongs to: electrically charged hydrophilic side chains, polar but uncharged side chains, nonpolar hydrophobic side chains, and special cases.

Proteins have different "layers" of structure: primary, secondary, tertiary, quaternary.

Proteins have a variety of function in cells. Major functions include acting as enzymes, receptors, transport molecules, regulatory proteins for gene expression, and so on. Enzymes are biological catalysts that speed up a chemical reaction without being permanently altered. They have "active sites" where the substrate/reactant binds, and they can be either activated or inhibited (competitive and/or noncompetitive inhibitors).

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COMPONENTS OF PROTEINS

Learning Outcomes

Identify the component parts of proteins

Proteins are one of the most abundant organic molecules in living systems and have the most diverse range of functions of all macromolecules. Proteins may be structural, regulatory, contractile, or protective; they may serve in transport, storage, or membranes; or they may be toxins or enzymes. Each cell in a living system may contain thousands of different proteins, each with a unique function. Their structures, like their functions, vary greatly. They are all, however, polymers of amino acids, arranged in a linear sequence.

Proteins have different shapes and molecular weights; some proteins are globular in shape whereas others are fibrous in nature. For example, hemoglobin is a globular protein, but collagen, found in our skin, is a fibrous protein. Protein shape is critical to its function. Changes in temperature, pH, and exposure to chemicals may lead to permanent changes in the shape of the protein, leading to a loss of function or denaturation (to be discussed in more detail later). All proteins are made up of different arrangements of the same 20 kinds of amino acids.

Amino acids are the monomers that make up proteins. Each amino acid has the same fundamental structure, which consists of a central carbon atom bonded to an amino group ($–NH_2$), a carboxyl group (–COOH), and a hydrogen atom. Every amino acid also has another variable atom or group of atoms bonded to the central carbon atom known as the R group. The R group is the only difference in structure between the 20 amino acids; otherwise, the amino acids are identical.

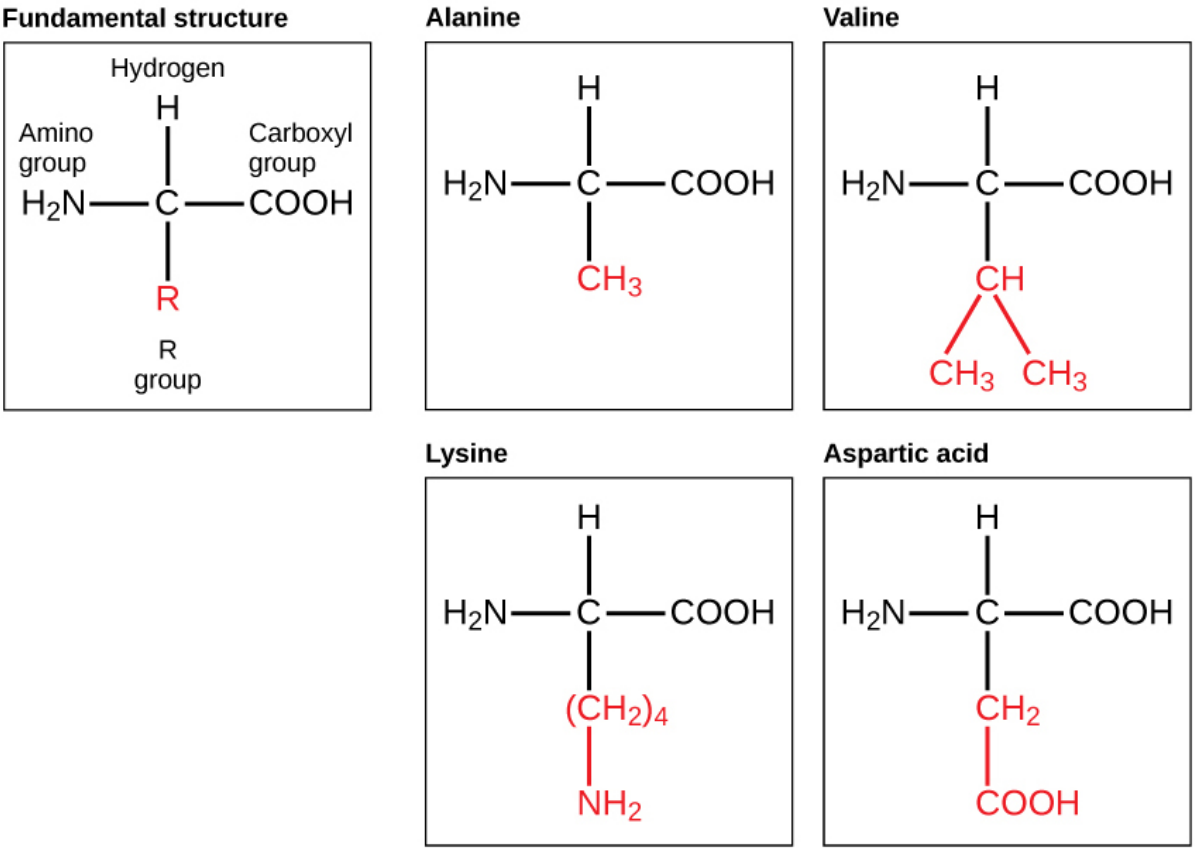

Figure 1. Amino acids are made up of a central carbon bonded to an amino group ($–NH_2$), a carboxyl group (–COOH), and a hydrogen atom. The central carbon's fourth bond varies among the different amino acids, as seen in these examples of alanine, valine, lysine, and aspartic acid.

The chemical nature of the R group determines the chemical nature of the amino acid within its protein (that is, whether it is acidic, basic, polar, or nonpolar).

The sequence and number of amino acids ultimately determine a protein's shape, size, and function. Each amino acid is attached to another amino acid by a covalent bond, known as a peptide bond, which is formed by a dehydration reaction. The carboxyl group of one amino acid and the amino group of a second amino acid combine, releasing a water molecule. The resulting bond is the peptide bond.

The products formed by such a linkage are called polypeptides. While the terms polypeptide and protein are sometimes used interchangeably, a **polypeptide** is technically a polymer of amino acids, whereas the term protein is used for a polypeptide or polypeptides that have combined together, have a distinct shape, and have a unique function.

The Evolutionary Significance of Cytochrome c

Cytochrome c is an important component of the electron transport chain, a part of cellular respiration, and it is normally found in the cellular organelle, the mitochondrion. This protein has a heme prosthetic group, and the central ion of the heme gets alternately reduced and oxidized during electron transfer. Because this essential protein's role in producing cellular energy is crucial, it has changed very little over millions of years. Protein sequencing has shown that there is a considerable amount of cytochrome c amino acid sequence homology among different species; in other words, evolutionary kinship can be assessed by measuring the similarities or differences among various species' DNA or protein sequences.

Scientists have determined that human cytochrome c contains 104 amino acids. For each cytochrome c molecule from different organisms that has been sequenced to date, 37 of these amino acids appear in the same position in all samples of cytochrome c. This indicates that there may have been a common ancestor. On comparing the human and chimpanzee protein sequences, no sequence difference was found. When human and rhesus monkey sequences were compared, the single difference found was in one amino acid. In another comparison, human to yeast sequencing shows a difference in the 44th position.

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PROTEIN STRUCTURE

Learning Outcomes

Define the different layers of protein structure

As discussed earlier, the shape of a protein is critical to its function. To understand how the protein gets its final shape or conformation, we need to understand the four levels of protein structure: primary, secondary, tertiary, and quaternary (Figure 2).

The unique sequence and number of amino acids in a polypeptide chain is its primary structure. The unique sequence for every protein is ultimately determined by the gene that encodes the protein. Any change in the gene sequence may lead to a different amino acid being added to the polypeptide chain, causing a change in protein

structure and function. In sickle cell anemia, the hemoglobin β chain has a single amino acid substitution, causing a change in both the structure and function of the protein. What is most remarkable to consider is that a hemoglobin molecule is made up of two alpha chains and two beta chains that each consist of about 150 amino acids. The molecule, therefore, has about 600 amino acids. The structural difference between a normal hemoglobin molecule and a sickle cell molecule—that dramatically decreases life expectancy—is a single amino acid of the 600.

Because of this change of one amino acid in the chain, the normally biconcave, or disc-shaped, red blood cells assume a crescent or "sickle" shape, which clogs arteries. This can lead to a myriad of serious health problems, such as breathlessness, dizziness, headaches, and abdominal pain for those who have this disease.

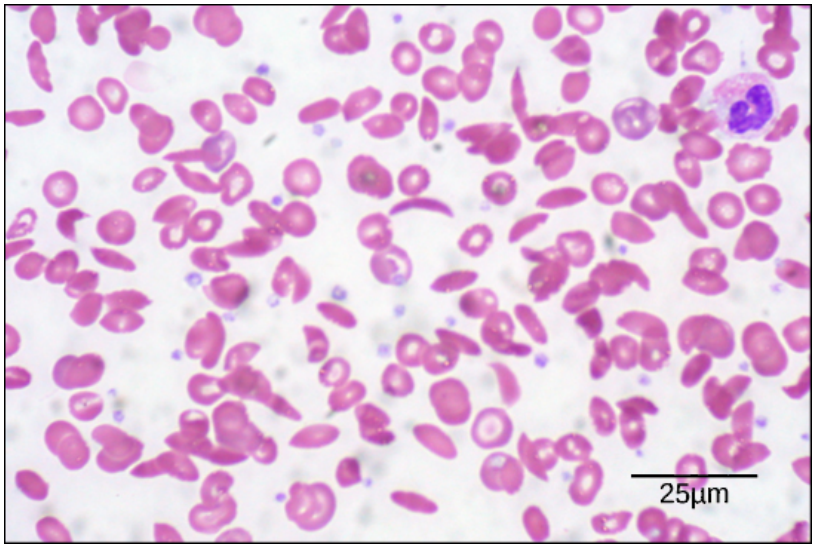

Figure 1. In this blood smear, visualized at 535x magnification using bright field microscopy, sickle cells are crescent shaped, while normal cells are disc-shaped. (credit: modification of work by Ed Uthman; scale-bar data from Matt Russell)

Folding patterns resulting from interactions between the non-R group portions of amino acids give rise to the **secondary structure** of the protein. The most common are the alpha (α)-helix and beta (β)-pleated sheet structures. Both structures are held in shape by hydrogen bonds. In the alpha helix, the bonds form between every fourth amino acid and cause a twist in the amino acid chain.

In the β-pleated sheet, the "pleats" are formed by hydrogen bonding between atoms on the backbone of the polypeptide chain. The R groups are attached to the carbons, and extend above and below the folds of the pleat. The pleated segments align parallel to each other, and hydrogen bonds form between the same pairs of atoms on each of the aligned amino acids. The α-helix and β-pleated sheet structures are found in many globular and fibrous proteins.

The unique three-dimensional structure of a polypeptide is known as its **tertiary structure.** This structure is caused by chemical interactions between various amino acids and regions of the polypeptide. Primarily, the interactions among R groups create the complex three-dimensional tertiary structure of a protein. There may be ionic bonds formed between R groups on different amino acids, or hydrogen bonding beyond that involved in the secondary structure. When protein folding takes place, the hydrophobic R groups of nonpolar amino acids lay in the interior of the protein, whereas the hydrophilic R groups lay on the outside. The former types of interactions are also known as hydrophobic interactions.

In nature, some proteins are formed from several polypeptides, also known as subunits, and the interaction of these subunits forms the **quaternary structure**. Weak interactions between the subunits help to stabilize the overall structure. For example, hemoglobin is a combination of four polypeptide subunits.

Each protein has its own unique sequence and shape held together by chemical interactions. If the protein is subject to changes in temperature, pH, or exposure to chemicals, the protein structure may change, losing its shape in what is known as **denaturation** as discussed earlier. Denaturation is often reversible because the primary structure is preserved if the denaturing agent is removed, allowing the protein to resume its function. Sometimes denaturation is irreversible, leading to a loss of function. One example of protein denaturation can be seen when an egg is fried or boiled. The albumin protein in the liquid egg white is denatured when placed in a hot pan, changing from a clear substance to an opaque white substance. Not all proteins are denatured at high temperatures; for instance, bacteria that survive in hot springs have proteins that are adapted to function at those temperatures.

The four levels of protein structure (primary, secondary, tertiary, and quaternary) are illustrated in Figure 2.

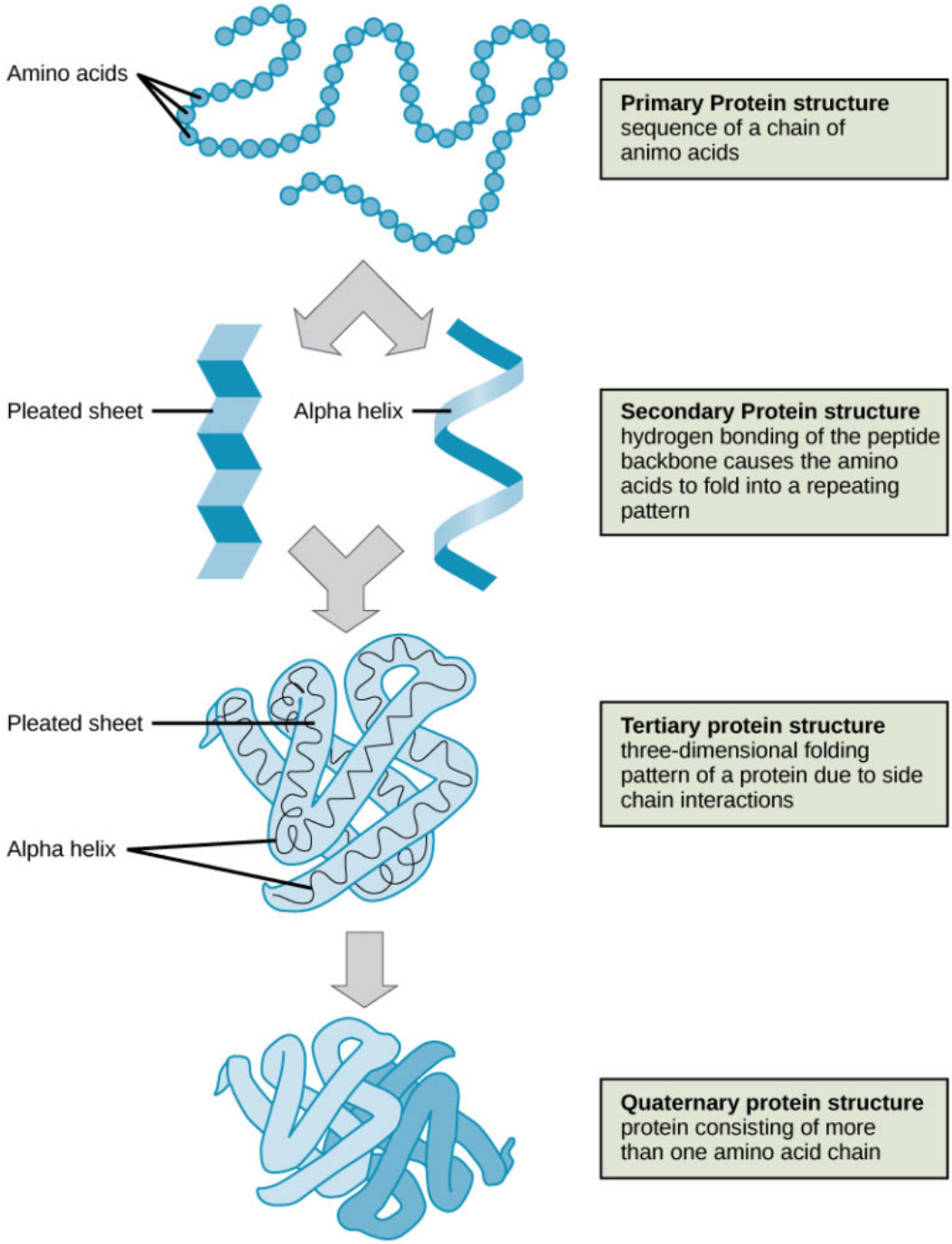

Figure 2. The four levels of protein structure can be observed in these illustrations. (credit: modification of work by National Human Genome Research Institute)

For an additional perspective on proteins, view this animation called "Biomolecules: The Proteins."

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FUNCTION OF PROTEINS

Learning Outcomes

Identify several major functions of proteins

The primary types and functions of proteins are listed in Table 1.

Table 1. Protein Types and Functions

Type	Examples	Functions
Digestive Enzymes	Amylase, lipase, pepsin, trypsin	Help in digestion of food by catabolizing nutrients into m
Transport	Hemoglobin, albumin	Carry substances in the blood or lymph throughout the b
Structural	Actin, tubulin, keratin	Construct different structures, like the cytoskeleton
Hormones	Insulin, thyroxine	Coordinate the activity of different body systems
Defense	Immunoglobulins	Protect the body from foreign pathogens
Contractile	Actin, myosin	Effect muscle contraction
Storage	Legume storage proteins, egg white (albumin)	Provide nourishment in early development of the embryo seedling

Two special and common types of proteins are enzymes and hormones. **Enzymes**, which are produced by living cells, are catalysts in biochemical reactions (like digestion) and are usually complex or conjugated proteins. Each enzyme is specific for the substrate (a reactant that binds to an enzyme) it acts on. The enzyme may help in breakdown, rearrangement, or synthesis reactions. Enzymes that break down their substrates are called catabolic enzymes, enzymes that build more complex molecules from their substrates are called anabolic enzymes, and enzymes that affect the rate of reaction are called catalytic enzymes. It should be noted that all enzymes increase the rate of reaction and, therefore, are considered to be organic catalysts. An example of an enzyme is salivary amylase, which hydrolyzes its substrate amylose, a component of starch.

Hormones are chemical-signaling molecules, usually small proteins or steroids, secreted by endocrine cells that act to control or regulate specific physiological processes, including growth, development, metabolism, and reproduction. For example, insulin is a protein hormone that helps to regulate the blood glucose level.

Proteins have different shapes and molecular weights; some proteins are globular in shape whereas others are fibrous in nature. For example, hemoglobin is a globular protein, but collagen, found in our skin, is a fibrous protein. Protein shape is critical to its function, and this shape is maintained by many different types of chemical bonds. Changes in temperature, pH, and exposure to chemicals may lead to permanent changes in the shape of the protein, leading to loss of function, known as denaturation. All proteins are made up of different arrangements of the same 20 types of amino acids.

In Summary: Function of Proteins

Proteins are a class of macromolecules that perform a diverse range of functions for the cell. They help in metabolism by providing structural support and by acting as enzymes, carriers, or hormones. The building blocks of proteins (monomers) are amino acids. Each amino acid has a central carbon that is linked to an amino group, a carboxyl group, a hydrogen atom, and an R group or side chain. There are 20 commonly occurring amino acids, each of which differs in the R group. Each amino acid is linked to its neighbors by a peptide bond. A long chain of amino acids is known as a polypeptide.

Proteins are organized at four levels: primary, secondary, tertiary, and (optional) quaternary. The primary structure is the unique sequence of amino acids. The local folding of the polypeptide to form structures such as the α helix and β-pleated sheet constitutes the secondary structure. The overall three-dimensional structure is the tertiary structure. When two or more polypeptides combine to form the complete protein structure, the configuration is known as the quaternary structure of a protein. Protein shape and function are intricately linked; any change in shape caused by changes in temperature or pH may lead to protein denaturation and a loss in function.

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INTRODUCTION TO NUCLEIC ACIDS

What you'll learn to do: Discuss nucleic acids and the role they play in DNA and RNA

Humans have two types of nucleic acids in their bodies: DNA and RNA. These molecules contain the set of instructions for our cells: they determine who and what we are. But what makes up our DNA?

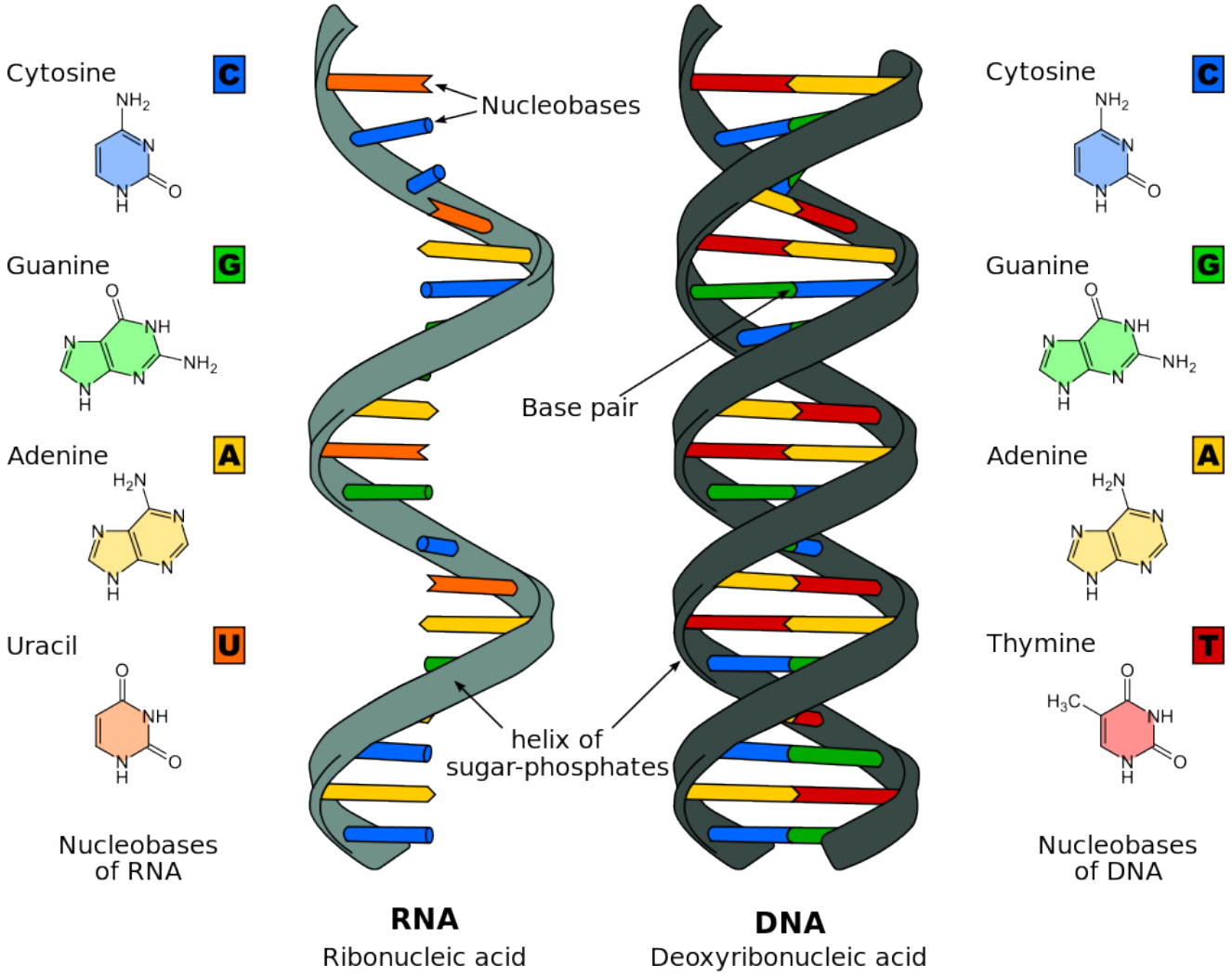

Figure 1. Spot the differences between DNA and RNA

In this outcome, we'll learn about the components of DNA and RNA and get a brief introduction to how they work.

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STRUCTURE OF NUCLEIC ACIDS

Learning Outcomes

Describe the basic structure of nucleic acids

Nucleic acids are key macromolecules in the continuity of life. They carry the genetic blueprint of a cell and carry instructions for the functioning of the cell.

The two main types of nucleic acids are deoxyribonucleic acid (DNA) and ribonucleic acid (RNA). DNA is the genetic material found in all living organisms, ranging from single-celled bacteria to multicellular mammals.

The other type of nucleic acid, RNA, is mostly involved in protein synthesis. The DNA molecules never leave the nucleus, but instead use an RNA intermediary to communicate with the rest of the cell. Other types of RNA are also involved in protein synthesis and its regulation.

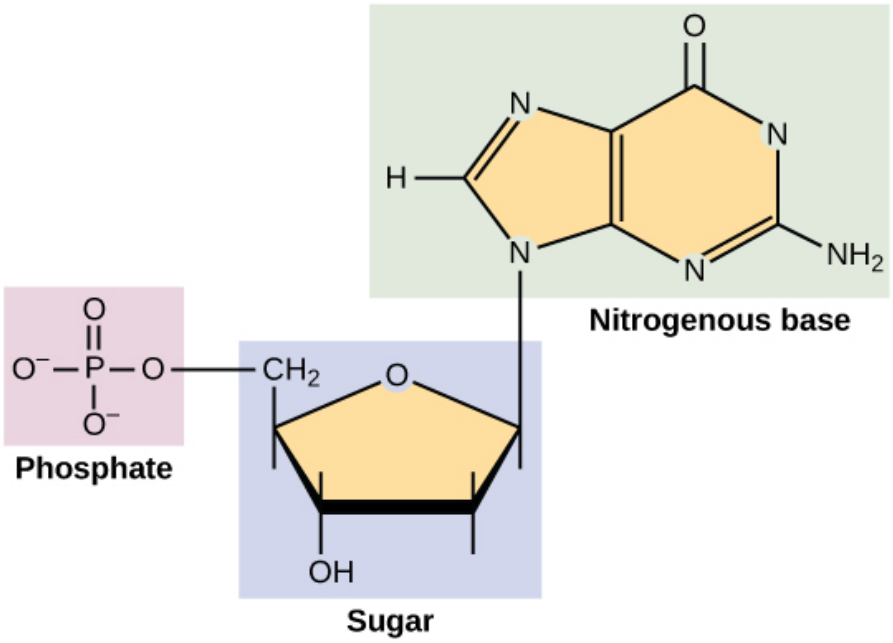

Figure 1. A nucleotide is made up of three components: a nitrogenous base, a pentose sugar, and a phosphate group.

DNA and RNA are made up of monomers known as nucleotides. The nucleotides combine with each other to form a polynucleotide, DNA or RNA. Each nucleotide is made up of three components: a nitrogenous base, a pentose (five-carbon) sugar, and a phosphate group (Figure 1). Each nitrogenous base in a nucleotide is attached to a sugar molecule, which is attached to a phosphate group. The nucleotides link together by phosphodiester bonds to form the polynucleotide.

DNA Double-Helical Structure

DNA has a double-helical structure (Figure 2). It is composed of two strands, or polymers, of nucleotides. The strands are formed with covalent bonds between phosphate and sugar groups of adjacent nucleotides.

The two strands are bonded to each other at their bases with hydrogen bonds, and the strands coil about each other along their length, hence the “double helix” description, which means a double spiral.

The alternating sugar and phosphate groups lie on the outside of each strand, forming the backbone of the DNA. The nitrogenous bases are stacked in the interior, like the steps of a staircase, and these bases pair; the pairs are bound to each other by hydrogen bonds. The bases pair in such a way that the distance between the backbones of the two strands is the same all along the molecule.

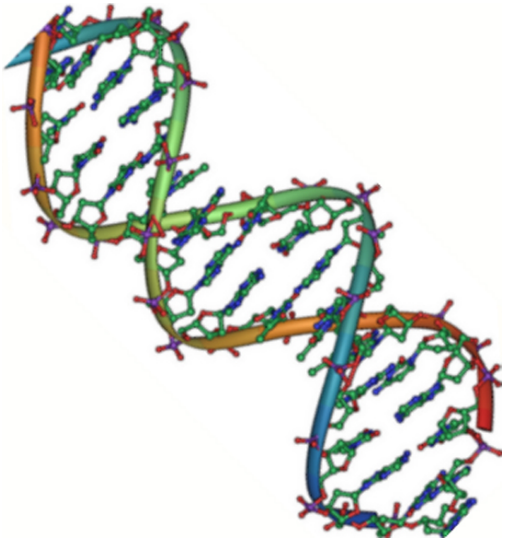

Figure 2. The double-helix model shows DNA as two parallel strands of intertwining molecules. (credit: Jerome Walker, Dennis Myts)

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DNA AND RNA

Learning Outcomes

Compare and contrast the structure of DNA and RNA

While DNA and RNA are similar, they have very distinct differences. Table 1 summarizes features of DNA and RNA.

Table 1. Features of DNA and RNA

	DNA	RNA
Function	Carries genetic information	Involved in protein synthesis
Location	Remains in the nucleus	Leaves the nucleus
Structure	DNA is double-stranded “ladder”: sugar-phosphate backbone, with base rungs.	Usually single-stranded
Sugar	Deoxyribose	Ribose
Pyrimidines	Cytosine, thymine	Cytosine, uracil
Purines	Adenine, guanine	Adenine, guanine

One other difference bears mention. There is only one type of DNA. DNA is the heritable information that is passed along to each generation of cells; its strands can be “unzipped” with small amount of energy when DNA needs to replicate, and DNA is transcribed into RNA. There are mutliple types of RNA: Messenger RNA is a temporary molecule that transports the information necessary to make a protein from the nucleus (where the DNA remains) to the cytoplasm, where the ribosomes are. Other kinds of RNA include ribosomal RNA (rRNA), transfer RNA (tRNA), small nuclear RNA (snRNA), and microRNA.

Even though the RNA is single stranded, most RNA types show extensive intramolecular base pairing between complementary sequences, creating a predictable three-dimensional structure essential for their function.

As you will learn later, information flow in an organism takes place from DNA to RNA to protein. DNA dictates the structure of mRNA in a process known as transcription, and RNA dictates the structure of protein in a process known as translation. This is known as the Central Dogma of Life, which holds true for all organisms; however, exceptions to the rule occur in connection with viral infections.

In Summary: DNA and RNA

Nucleic acids are molecules made up of nucleotides that direct cellular activities such as cell division and protein synthesis. Each nucleotide is made up of a pentose sugar, a nitrogenous base, and a phosphate group. There are two types of nucleic acids: DNA and RNA. DNA carries the genetic blueprint of the cell and is passed on from parents to offspring (in the form of chromosomes). It has a double-helical structure with the two strands running in opposite directions, connected by hydrogen bonds, and complementary to each other. RNA is single-stranded and is made of a pentose sugar (ribose), a nitrogenous base, and a phosphate group. RNA is involved in protein synthesis and its regulation. Messenger RNA (mRNA) is copied from the DNA, is exported from the nucleus to the cytoplasm, and contains information for the construction of proteins. Ribosomal RNA (rRNA) is a part of the ribosomes at the site of protein synthesis, whereas transfer RNA (tRNA) carries the amino acid to the site of protein synthesis. microRNA regulates the use of mRNA for protein synthesis.

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INTRODUCTION TO COMPARING BIOLOGICAL MACROMOLECULES

What you'll learn to do: Discuss macromolecules and the differences between the four classes

As we've learned, there are four major classes of biological macromolecules:

- Proteins (polymers of amino acids)
- Carbohydrates (polymers of sugars)
- Lipids (polymers of lipid monomers)
- Nucleic acids (DNA and RNA; polymers of nucleotides)

Let's take a closer look at the differences between the difference classes.

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DIFFERENT TYPES OF BIOLOGICAL MACROMOLECULES

Learning Outcomes

- Distinguish between the four classes of macromolecules

Carbohydrates are a group of macromolecules that are a vital energy source for the cell, provide structural support to many organisms, and can be found on the surface of the cell as receptors or for cell recognition. Carbohydrates are classified as monosaccharides, disaccharides, and polysaccharides, depending on the number of monomers in the molecule.

Lipids are a class of macromolecules that are nonpolar and hydrophobic in nature. Major types include fats and oils, waxes, phospholipids, and steroids. Fats and oils are a stored form of energy and can include triglycerides. Fats and oils are usually made up of fatty acids and glycerol.

Proteins are a class of macromolecules that can perform a diverse range of functions for the cell. They help in metabolism by providing structural support and by acting as enzymes, carriers or as hormones. The building blocks of proteins are amino acids. Proteins are organized at four levels: primary, secondary, tertiary, and quaternary. Protein shape and function are intricately linked; any change in shape caused by changes in temperature, pH, or chemical exposure may lead to protein denaturation and a loss of function.

Nucleic acids are molecules made up of repeating units of nucleotides that direct cellular activities such as cell division and protein synthesis. Each nucleotide is made up of a pentose sugar, a nitrogenous base, and a phosphate group. There are two types of nucleic acids: DNA and RNA.

Practice Questions

Explain at least three functions that lipids serve in plants and/or animals.

Answer

Fat serves as a valuable way for animals to store energy. It can also provide insulation. Phospholipids and steroids are important components of cell membranes.

Explain what happens if even one amino acid is substituted for another in a polypeptide chain. Provide a specific example.

Answer

A change in gene sequence can lead to a different amino acid being added to a polypeptide chain instead of the normal one. This causes a change in protein structure and function. For example, in sickle cell anemia, the hemoglobin β chain has a single amino acid substitution. Because of this change, the disc-shaped red blood cells assume a crescent shape, which can result in serious health problems.

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PUTTING IT TOGETHER: IMPORTANT BIOLOGICAL MACROMOLECULES

Watch This Video

Watch this video online: https://youtu.be/H8WJ2KENlK0

Now that we've learned about the different macromolecules our bodies need and use, let's return to our the questions we asked at the beginning of the chapter about healthy diets:

Think about It

- Is it even possible for a person to cut all carbs out of his or her diet?
- Is it actually healthy to remove an entire class of molecules from the diet?
- Fats and cholesterol are strictly bad—right?

See Our Thoughts

It is, in fact, impossible to have a no carb or no fat diet. These molecules are in all cells and cells makes up what we eat. More importantly though, each of these biological macromolecules has a very important role to play. If you cut too much fat from your diet, for example, it is possible for your fat stores dip low enough that your hair will fall out!

Even the much maligned cholesterol is a requirement for a healthy body and lifestyle: without sufficient cholesterol, your body doesn't make enough sex hormones (estrogen or testosterone depending on if you're male or female). The trick is to make healthy choices overall without too much of any biological macromolecules—after all, there can certainly be too much of a good thing.

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MODULE 4: CELLULAR STRUCTURE

WHY IT MATTERS: CELLULAR STRUCTURE

Why is it important to learn about cells?

The cell is the basic structural, functional, and biological unit of all known living organisms: all living things are made up of cells (or a single cell, in some cases). However, it's important to remember that cells are simply the smallest unit that can act by themselves. Cells are composed of many different subunits that work in harmony. Each of these different units—membranes, organelles, filaments, etc.—performs a unique function that facilitates life.

Inherited Diseases

When a specific organelle performs incorrectly, it can result in various diseases. For example, the following diseases are linked directly to specific cellular components:

- **Pompe Disease:** characterized by excess accumulation of glycogen in muscle cells
- **Leigh Disease:** progressive disorder of lesions (dead or dying cells) in the brain
- **Emery-Dreifuss muscular dystrophy:** wasting and weakness in muscles of the shoulders, upper arms, and calf muscles

What cellular components could each of these be connected to? Understanding how cellular components work can help you understand health issues related to cellular function.

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INTRODUCTION TO CELL THEORY

What you'll learn to do: State the basic principles of the unified cell theory

A cell is the smallest unit of a living thing. A living thing, whether made of one cell (like bacteria) or many cells (like a human), is called an organism. Thus, cells are the basic building blocks of all organisms. In this outcome we will learn about the discovery and origin of cells.

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CELL THEORY

Learning Outcomes

State the basic principles of the unified cell theory

A cell is the smallest unit of life. Most cells are so tiny that they cannot be seen with the naked eye. Therefore, scientists use microscopes to study cells. The first microscopes were used in the 1600s by Antony van Leeuwenhoek, a Dutch shopkeeper who had great skill in crafting lenses. Despite the limitations of his now-ancient lenses, van Leeuwenhoek observed the movements of protista (a type of single-celled organism) and sperm, which he collectively termed “animalcules.”

In a 1665 publication called *Micrographia*, experimental scientist Robert Hooke coined the term “cell” for the box-like structures he observed when viewing cork tissue through a lens. In the 1670s, van Leeuwenhoek discovered bacteria and protozoa. Later advances in lenses, microscope construction, and staining techniques enabled other scientists to see some components inside cells.

By the late 1830s, botanist Matthias Schleiden and zoologist Theodor Schwann were studying tissues and proposed the **unified cell theory**, which states that all living things are composed of one or more cells, the cell is the basic unit of life, and new cells arise from existing cells. Rudolf Virchow later made important contributions to this theory.

Microscopes

The microscopes we use today are far more complex than those used in the 1600s and 1800s. There are two primary types of modern microscopes used: light microscopes and electron microscopes. Electron microscopes provide higher magnification, higher resolution, and more detail than light microscopes. However, a light microscope is required to study living cells as the method used to prepare the specimen for viewing with an electron microscope kills the specimen.

Cytotechnologist

Have you ever heard of a medical test called a Pap smear (shown in Figure 1)? In this test, a doctor takes a small sample of cells from the uterine cervix of a patient and sends it to a medical lab where a cytotechnologist stains the cells and examines them for any changes that could indicate cervical cancer or a microbial infection.

Cytotechnologists (*cyto* = “cell”) are professionals who study cells via microscopic examinations and other laboratory tests. They are trained to determine which cellular changes are within normal limits and which are abnormal. Their focus is not limited to cervical cells; they study cellular specimens that come from all organs. When they notice abnormalities, they consult a pathologist, who is a medical doctor who can make a clinical diagnosis.

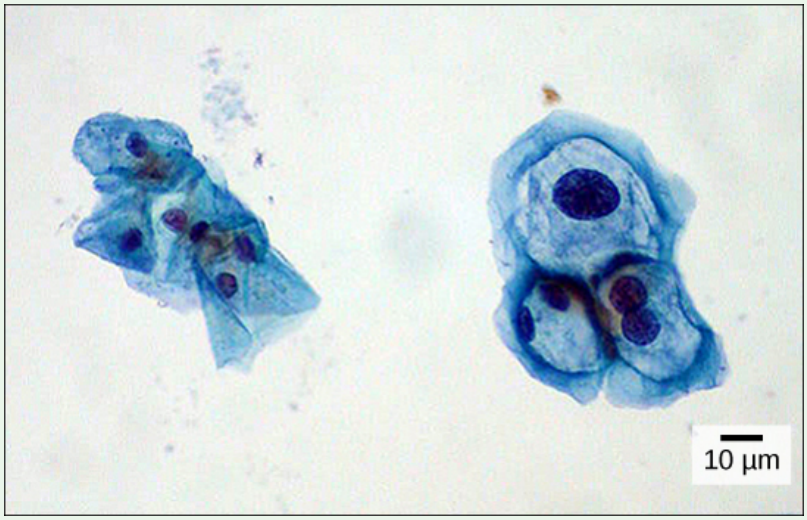

Figure 1. These uterine cervix cells, viewed through a light microscope, were obtained from a Pap smear. Normal cells are on the left. The cells on the right are infected with human papillomavirus (HPV). Notice that the infected cells are larger; also, two of these cells each have two nuclei instead of one, the normal number. (credit: modification of work by Ed Uthman, MD; scale-bar data from Matt Russell)

Cytotechnologists play a vital role in saving people's lives. When abnormalities are discovered early, a patient's treatment can begin sooner, which usually increases the chances of a successful outcome.

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INTRODUCTION TO PROKARYOTES AND EUKARYOTES

What you'll learn to do: Compare prokaryotes and eukaryotes

In Introduction to Biology, we discussed the diversity of life on earth and mentioned how there are over 1.9 million species of living organisms on earth today. All these species of organisms have one of two different types of cells. In this section, we will compare the two cell types: prokaryotic and eukaryotic.

Within these two broad categories of cells, there are many diverse life forms. For example, both animal and plant cells are classified as eukaryotic cells, whereas all the many bacterial cells are classified as prokaryotic.

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COMPARING PROKARYOTIC AND EUKARYOTIC CELLS

Learning Outcomes

- Identify features common to all cells
- Compare and contrast prokaryotic cells and eukaryotic cells

Cells fall into one of two broad categories: prokaryotic and eukaryotic. The predominantly single-celled organisms of the domains Bacteria and Archaea are classified as prokaryotes (*pro–* = before; *–karyon–* = nucleus). Animal cells, plant cells, fungi, and protists are eukaryotes (*eu–* = true).

Components of Prokaryotic Cells

All cells share four common components: 1) a plasma membrane, an outer covering that separates the cell's interior from its surrounding environment; 2) cytoplasm, consisting of a jelly-like region within the cell in which other cellular components are found; 3) DNA, the genetic material of the cell; and 4) ribosomes, particles that synthesize proteins. However, prokaryotes differ from eukaryotic cells in several ways.

A prokaryotic cell is a simple, single-celled (unicellular) organism that lacks a nucleus, or any other membrane-bound organelle. We will shortly come to see that this is significantly different in eukaryotes. Prokaryotic DNA is found in the central part of the cell: a darkened region called the nucleoid (Figure 1).

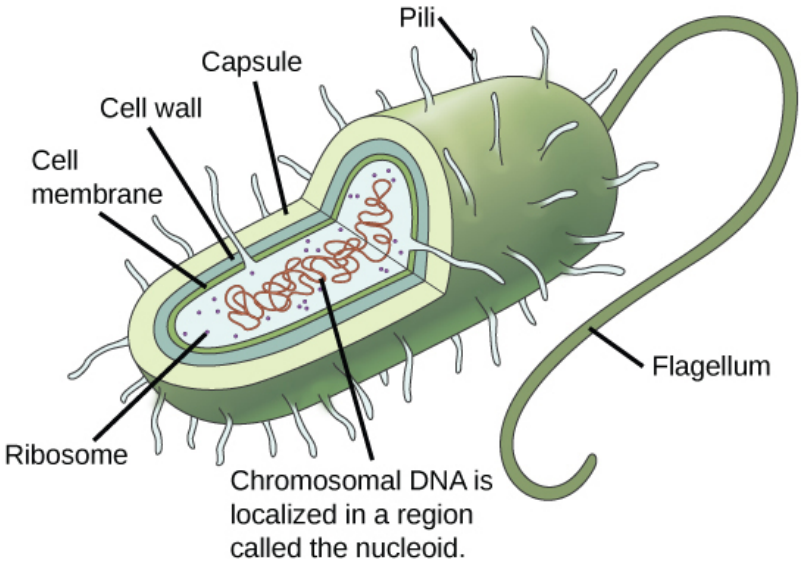

Figure 1. This figure shows the generalized structure of a prokaryotic cell.

Unlike Archaea and eukaryotes, bacteria have a cell wall made of peptidoglycan, comprised of sugars and amino acids, and many have a polysaccharide capsule (Figure 1). The cell wall acts as an extra layer of protection, helps the cell maintain its shape, and prevents dehydration. The capsule enables the cell to attach to surfaces in its environment. Some prokaryotes have flagella, pili, or fimbriae. Flagella are used for locomotion. Pili are used to exchange genetic material during a type of reproduction called conjugation. Fimbriae are protein appendages used by bacteria to attach to other cells.

Eukaryotic Cells

In nature, the relationship between form and function is apparent at all levels, including the level of the cell, and this will become clear as we explore eukaryotic cells. The principle "form follows function" is found in many contexts. For example, birds and fish have streamlined bodies that allow them to move quickly through the

medium in which they live, be it air or water. It means that, in general, one can deduce the function of a structure by looking at its form, because the two are matched.

A eukaryotic cell is a cell that has a membrane-bound nucleus and other membrane-bound compartments or sacs, called organelles, which have specialized functions. The word eukaryotic means “true kernel” or “true nucleus,” alluding to the presence of the membrane-bound nucleus in these cells. The word “organelle” means “little organ,” and, as already mentioned, organelles have specialized cellular functions, just as the organs of your body have specialized functions.

Watch this video to see the functionality of plant and animal cells.

Cell Size

At 0.1–5.0 µm in diameter, prokaryotic cells are significantly smaller than eukaryotic cells, which have diameters ranging from 10–100 µm (Figure 2). The small size of prokaryotes allows ions and organic molecules that enter them to quickly spread to other parts of the cell. Similarly, any wastes produced within a prokaryotic cell can quickly move out. However, larger eukaryotic cells have evolved different structural adaptations to enhance cellular transport. Indeed, the large size of these cells would not be possible without these adaptations. In general, cell size is limited because volume increases much more quickly than does cell surface area. As a cell becomes larger, it becomes more and more difficult for the cell to acquire sufficient materials to support the processes inside the cell, because the relative size of the surface area through which materials must be transported declines.

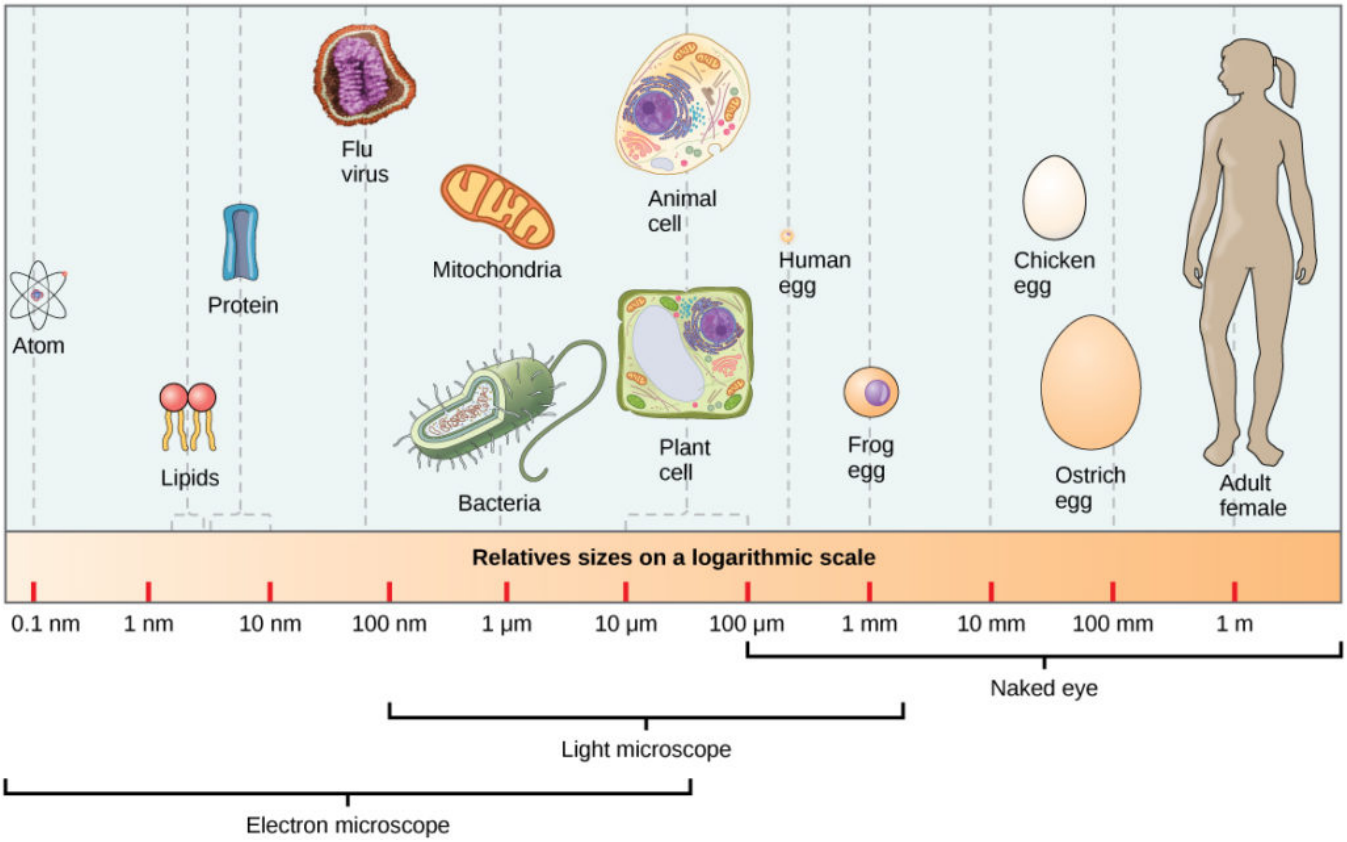

Figure 2. This figure shows the relative sizes of different kinds of cells and cellular components. An adult human is shown for comparison.

In Summary: Comparing Prokaryotic and Eukaryotic Cells

Prokaryotes are predominantly single-celled organisms of the domains Bacteria and Archaea. All prokaryotes have plasma membranes, cytoplasm, ribosomes, a cell wall, DNA, and lack membrane-bound organelles. Many also have polysaccharide capsules. Prokaryotic cells range in diameter from 0.1–5.0 µm.

Like a prokaryotic cell, a eukaryotic cell has a plasma membrane, cytoplasm, and ribosomes, but a eukaryotic cell is typically larger than a prokaryotic cell, has a true nucleus (meaning its DNA is surrounded by a membrane), and has other membrane-bound organelles that allow for compartmentalization of functions. Eukaryotic cells tend to be 10 to 100 times the size of prokaryotic cells.

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EUKARYOTIC ORIGINS

Learning Outcomes

- List the unifying characteristics of eukaryotes
- Discuss the origins of eukaryotic life

Living things fall into three large groups: Archaea, Bacteria, and Eukarya. The first two have prokaryotic cells, and the third contains all eukaryotes. A relatively sparse fossil record is available to help discern what the first members of each of these lineages looked like, so it is possible that all the events that led to the last common ancestor of extant eukaryotes will remain unknown. However, comparative biology of extant organisms and the limited fossil record provide some insight into the history of Eukarya.

The earliest fossils found appear to be Bacteria, most likely cyanobacteria. They are about 3.5 billion years old and are recognizable because of their relatively complex structure and, for prokaryotes, relatively large cells. Most other prokaryotes have small cells, 1 or 2 µm in size, and would be difficult to pick out as fossils. Most living eukaryotes have cells measuring 10 µm or greater. Structures this size, which might be fossils, appear in the geological record about 2.1 billion years ago.

Characteristics of Eukaryotes

Data from these fossils have led comparative biologists to the conclusion that living eukaryotes are all descendants of a single common ancestor. Mapping the characteristics found in all major groups of eukaryotes reveals that the following characteristics must have been present in the last common ancestor, because these characteristics are present in at least some of the members of each major lineage.

1. Cells with nuclei surrounded by a nuclear envelope with nuclear pores. This is the single characteristic that is both necessary and sufficient to define an organism as a eukaryote. All extant eukaryotes have cells with nuclei.
2. Mitochondria. Some extant eukaryotes have very reduced remnants of mitochondria in their cells, whereas other members of their lineages have “typical” mitochondria.

3. A cytoskeleton containing the structural and motility components called actin microfilaments and microtubules. All extant eukaryotes have these cytoskeletal elements.
4. Flagella and cilia, organelles associated with cell motility. Some extant eukaryotes lack flagella and/or cilia, but they are descended from ancestors that possessed them.
5. Chromosomes, each consisting of a linear DNA molecule coiled around basic (alkaline) proteins called histones. The few eukaryotes with chromosomes lacking histones clearly evolved from ancestors that had them.
6. Mitosis, a process of nuclear division wherein replicated chromosomes are divided and separated using elements of the cytoskeleton. Mitosis is universally present in eukaryotes.
7. Sex, a process of genetic recombination unique to eukaryotes in which diploid nuclei at one stage of the life cycle undergo meiosis to yield haploid nuclei and subsequent karyogamy, a stage where two haploid nuclei fuse together to create a diploid zygote nucleus.
8. Members of all major lineages have cell walls, and it might be reasonable to conclude that the last common ancestor could make cell walls during some stage of its life cycle. However, not enough is known about eukaryotes' cell walls and their development to know how much homology exists among them. If the last common ancestor could make cell walls, it is clear that this ability must have been lost in many groups.

Endosymbiosis and the Evolution of Eukaryotes

In order to understand eukaryotic organisms fully, it is necessary to understand that all living eukaryotes are descendants of a chimeric organism that was a composite of a host cell and the cell(s) of an alpha-proteobacterium that "took up residence" inside it. This major theme in the origin of eukaryotes is known as endosymbiosis, one cell engulfing another such that the engulfed cell survives and both cells benefit. Over many generations, a symbiotic relationship can result in two organisms that depend on each other so completely that neither could survive on its own. Endosymbiotic events likely contributed to the origin of the last common ancestor of today's eukaryotes and to later diversification in certain lineages of eukaryotes.

Endosymbiotic Theory

As cell biology developed in the twentieth century, it became clear that mitochondria were the organelles responsible for producing ATP using aerobic respiration. In the 1960s, American biologist Lynn Margulis developed endosymbiotic theory, which states that eukaryotes may have been a product of one cell engulfing another, one living within another, and evolving over time until the separate cells were no longer recognizable as such. In 1967, Margulis introduced new work on the theory and substantiated her findings through microbiological evidence. Although Margulis' work initially was met with resistance, this once-revolutionary hypothesis is now widely (but not completely) accepted, with work progressing on uncovering the steps involved in this evolutionary process and the key players involved. Much still remains to be discovered about the origins of the cells that now make up the cells in all living eukaryotes.

Broadly, it has become clear that many of our nuclear genes and the molecular machinery responsible for replication and expression appear closely related to those in Archaea. On the other hand, the metabolic organelles and genes responsible for many energy-harvesting processes had their origins in bacteria. Much remains to be clarified about how this relationship occurred; this continues to be an exciting field of discovery in biology. For instance, it is not known whether the endosymbiotic event that led to mitochondria occurred before or after the host cell had a nucleus. Such organisms would be among the extinct precursors of the last common ancestor of eukaryotes.

Mitochondria

One of the major features distinguishing prokaryotes from eukaryotes is the presence of mitochondria. Eukaryotic cells may contain anywhere from one to several thousand mitochondria, depending on the cell's level of energy consumption. Each mitochondrion measures 1 to 10 or greater micrometers in length and exists in the cell as an organelle that can be ovoid to worm-shaped to intricately branched (Figure 1).

Mitochondria arise from the division of existing mitochondria; they may fuse together; and they may be moved around inside the cell by interactions with the cytoskeleton. However, mitochondria cannot survive outside the cell. As the atmosphere was oxygenated by photosynthesis, and as successful aerobic prokaryotes evolved, evidence suggests that an ancestral cell with some membrane compartmentalization engulfed a free-living aerobic prokaryote, specifically an alpha-proteobacterium, thereby giving the host cell the ability to use oxygen to release energy stored in nutrients.

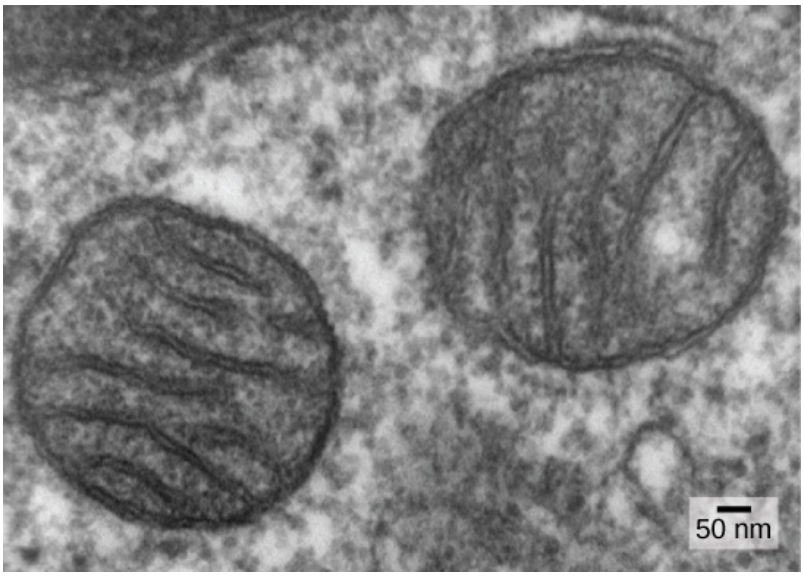

Figure 1. In this transmission electron micrograph of mitochondria in a mammalian lung cell, the cristae, infoldings of the mitochondrial inner membrane, can be seen in cross-section. (credit: Louise Howard)

Mitochondria divide independently by a process that resembles binary fission in prokaryotes. Specifically, mitochondria are not formed from scratch (de novo) by the eukaryotic cell; they reproduce within it and are distributed with the cytoplasm when a cell divides or two cells fuse. Therefore, although these organelles are highly integrated into the eukaryotic cell, they still reproduce as if they are independent organisms within the cell. However, their reproduction is synchronized with the activity and division of the cell. Mitochondria have their own (usually) circular DNA chromosome that is stabilized by attachments to the inner membrane and carries genes similar to genes expressed by alpha-proteobacteria. Mitochondria also have special ribosomes and transfer RNAs that resemble these components in prokaryotes. These features all support that mitochondria were once free-living prokaryotes.

Mitochondria that carry out aerobic respiration have their own genomes, with genes similar to those in alpha-proteobacteria. However, many of the genes for respiratory proteins are located in the nucleus. When these genes are compared to those of other organisms, they appear to be of alpha-proteobacterial origin. Additionally, in some eukaryotic groups, such genes are found in the mitochondria, whereas in other groups, they are found in the nucleus. This has been interpreted as evidence that genes have been transferred from the endosymbiont chromosome to the host genome. This loss of genes by the endosymbiont is probably one explanation why mitochondria cannot live without a host.

Plastids

Some groups of eukaryotes are photosynthetic. Their cells contain, in addition to the standard eukaryotic organelles, another kind of organelle called a plastid. When such cells are carrying out photosynthesis, their plastids are rich in the pigment chlorophyll *a* and a range of other pigments, called accessory pigments, which are involved in harvesting energy from light. Photosynthetic plastids are called chloroplasts (Figure 2).

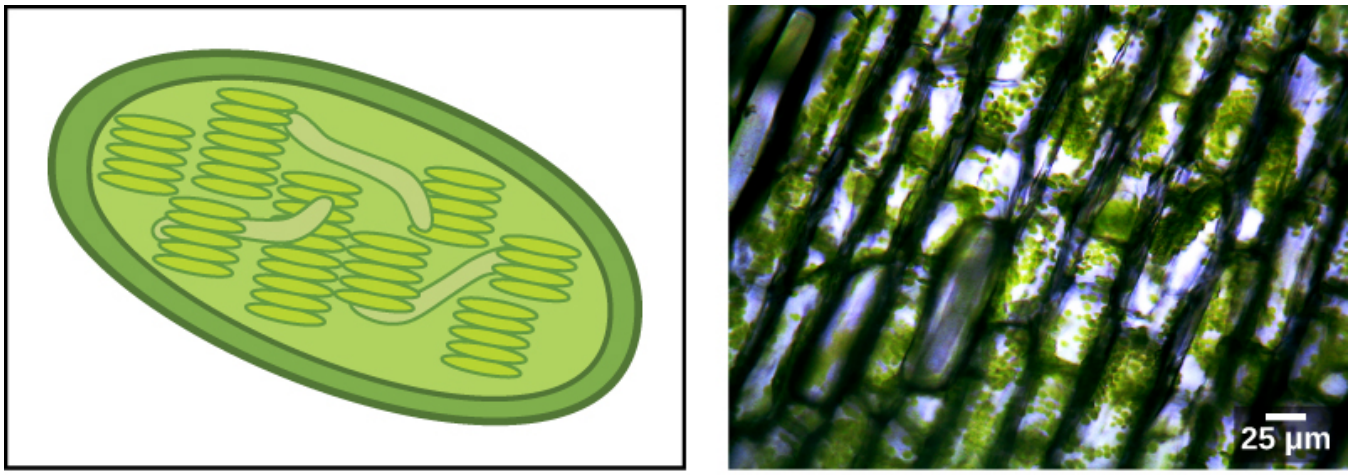

Figure 2. (a) This chloroplast cross-section illustrates its elaborate inner membrane organization. Stacks of thylakoid membranes compartmentalize photosynthetic enzymes and provide scaffolding for chloroplast DNA. (b) In this micrograph of Elodea sp., the chloroplasts can be seen as small green spheres. (credit b: modification of work by Brandon Zierer; scale-bar data from Matt Russell)

Like mitochondria, plastids appear to have an endosymbiotic origin. This hypothesis was also championed by Lynn Margulis. Plastids are derived from cyanobacteria that lived inside the cells of an ancestral, aerobic, heterotrophic eukaryote. This is called primary endosymbiosis, and plastids of primary origin are surrounded by two membranes. The best evidence is that this has happened twice in the history of eukaryotes. In one case, the common ancestor of the major lineage/supergroup Archaeplastida took on a cyanobacterial endosymbiont; in the other, the ancestor of the small amoeboid rhizarian taxon, *Paulinella*, took on a different cyanobacterial endosymbiont. Almost all photosynthetic eukaryotes are descended from the first event, and only a couple of species are derived from the other.

Cyanobacteria are a group of Gram-negative bacteria with all the conventional structures of the group. However, unlike most prokaryotes, they have extensive, internal membrane-bound sacs called thylakoids. Chlorophyll is a component of these membranes, as are many of the proteins of the light reactions of photosynthesis. Cyanobacteria also have the peptidoglycan wall and lipopolysaccharide layer associated with Gram-negative bacteria.

Chloroplasts of primary origin have thylakoids, a circular DNA chromosome, and ribosomes similar to those of cyanobacteria. Each chloroplast is surrounded by two membranes. In the group of Archaeplastida called the glaucophytes and in *Paulinella*, a thin peptidoglycan layer is present between the outer and inner plastid membranes. All other plastids lack this relictual cyanobacterial wall. The outer membrane surrounding the plastid is thought to be derived from the vacuole in the host, and the inner membrane is thought to be derived from the plasma membrane of the symbiont.

There is also, as with the case of mitochondria, strong evidence that many of the genes of the endosymbiont were transferred to the nucleus. Plastids, like mitochondria, cannot live independently outside the host. In addition, like mitochondria, plastids are derived from the division of other plastids and never built from scratch. Researchers have suggested that the endosymbiotic event that led to Archaeplastida occurred 1 to 1.5 billion years ago, at least 5 hundred million years after the fossil record suggests that eukaryotes were present.

Practice Question

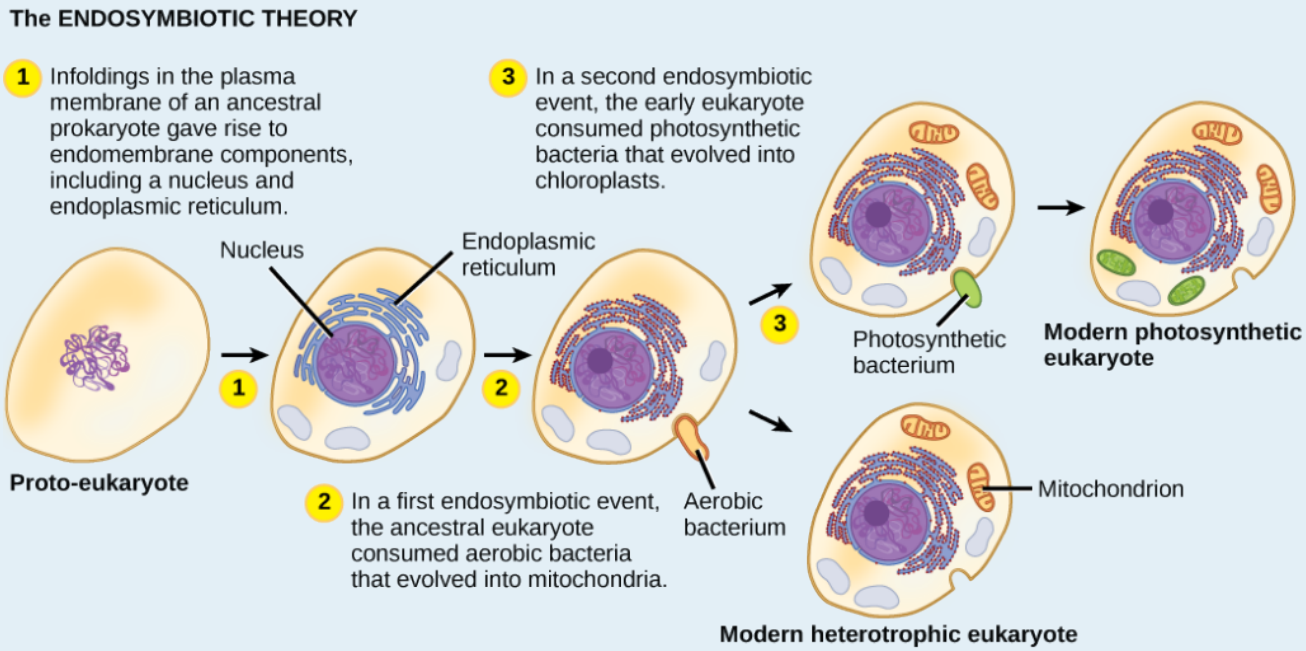

Figure 3. The first eukaryote may have originated from an ancestral prokaryote that had undergone membrane proliferation, compartmentalization of cellular function (into a nucleus, lysosomes, and an endoplasmic reticulum), and the establishment of endosymbiotic relationships with an aerobic prokaryote, and, in some cases, a photosynthetic prokaryote, to form mitochondria and chloroplasts, respectively.

What evidence is there that mitochondria were incorporated into the ancestral eukaryotic cell before chloroplasts?

Answer

All eukaryotic cells have mitochondria, but not all eukaryotic cells have chloroplasts.

Secondary Endosymbiosis in Chlorarachniophytes

Endosymbiosis involves one cell engulfing another to produce, over time, a coevolved relationship in which neither cell could survive alone. The chloroplasts of red and green algae, for instance, are derived from the engulfment of a photosynthetic cyanobacterium by an early prokaryote.

This leads to the question of the possibility of a cell containing an endosymbiont to itself become engulfed, resulting in a secondary endosymbiosis. Molecular and morphological evidence suggest that the chlorarachniophyte protists are derived from a secondary endosymbiotic event. Chlorarachniophytes are rare algae indigenous to tropical seas and sand that can be classified into the rhizarian supergroup. Chlorarachniophytes extend thin cytoplasmic strands, interconnecting themselves with other chlorarachniophytes, in a cytoplasmic network. These protists are thought to have originated when a eukaryote engulfed a green alga, the latter of which had already established an endosymbiotic relationship with a photosynthetic cyanobacterium (Figure 4).

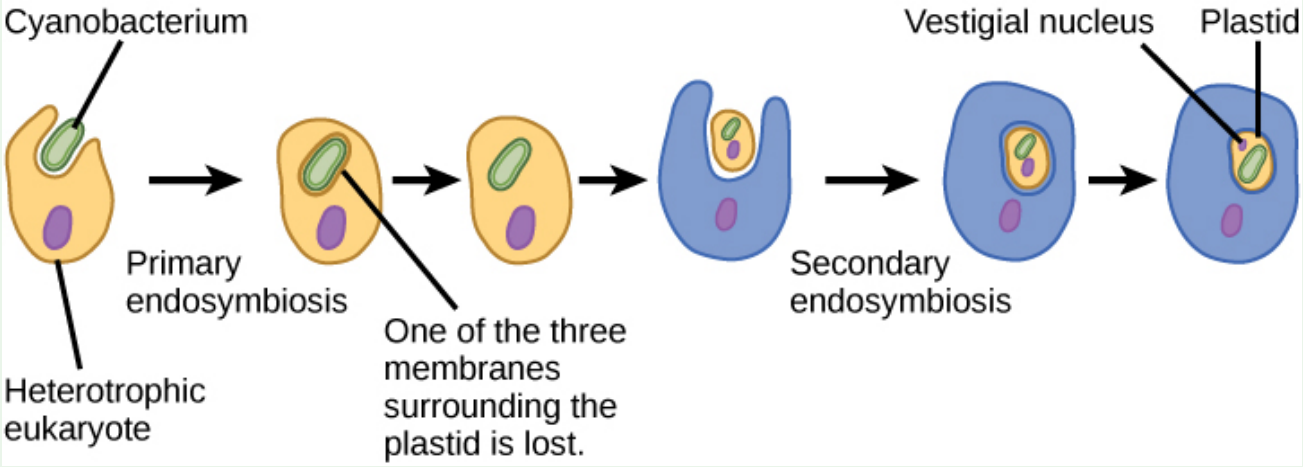

Figure 4. The hypothesized process of endosymbiotic events leading to the evolution of chlorarachniophytes is shown. In a primary endosymbiotic event, a heterotrophic eukaryote consumed a cyanobacterium. In a secondary endosymbiotic event, the cell resulting from primary endosymbiosis was consumed by a second cell. The resulting organelle became a plastid in modern chlorarachniophytes.

Several lines of evidence support that chlorarachniophytes evolved from secondary endosymbiosis. The chloroplasts contained within the green algal endosymbionts still are capable of photosynthesis, making chlorarachniophytes photosynthetic. The green algal endosymbiont also exhibits a stunted vestigial nucleus. In fact, it appears that chlorarachniophytes are the products of an evolutionarily recent secondary endosymbiotic event.

The plastids of chlorarachniophytes are surrounded by four membranes: The first two correspond to the inner and outer membranes of the photosynthetic cyanobacterium, the third corresponds to the green alga, and the fourth corresponds to the vacuole that surrounded the green alga when it was engulfed by the chlorarachniophyte ancestor. In other lineages that involved secondary endosymbiosis, only three membranes can be identified around plastids. This is currently rectified as a sequential loss of a membrane during the course of evolution.

The process of secondary endosymbiosis is not unique to chlorarachniophytes. In fact, secondary endosymbiosis of green algae also led to euglenid protists, whereas secondary endosymbiosis of red algae led to the evolution of dinoflagellates, apicomplexans, and stramenopiles.

In Summary: Eukaryotic Origins

The oldest fossil evidence of eukaryotes is about 2 billion years old. Fossils older than this all appear to be prokaryotes. It is probable that today's eukaryotes are descended from an ancestor that had a prokaryotic organization. The last common ancestor of today's Eukarya had several characteristics, including cells with nuclei that divided mitotically and contained linear chromosomes where the DNA was associated with histones, a cytoskeleton and endomembrane system, and the ability to make cilia/flagella during at least part of its life cycle. It was aerobic because it had mitochondria that were the result of an aerobic alpha-proteobacterium that lived inside a host cell. Whether this host had a nucleus at the time of the initial symbiosis remains unknown. The last common ancestor may have had a cell wall for at least part of its life cycle, but more data are needed to confirm this hypothesis. Today's eukaryotes are very diverse in their shapes, organization, life cycles, and number of cells per individual.

Practice Questions

Refer back to Figure 3. What evidence is there that mitochondria were incorporated into the ancestral eukaryotic cell before chloroplasts?

Answer

All eukaryotic cells have mitochondria, but not all eukaryotic cells have chloroplasts.

Describe the hypothesized steps in the origin of eukaryotic cells.

Answer

Eukaryotic cells arose through endosymbiotic events that gave rise to the energy-producing organelles within the eukaryotic cells such as mitochondria and chloroplasts. The nuclear genome of eukaryotes is related most closely to the Archaea, so it may have been an early archaean that engulfed a bacterial cell that evolved into a mitochondrion. Mitochondria appear to have originated from an alpha-proteobacterium, whereas chloroplasts originated as a cyanobacterium. There is also evidence of secondary endosymbiotic events. Other cell components may also have resulted from endosymbiotic events.

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INTRODUCTION TO ORGANELLES

What you'll learn to do: Identify membrane-bound organelles found in eukaryotic cells

Have you ever heard the phrase "form follows function?" It's a philosophy practiced in many industries. In architecture, this means that buildings should be constructed to support the activities that will be carried out inside them. For example, a skyscraper should be built with several elevator banks; a hospital should be built so that its emergency room is easily accessible.

Our natural world originated the principle of form following function, especially in cell biology, and this will become clear as we explore eukaryotic cells. Unlike prokaryotic cells, **eukaryotic cells** have:

1. a membrane-bound nucleus
2. numerous membrane-bound **organelles**—such as the endoplasmic reticulum, Golgi apparatus, chloroplasts, mitochondria, and others
3. several, rod-shaped chromosomes

Because a eukaryotic cell's nucleus is surrounded by a membrane, it is often said to have a "true nucleus." The word "organelle" means "little organ," and, as already mentioned, organelles have specialized cellular functions, just as the organs of your body have specialized functions.

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CYTOPLASM

Learning Outcomes

Describe the basic composition of cytoplasm

Before we begin looking at individual organelles, we do need to briefly address the matrix in which they sit: the **cytoplasm**. The part of the cell referred to as cytoplasm is slightly different in eukaryotes and prokaryotes. In eukaryotic cells, which have a nucleus, the cytoplasm is everything between the plasma membrane and the nuclear envelope. In prokaryotes, which lack a nucleus, cytoplasm simply means everything found inside the plasma membrane.

One major component of the cytoplasm in both prokaryotes and eukaryotes is the gel-like **cytosol**, a water-based solution that contains ions, small molecules, and macromolecules. In eukaryotes, the cytoplasm also includes membrane-bound organelles, which are suspended in the cytosol. The cytoskeleton, a network of fibers that supports the cell and gives it shape, is also part of the cytoplasm and helps to organize cellular components.

Even though the cytosol is mostly water, it has a semi-solid, Jello-like consistency because of the many proteins suspended in it. The cytosol contains a rich broth of macromolecules and smaller organic molecules, including glucose and other simple sugars, polysaccharides, amino acids, nucleic acids, and fatty acids. Ions of sodium, potassium, calcium, and other elements are also found in the cytosol. Many metabolic reactions, including protein synthesis, take place in this part of the cell.

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THE ENDOMEMBRANE SYSTEM

Learning Outcomes

- Describe the structure and function of the nucleus and nuclear membrane
- Describe the structure, function, and components of the endomembrane system

The endomembrane system (*endo* = within) is a group of membranes and organelles (Figure 1) in eukaryotic cells that work together to modify, package, and transport lipids and proteins. It includes the nuclear envelope, lysosomes (which only appear in animal cells), vesicles, the endoplasmic reticulum, and Golgi apparatus, which we will cover shortly.

The Nucleus

Typically, the nucleus is the most prominent organelle in a cell (Figure 1). The nucleus (plural = nuclei) houses the cell's DNA in the form of chromatin and directs the synthesis of ribosomes and proteins. Let us look at it in more detail (Figure 1).

The nuclear envelope is a double-membrane structure that constitutes the outermost portion of the nucleus (Figure 1). Both the inner and outer membranes of the nuclear envelope are phospholipid bilayers.

The nuclear envelope is punctuated with pores that control the passage of ions, molecules, and RNA between the nucleoplasm and the cytoplasm.

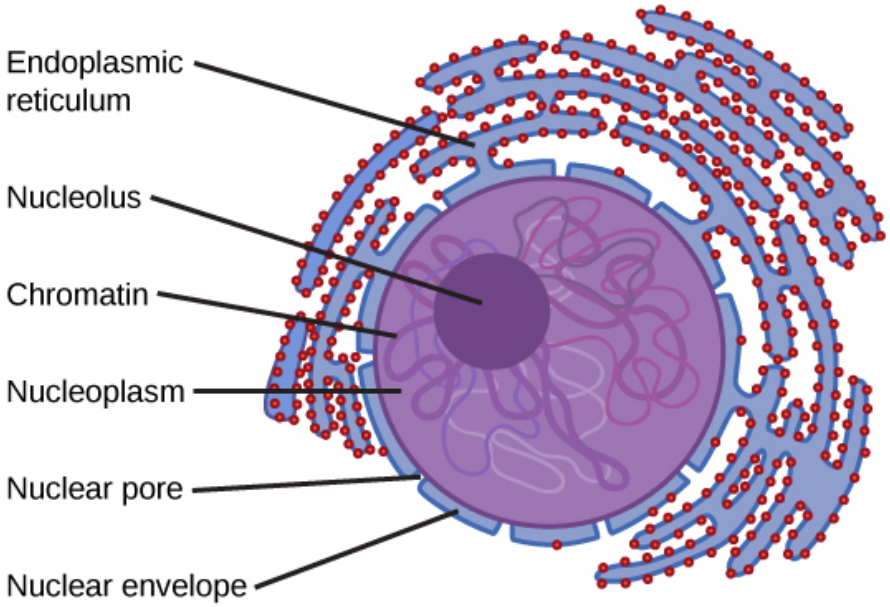

Figure 1. The outermost boundary of the nucleus is the nuclear envelope. Notice that the nuclear envelope consists of two phospholipid bilayers (membranes)—an outer membrane and an inner membrane—in contrast to the plasma membrane, which consists of only one phospholipid bilayer. (credit: modification of work by NIGMS, NIH)

To understand chromatin, it is helpful to first consider chromosomes. Chromosomes are structures within the nucleus that are made up of DNA, the hereditary material, and proteins. This combination of DNA and proteins is called chromatin. In eukaryotes, chromosomes are linear structures. Every species has a specific number of chromosomes in the nucleus of its body cells. For example, in humans, the chromosome number is 46, whereas in fruit flies, the chromosome number is eight.

Chromosomes are only visible and distinguishable from one another when the cell is getting ready to divide. When the cell is in the growth and maintenance phases of its life cycle, the chromosomes resemble an unwound, jumbled bunch of threads, which is the chromatin.

We already know that the nucleus directs the synthesis of ribosomes, but how does it do this? Some chromosomes have sections of DNA that encode ribosomal RNA. A darkly staining area within the nucleus, called the nucleolus (plural = nucleoli), aggregates the ribosomal RNA with associated proteins to assemble the ribosomal subunits that are then transported through the nuclear pores into the cytoplasm.

The Endoplasmic Reticulum

The endoplasmic reticulum (ER) (Figure 1) is a series of interconnected membranous tubules that collectively modify proteins and synthesize lipids. However, these two functions are performed in separate areas of the endoplasmic reticulum: the rough endoplasmic reticulum and the smooth endoplasmic reticulum, respectively.

The hollow portion of the ER tubules is called the lumen or cisternal space. The membrane of the ER, which is a phospholipid bilayer embedded with proteins, is continuous with the nuclear envelope.

The rough endoplasmic reticulum (RER) is so named because the ribosomes attached to its cytoplasmic surface give it a studded appearance when viewed through an electron microscope.

The ribosomes synthesize proteins while attached to the ER, resulting in transfer of their newly synthesized proteins into the lumen of the RER where they undergo modifications such as folding or addition of sugars. The RER also makes phospholipids for cell membranes.

If the phospholipids or modified proteins are not destined to stay in the RER, they will be packaged within vesicles and transported from the RER by budding from the membrane (Figure 1). Since the RER is engaged in modifying proteins that will be secreted from the cell, it is abundant in cells that secrete proteins, such as the liver.

The smooth endoplasmic reticulum (SER) is continuous with the RER but has few or no ribosomes on its cytoplasmic surface (see Figure 1). The SER's functions include synthesis of carbohydrates, lipids (including phospholipids), and steroid hormones; detoxification of medications and poisons; alcohol metabolism; and storage of calcium ions.

The Golgi Apparatus

We have already mentioned that vesicles can bud from the ER, but where do the vesicles go? Before reaching their final destination, the lipids or proteins within the transport vesicles need to be sorted, packaged, and tagged so that they wind up in the right place. The sorting, tagging, packaging, and distribution of lipids and proteins take place in the Golgi apparatus (also called the Golgi body), a series of flattened membranous sacs (Figure 2).

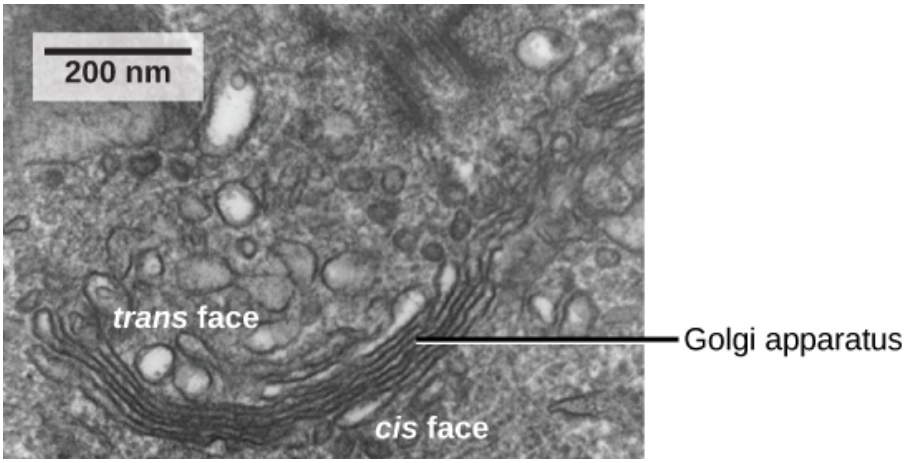

Figure 2. The Golgi apparatus in this transmission electron micrograph of a white blood cell is visible as a stack of semicircular flattened rings in the lower portion of this image. Several vesicles can be seen near the Golgi apparatus. (credit: modification of work by Louisa Howard; scale-bar data from Matt Russell)

The Golgi apparatus has a receiving (*cis*) face near the endoplasmic reticulum and a releasing (*trans*) face on the side away from the ER, toward the cell membrane. The transport vesicles that form from the ER travel to the receiving face, fuse with it, and empty their contents into the lumen of the Golgi apparatus. As the proteins and lipids travel through the Golgi, they undergo further modifications. The most frequent modification is the addition of short chains of sugar molecules. The newly modified proteins and lipids are then tagged with small molecular groups so that they are routed to their proper destinations.

Finally, the modified and tagged proteins are packaged into vesicles that bud from the opposite face of the Golgi. While some of these vesicles, transport vesicles, deposit their contents into other parts of the cell where they will be used, others, secretory vesicles, fuse with the plasma membrane and release their contents outside the cell.

The amount of Golgi in different cell types again illustrates that form follows function within cells. Cells that engage in a great deal of secretory activity (such as cells of the salivary glands that secrete digestive enzymes or cells of the immune system that secrete antibodies) have an abundant number of Golgi.

In plant cells, the Golgi has an additional role of synthesizing polysaccharides, some of which are incorporated into the cell wall and some of which are used in other parts of the cell.

Practice Question

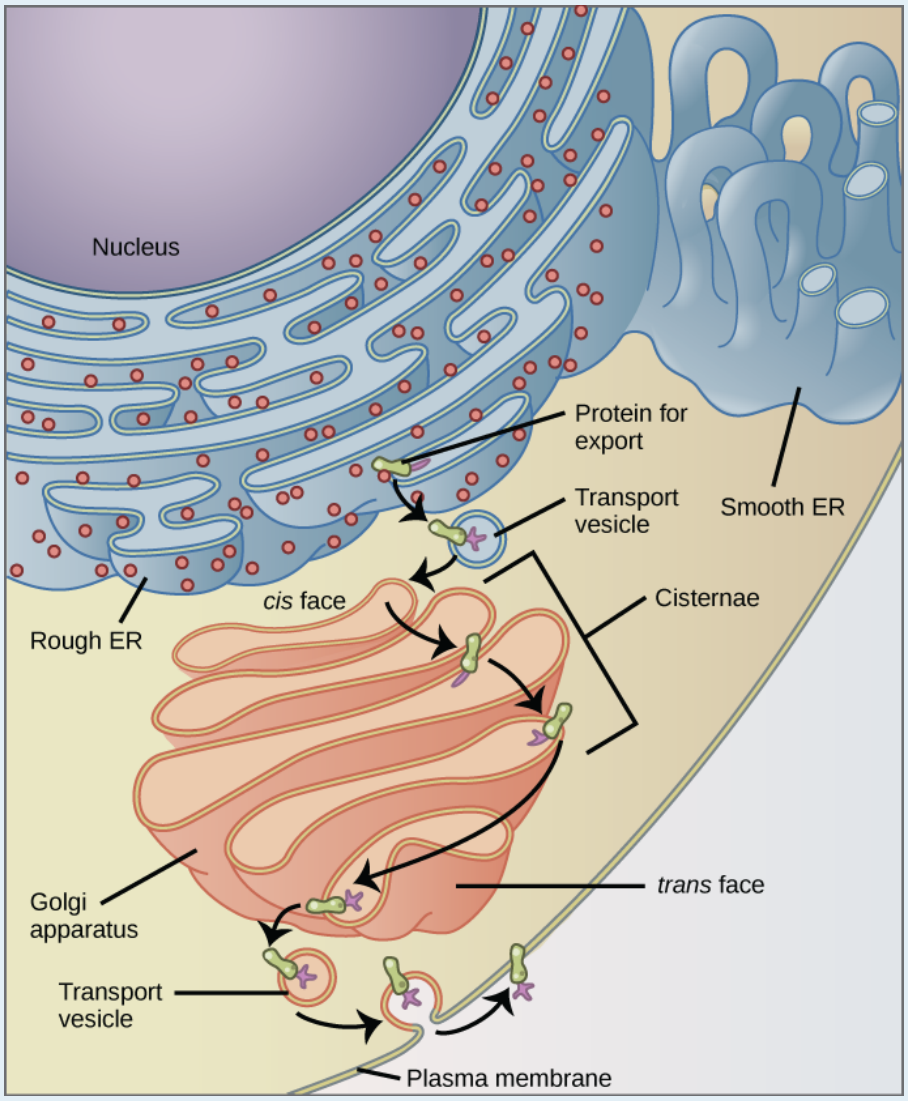

Figure 3. The endomembrane system works to modify, package, and transport lipids and proteins. (credit: modification of work by Magnus Manske)

Why does the *cis* face of the Golgi not face the plasma membrane?

Answer

Because that face receives chemicals from the ER, which is toward the center of the cell.

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RIBOSOMES, MITOCHONDRIA, VESICLES, AND PEROXISOMES

Learning Outcomes

- Describe the structure and function of ribosomes
- Describe the structure and function of mitochondria
- Describe the structure and functions of vesicles
- Describe the structure and function of peroxisomes

Ribosomes

Ribosomes are the cellular structures responsible for protein synthesis. When viewed through an electron microscope, free ribosomes appear as either clusters or single tiny dots floating freely in the cytoplasm. Ribosomes may be attached to either the cytoplasmic side of the plasma membrane or the cytoplasmic side of the endoplasmic reticulum (Figure 1).

Electron microscopy has shown that ribosomes consist of large and small subunits. Ribosomes are enzyme complexes that are responsible for protein synthesis and are composed of both proteins and RNA.

Because protein synthesis is essential for all cells, ribosomes are found in practically every cell, although they are smaller in prokaryotic cells. They are particularly abundant in immature red blood cells for the synthesis of hemoglobin, which functions in the transport of oxygen throughout the body.

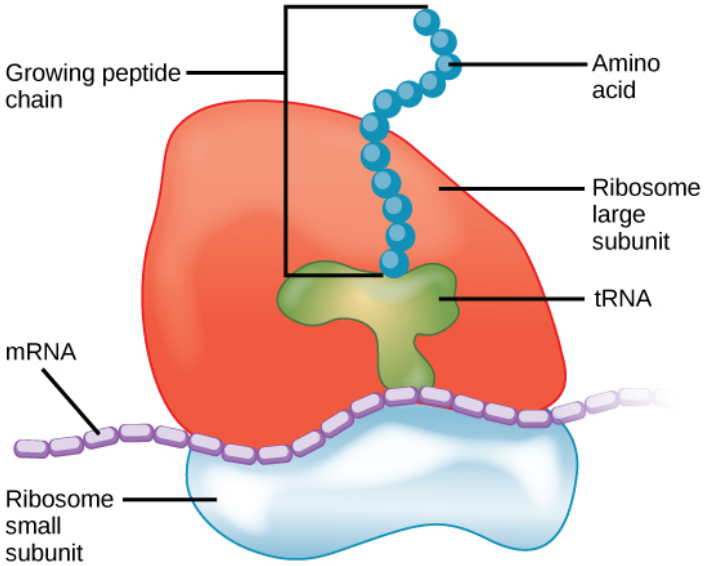

Figure 1. Ribosomes are made up of a large subunit (top) and a small subunit (bottom). During protein synthesis, ribosomes assemble amino acids into proteins.

Mitochondria

Mitochondria (singular = mitochondrion) are often called the “powerhouses” or “energy factories” of a cell because they are responsible for making adenosine triphosphate (ATP), the cell’s main energy-carrying molecule. The formation of ATP from the breakdown of glucose is known as cellular respiration. Mitochondria are oval-shaped, double-membrane organelles (Figure 2) that have their own ribosomes and DNA. Each membrane is a phospholipid bilayer embedded with proteins. The inner layer has folds called cristae, which increase the surface area of the inner membrane. The area surrounded by the folds is called the mitochondrial matrix. The cristae and the matrix have different roles in cellular respiration.

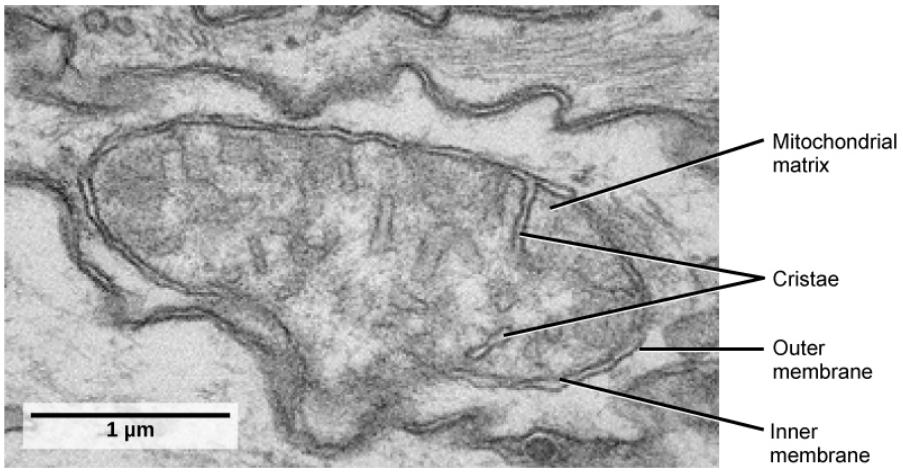

Figure 2. This transmission electron micrograph shows a mitochondrion as viewed with an electron microscope. Notice the inner and outer membranes, the cristae, and the mitochondrial matrix. (credit: modification of work by Matthew Britton; scale-bar data from Matt Russell)

In keeping with our theme of form following function, it is important to point out that muscle cells have a very high concentration of mitochondria because muscle cells need a lot of energy to contract.

Vesicles

Vesicles are membrane-bound sacs that function in storage and transport. The membrane of a vesicle can fuse with the membranes of other cellular components.

Vesicles perform a variety of functions. Because they are separated from the cytosol, the inside of a vesicle can be different from the cytosolic environment. For this reason, vesicles are a basic tool used by the cell for organizing cellular substances. Vesicles are involved in metabolism, transport, buoyancy control, and enzyme storage. They can also act as chemical reaction chambers.

Peroxisomes

Peroxisomes are small, round organelles enclosed by single membranes. They carry out oxidation reactions that break down fatty acids and amino acids. They also detoxify many poisons that may enter the body. Alcohol is detoxified by peroxisomes in liver cells. A byproduct of these oxidation reactions is hydrogen peroxide, H_2O_2, which is contained within the peroxisomes to prevent the chemical from causing damage to cellular components outside of the organelle. Hydrogen peroxide is safely broken down by peroxisomal enzymes into water and oxygen.

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THE CYTOSKELETON, FLAGELLA AND CILIA, AND THE PLASMA MEMBRANE

Learning Outcomes

- Demonstrate familiarity with various components of the cytoskeleton
- Describe the structure and functions of flagella and cilia
- Explain the structure and function of cell membranes

The Cytoskeleton

If you were to remove all the organelles from a cell, would the plasma membrane and the cytoplasm be the only components left? No. Within the cytoplasm, there would still be ions and organic molecules, plus a network of protein fibers that helps to maintain the shape of the cell, secures certain organelles in specific positions, allows cytoplasm and vesicles to move within the cell, and enables unicellular organisms to move independently. Collectively, this network of protein fibers is known as the cytoskeleton. There are three types of fibers within the cytoskeleton: microfilaments, also known as actin filaments, intermediate filaments, and microtubules (Figure 1).

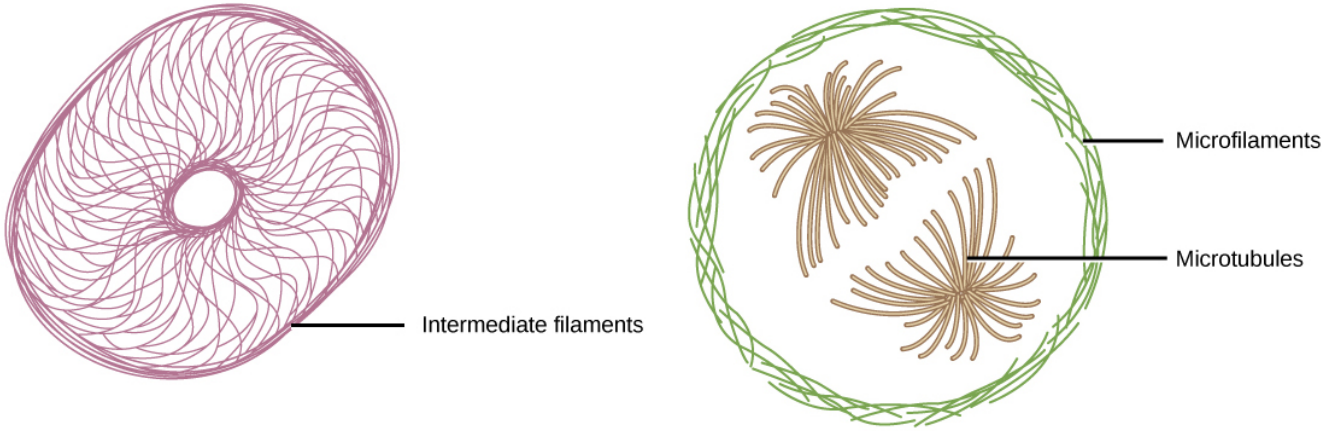

Figure 1. Microfilaments, intermediate filaments, and microtubules compose a cell's cytoskeleton.

Microfilaments are the thinnest of the cytoskeletal fibers and function in moving cellular components, for example, during cell division. They also maintain the structure of microvilli, the extensive folding of the plasma membrane found in cells dedicated to absorption. These components are also common in muscle cells and are responsible for muscle cell contraction.

Intermediate filaments are of intermediate diameter and have structural functions, such as maintaining the shape of the cell and anchoring organelles. Keratin, the compound that strengthens hair and nails, forms one type of intermediate filament.

Microtubules are the thickest of the cytoskeletal fibers. These are hollow tubes that can dissolve and reform quickly. Microtubules guide organelle movement and are the structures that pull chromosomes to their poles during cell division. They are also the structural components of flagella and cilia. In cilia and flagella, the microtubules are organized as a circle of nine double microtubules on the outside and two microtubules in the center.

The centrosome is a region near the nucleus of animal cells that functions as a microtubule-organizing center. It contains a pair of centrioles, two structures that lie perpendicular to each other. Each centriole is a cylinder of nine triplets of microtubules. The centrosome replicates itself before a cell divides, and the centrioles play a role in pulling the duplicated chromosomes to opposite ends of the dividing cell. However, the exact function of the centrioles in cell division is not clear, since cells that have the centrioles removed can still divide, and plant cells, which lack centrioles, are capable of cell division.

Flagella and Cilia

Flagella (singular = *flagellum*) are long, hair-like structures that extend from the plasma membrane and are used to move an entire cell, (for example, sperm, *Euglena*). When present, the cell has just one flagellum or a few flagella. When cilia (singular = *cilium*) are present, however, they are many in number and extend along the entire surface of the plasma membrane. They are short, hair-like structures that are used to move entire cells (such as paramecium) or move substances along the outer surface of the cell (for example, the cilia of cells lining the Fallopian tubes that move the ovum toward the uterus, or cilia lining the cells of the respiratory tract that move particulate matter toward the throat that mucus has trapped).

The Plasma Membrane

Like prokaryotes, eukaryotic cells have a plasma membrane (Figure 2) made up of a phospholipid bilayer with embedded proteins that separates the internal contents of the cell from its surrounding environment. A phospholipid is a lipid molecule composed of two fatty acid chains and a phosphate group. The plasma membrane regulates the passage of some substances, such as organic molecules, ions, and water, preventing the passage of some to maintain internal conditions, while actively bringing in or removing others. Other compounds move passively across the membrane.

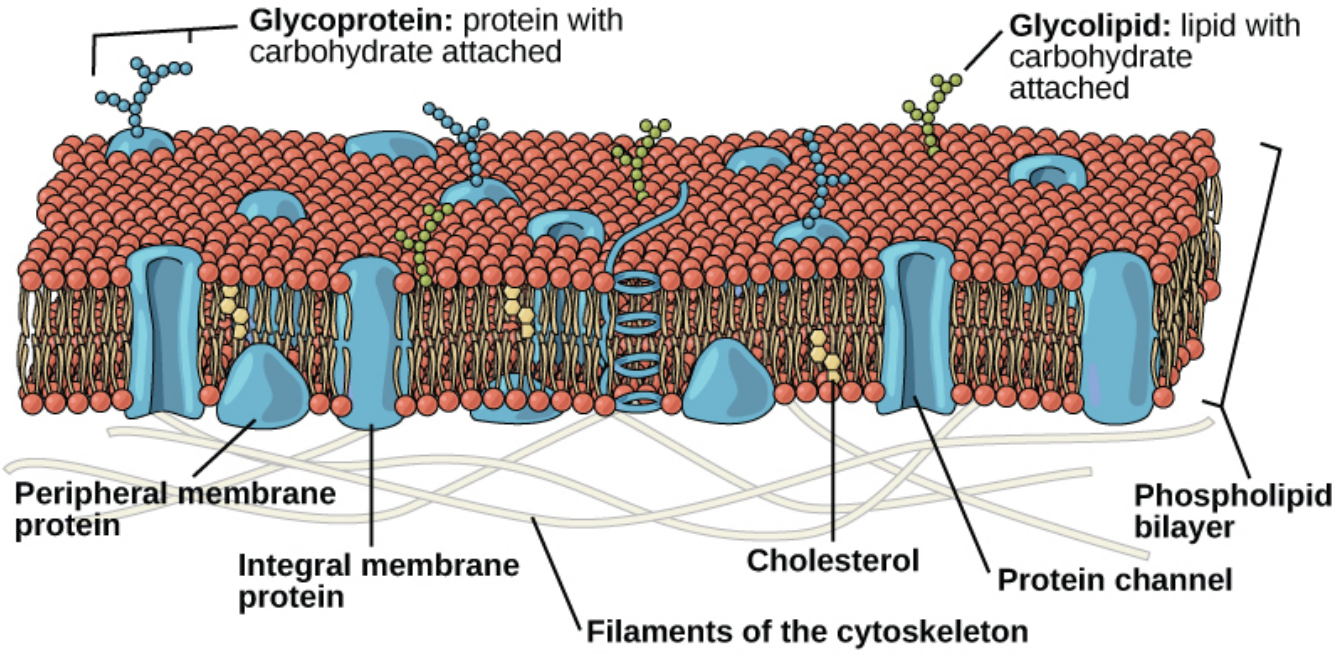

Figure 2. The plasma membrane is a phospholipid bilayer with embedded proteins. There are other components, such as cholesterol and carbohydrates, which can be found in the membrane in addition to phospholipids and protein.

The plasma membranes of cells that specialize in absorption are folded into fingerlike projections called microvilli (singular = microvillus). This folding increases the surface area of the plasma membrane. Such cells are typically found lining the small intestine, the organ that absorbs nutrients from digested food. This is an excellent example of form matching the function of a structure. People with celiac disease have an immune response to gluten, which is a protein found in wheat, barley, and rye. The immune response damages microvilli, and thus, afflicted

individuals cannot absorb nutrients. This leads to malnutrition, cramping, and diarrhea. Patients suffering from celiac disease must follow a gluten-free diet.

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ANIMAL CELLS VERSUS PLANT CELLS

Learning Outcomes

- Identify key organelles present only in plant cells, including chloroplasts and vacuoles
- Identify key organelles present only in animal cells, including centrosomes and lysosomes

At this point, it should be clear that eukaryotic cells have a more complex structure than do prokaryotic cells. Organelles allow for various functions to occur in the cell at the same time. Despite their fundamental similarities, there are some striking differences between animal and plant cells (see Figure 1).

Animal cells have centrioles, centrosomes (discussed under the cytoskeleton), and lysosomes, whereas plant cells do not. Plant cells have a cell wall, chloroplasts, plasmodesmata, and plastids used for storage, and a large central vacuole, whereas animal cells do not.

Practice Question

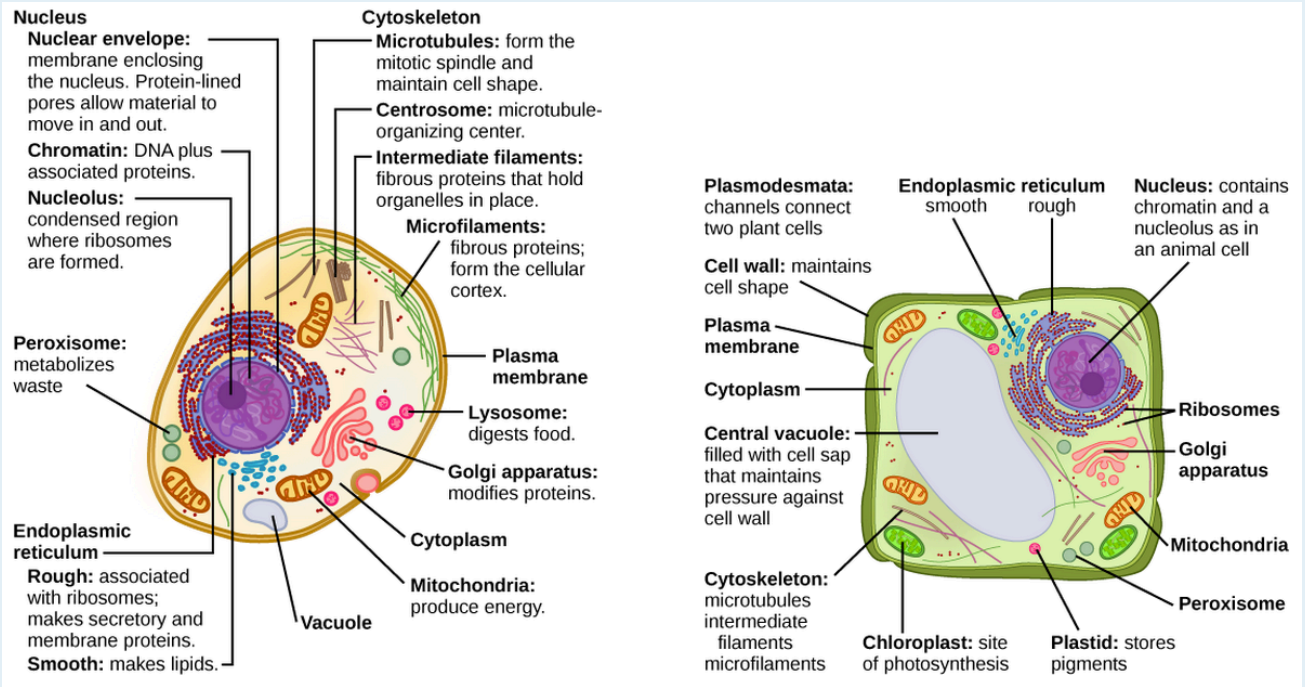

Figure 1. (a) A typical animal cell and (b) a typical plant cell.

What structures does a plant cell have that an animal cell does not have? What structures does an animal cell have that a plant cell does not have?

Answer

Plant cells have plasmodesmata, a cell wall, a large central vacuole, chloroplasts, and plastids. Animal cells have lysosomes and centrosomes.

Plant Cells

The Cell Wall

In Figure 1b, the diagram of a plant cell, you see a structure external to the plasma membrane called the cell wall. The cell wall is a rigid covering that protects the cell, provides structural support, and gives shape to the cell. Fungal and protist cells also have cell walls.

While the chief component of prokaryotic cell walls is peptidoglycan, the major organic molecule in the plant cell wall is cellulose (Figure 2), a polysaccharide made up of long, straight chains of glucose units. When nutritional information refers to dietary fiber, it is referring to the cellulose content of food.

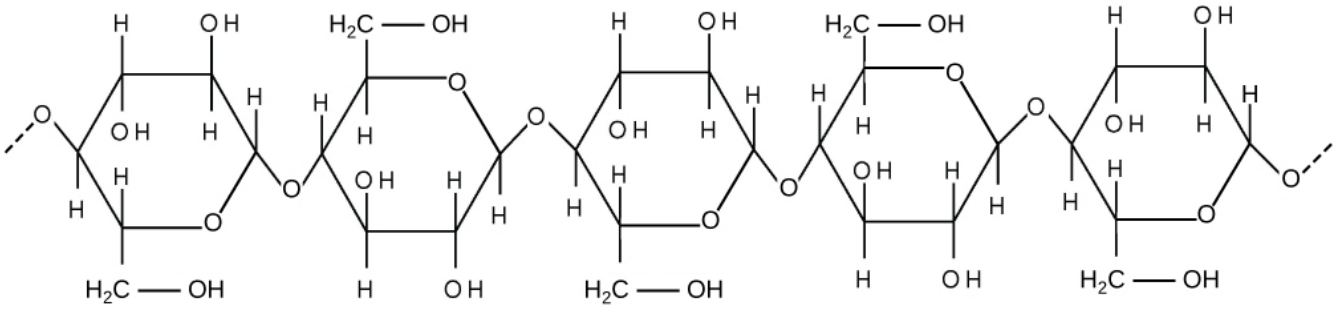

Figure 2. Cellulose is a long chain of β-glucose molecules connected by a 1–4 linkage. The dashed lines at each end of the figure indicate a series of many more glucose units. The size of the page makes it impossible to portray an entire cellulose molecule.

Chloroplasts

Like mitochondria, chloroplasts also have their own DNA and ribosomes. Chloroplasts function in photosynthesis and can be found in photoautotrophic eukaryotic cells such as plants and algae. In photosynthesis, carbon dioxide, water, and light energy are used to make glucose and oxygen. This is the major difference between plants and animals: Plants (autotrophs) are able to make their own food, like glucose, whereas animals (heterotrophs) must rely on other organisms for their organic compounds or food source.

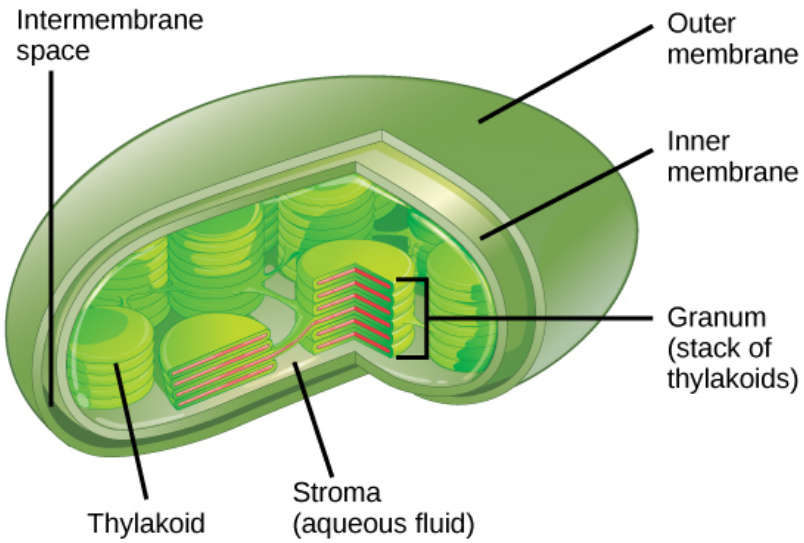

Figure 3. This simplified diagram of a chloroplast shows the outer membrane, inner membrane, thylakoids, grana, and stroma.

Like mitochondria, chloroplasts have outer and inner membranes, but within the space enclosed by a chloroplast's inner membrane is a set of interconnected and stacked, fluid-filled membrane sacs called thylakoids (Figure 3). Each stack of thylakoids is called a granum (plural = *grana*). The fluid enclosed by the inner membrane and surrounding the grana is called the stroma.

The chloroplasts contain a green pigment called chlorophyll, which captures the energy of sunlight for photosynthesis. Like plant cells, photosynthetic protists also have chloroplasts. Some bacteria also perform photosynthesis, but they do not have chloroplasts. Their photosynthetic pigments are located in the thylakoid membrane within the cell itself.

Endosymbiosis

We have mentioned that both mitochondria and chloroplasts contain DNA and ribosomes. Have you wondered why? Strong evidence points to endosymbiosis as the explanation.

Symbiosis is a relationship in which organisms from two separate species live in close association and typically exhibit specific adaptations to each other. Endosymbiosis (*endo-*= within) is a relationship in which one organism lives inside the other. Endosymbiotic relationships abound in nature. Microbes that produce vitamin K live inside the human gut. This relationship is beneficial for us because we are unable to synthesize vitamin K. It is also beneficial for the microbes because they are protected from other organisms and are provided a stable habitat and abundant food by living within the large intestine.

Scientists have long noticed that bacteria, mitochondria, and chloroplasts are similar in size. We also know that mitochondria and chloroplasts have DNA and ribosomes, just as bacteria do. Scientists believe that host cells and bacteria formed a mutually beneficial endosymbiotic relationship when the host cells ingested aerobic bacteria and cyanobacteria but did not destroy them. Through evolution, these ingested bacteria became more specialized in their functions, with the aerobic bacteria becoming mitochondria and the photosynthetic bacteria becoming chloroplasts.

The Central Vacuole

Previously, we mentioned vacuoles as essential components of plant cells. If you look at Figure 1b, you will see that plant cells each have a large, central vacuole that occupies most of the cell. The central vacuole plays a key role in regulating the cell's concentration of water in changing environmental conditions. In plant cells, the liquid inside the central vacuole provides turgor pressure, which is the outward pressure caused by the fluid inside the cell. Have you ever noticed that if you forget to water a plant for a few days, it wilts? That is because as the water

concentration in the soil becomes lower than the water concentration in the plant, water moves out of the central vacuoles and cytoplasm and into the soil. As the central vacuole shrinks, it leaves the cell wall unsupported. This loss of support to the cell walls of a plant results in the wilted appearance. When the central vacuole is filled with water, it provides a low energy means for the plant cell to expand (as opposed to expending energy to actually increase in size). Additionally, this fluid can deter herbivory since the bitter taste of the wastes it contains discourages consumption by insects and animals. The central vacuole also functions to store proteins in developing seed cells.

Animal Cells

Lysosomes

In animal cells, the lysosomes are the cell's "garbage disposal." Digestive enzymes within the lysosomes aid the breakdown of proteins, polysaccharides, lipids, nucleic acids, and even worn-out organelles. In single-celled eukaryotes, lysosomes are important for digestion of the food they ingest and the recycling of organelles. These enzymes are active at a much lower pH (more acidic) than those located in the cytoplasm. Many reactions that take place in the cytoplasm could not occur at a low pH, thus the advantage of compartmentalizing the eukaryotic cell into organelles is apparent.

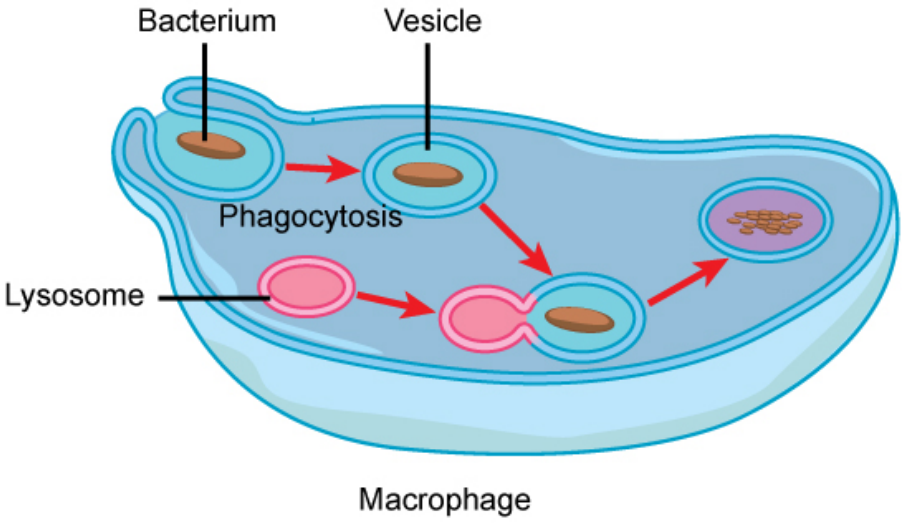

Figure 4. A macrophage has phagocytized a potentially pathogenic bacterium into a vesicle, which then fuses with a lysosome within the cell so that the pathogen can be destroyed. Other organelles are present in the cell, but for simplicity, are not shown.

Lysosomes also use their hydrolytic enzymes to destroy disease-causing organisms that might enter the cell. A good example of this occurs in a group of white blood cells called macrophages, which are part of your body's immune system. In a process known as phagocytosis, a section of the plasma membrane of the macrophage invaginates (folds in) and engulfs a pathogen. The invaginated section, with the pathogen inside, then pinches itself off from the plasma membrane and becomes a vesicle. The vesicle fuses with a lysosome. The lysosome's hydrolytic enzymes then destroy the pathogen (Figure 4).

Extracellular Matrix of Animal Cells

Most animal cells release materials into the extracellular space. The primary components of these materials are glycoproteins and the protein collagen. Collectively, these materials are called the extracellular matrix (Figure 5). Not only does the extracellular matrix hold the cells together to form a tissue, but it also allows the cells within the tissue to communicate with each other.

Blood clotting provides an example of the role of the extracellular matrix in cell communication. When the cells lining a blood vessel are damaged, they display a protein receptor called tissue factor. When tissue factor binds with another factor in the extracellular matrix, it causes platelets to adhere to the wall of the damaged blood vessel, stimulates adjacent smooth muscle cells in the blood vessel to contract (thus constricting the blood vessel), and initiates a series of steps that stimulate the platelets to produce clotting factors.

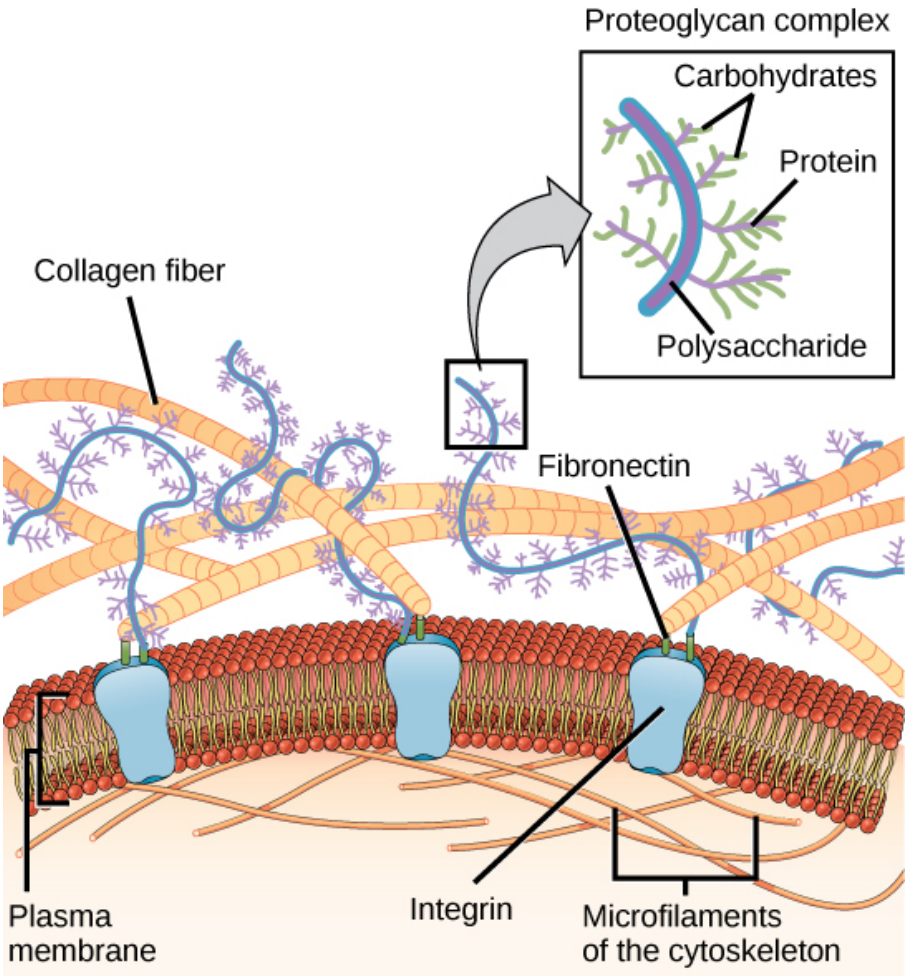

Figure 5. The extracellular matrix consists of a network of substances secreted by cells.

Intercellular Junctions

Cells can also communicate with each other by direct contact, referred to as intercellular junctions. There are some differences in the ways that plant and animal cells do this. Plasmodesmata (singular = plasmodesma) are junctions between plant cells, whereas animal cell contacts include tight and gap junctions, and desmosomes.

In general, long stretches of the plasma membranes of neighboring plant cells cannot touch one another because they are separated by the cell walls surrounding each cell. Plasmodesmata are numerous channels that pass between the cell walls of adjacent plant cells, connecting their cytoplasm and enabling signal molecules and nutrients to be transported from cell to cell (Figure 6a).

A tight junction is a watertight seal between two adjacent animal cells (Figure 6b). Proteins hold the cells tightly against each other. This tight adhesion prevents materials from leaking between the cells. Tight junctions are typically found in the epithelial tissue that lines internal organs and cavities, and composes most of the skin. For example, the tight junctions of the epithelial cells lining the urinary bladder prevent urine from leaking into the extracellular space.

Also found only in animal cells are desmosomes, which act like spot welds between adjacent epithelial cells (Figure 6c). They keep cells together in a sheet-like formation in organs and tissues that stretch, like the skin, heart, and muscles.

Gap junctions in animal cells are like plasmodesmata in plant cells in that they are channels between adjacent cells that allow for the transport of ions, nutrients, and other substances that enable cells to communicate (Figure 6d). Structurally, however, gap junctions and plasmodesmata differ.

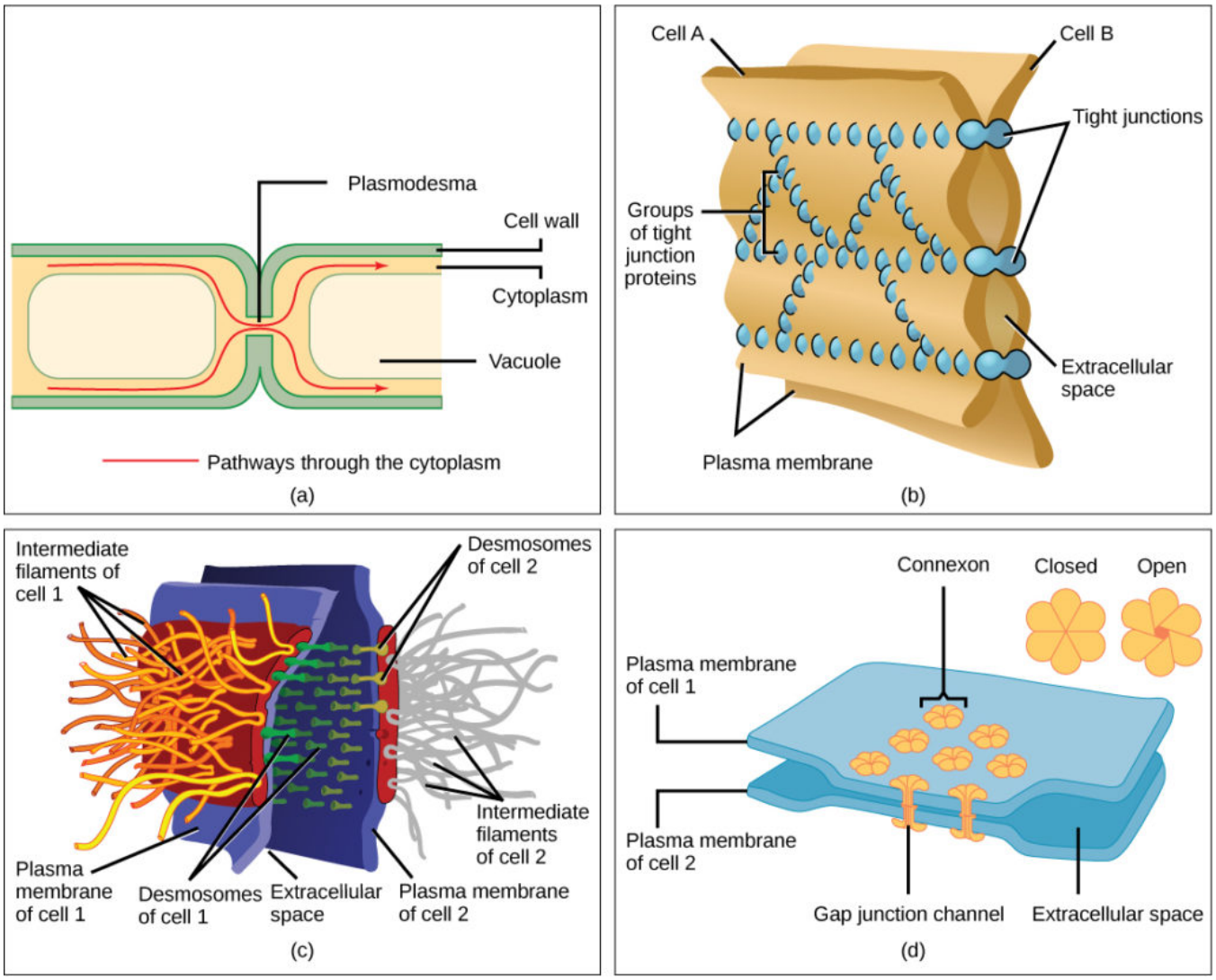

Figure 6. There are four kinds of connections between cells. (a) A plasmodesma is a channel between the cell walls of two adjacent plant cells. (b) Tight junctions join adjacent animal cells. (c) Desmosomes join two animal cells together. (d) Gap junctions act as channels between animal cells. (credit b, c, d: modification of work by Mariana Ruiz Villareal)

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SUMMARY: ORGANELLES

Learning Outcomes

- Describe the basic composition of cytoplasm
- Describe the structure and function of the nucleus and nuclear membrane
- Describe the structure, function, and components of the endomembrane system
- Describe the structure and function of ribosomes
- Describe the structure and function of mitochondria
- Describe the structure and functions of vesicles
- Describe the structure and function of peroxisomes
- Demonstrate familiarity with various components of the cytoskeleton
- Describe the structure and functions of flagella and cilia
- Explain the structure and function of cell membranes
- Identify key organelles present only in plant cells, including chloroplasts and vacuoles
- Identify key organelles present only in animal cells, including centrosomes and lysosomes

Table 1 provides the components of prokaryotic and eukaryotic cells and their respective functions.

Table 1. Components of Prokaryotic and Eukaryotic Cells and Their Functions

Cell Component	Function	Present in Prokaryotes?	Present in Animal Cells?	Present in Plant Cells?
Plasma membrane	Separates cell from external environment; controls passage of organic molecules, ions, water, oxygen, and wastes into and out of the cell	Yes	Yes	Yes
Cytoplasm	Provides structure to cell; site of many metabolic reactions; medium in which organelles are found	Yes	Yes	Yes
Nucleoid	Location of DNA	Yes	No	No
Nucleus	Cell organelle that houses DNA and directs synthesis of ribosomes and proteins	No	Yes	Yes
Ribosomes	Protein synthesis	Yes	Yes	Yes
Mitochondria	ATP production/cellular respiration	No	Yes	Yes
Peroxisomes	Oxidizes and breaks down fatty acids and amino acids, and detoxifies poisons	No	Yes	Yes
Vesicles and vacuoles	Storage and transport; digestive function in plant cells	No	Yes	Yes
Centrosome	Unspecified role in cell division in animal cells; source of microtubules in animal cells	No	Yes	No

Table 1. Components of Prokaryotic and Eukaryotic Cells and Their Functions				
Cell Component	Function	Present in Prokaryotes?	Present in Animal Cells?	Present in Plant Cells?
Lysosomes	Digestion of macromolecules; recycling of worn-out organelles	No	Yes	No
Cell wall	Protection, structural support and maintenance of cell shape	Yes, primarily peptidoglycan in bacteria but not Archaea	No	Yes, primarily cellulose
Chloroplasts	Photosynthesis	No	No	Yes
Endoplasmic reticulum	Modifies proteins and synthesizes lipids	No	Yes	Yes
Golgi apparatus	Modifies, sorts, tags, packages, and distributes lipids and proteins	No	Yes	Yes
Cytoskeleton	Maintains cell's shape, secures organelles in specific positions, allows cytoplasm and vesicles to move within the cell, and enables unicellular organisms to move independently	Yes	Yes	Yes
Flagella	Cellular locomotion	Some	Some	No, except for some plant sperm.
Cilia	Cellular locomotion, movement of particles along extracellular surface of plasma membrane, and filtration	No	Some	No

Like a prokaryotic cell, a eukaryotic cell has a plasma membrane, cytoplasm, and ribosomes, but a eukaryotic cell is typically larger than a prokaryotic cell, has a true nucleus (meaning its DNA is surrounded by a membrane), and has other membrane-bound organelles that allow for compartmentalization of functions. The plasma membrane is a phospholipid bilayer embedded with proteins. The nucleolus within the nucleus is the site for ribosome assembly. Ribosomes are found in the cytoplasm or are attached to the cytoplasmic side of the plasma membrane or endoplasmic reticulum. They perform protein synthesis. Mitochondria perform cellular respiration and produce ATP. Peroxisomes break down fatty acids, amino acids, and some toxins. Vesicles and vacuoles are storage and transport compartments. In plant cells, vacuoles also help break down macromolecules.

Animal cells also have a centrosome and lysosomes. The centrosome has two bodies, the centrioles, with an unknown role in cell division. Lysosomes are the digestive organelles of animal cells.

Plant cells have a cell wall, chloroplasts, and a central vacuole. The plant cell wall, whose primary component is cellulose, protects the cell, provides structural support, and gives shape to the cell. Photosynthesis takes place in chloroplasts. The central vacuole expands, enlarging the cell without the need to produce more cytoplasm.

The endomembrane system includes the nuclear envelope, the endoplasmic reticulum, Golgi apparatus, lysosomes, vesicles, as well as the plasma membrane. These cellular components work together to modify, package, tag, and transport membrane lipids and proteins.

The cytoskeleton has three different types of protein elements. Microfilaments provide rigidity and shape to the cell, and facilitate cellular movements. Intermediate filaments bear tension and anchor the nucleus and other organelles in place. Microtubules help the cell resist compression, serve as tracks for motor proteins that move vesicles through the cell, and pull replicated chromosomes to opposite ends of a dividing cell. They are also the structural elements of centrioles, flagella, and cilia.

Animal cells communicate through their extracellular matrices and are connected to each other by tight junctions, desmosomes, and gap junctions. Plant cells are connected and communicate with each other by plasmodesmata.

Practice Question

In the context of cell biology, what do we mean by form follows function? What are at least two examples of this concept?

Answer

"Form follows function" refers to the idea that the function of a body part dictates the form of that body part. As an example, organisms like birds or fish that fly or swim quickly through the air or water have streamlined bodies that reduce drag. At the level of the cell, in tissues involved in secretory functions, such as the salivary glands, the cells have abundant Golgi.

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PUTTING IT TOGETHER: CELLULAR STRUCTURE

In this chapter, we've learned about several different cell components. View this video from Khan Academy to learn more about cells.

Watch this video online: https://youtu.be/jVSUQmtTk7Y

Table 1 provides a summary of all the components we covered, as well as their functions and locations.

Table 1. Components of Prokaryotic and Eukaryotic Cells

Cell Component	Function	Present in Prokaryotes?	Present in Animal Cells?	Present in Plant Cells?
Cytoplasm	Provides turgor pressure to plant cells as fluid inside the central vacuole; site of many metabolic reactions; medium in which organelles are found	Yes	Yes	Yes
Nucleus	Cell organelle that houses DNA and directs synthesis of ribosomes and proteins	No	Yes	Yes

Table 1. Components of Prokaryotic and Eukaryotic Cells				
Cell Component	Function	Present in Prokaryotes?	Present in Animal Cells?	Present in Plant Cells?
Nucleolus	Darkened area within the nucleus where ribosomal subunits are synthesized.	No	Yes	Yes
Ribosomes	Protein synthesis	Yes	Yes	Yes
Mitochondria	ATP production/cellular respiration	No	Yes	Yes
Peroxisomes	Oxidizes and thus breaks down fatty acids and amino acids, and detoxifies poisons	No	Yes	Yes
Endoplasmic reticulum	Modifies proteins and synthesizes lipids	No	Yes	Yes
Golgi apparatus	Modifies, sorts, tags, packages, and distributes lipids and proteins	No	Yes	Yes
Vesicles and vacuoles	Storage and transport; digestive function in plant cells	No	Yes	Yes
Centrosome	Unspecified role in cell division in animal cells; source of microtubules in animal cells	No	Yes	No
Lysosomes	Digestion of macromolecules; recycling of worn-out organelles	No	Yes	No
Chloroplasts	Photosynthesis	No	No	Yes
Cytoskeleton	Maintains cell's shape, secures organelles in specific positions, allows cytoplasm and vesicles to move within cell, and enables unicellular organisms to move independently	Yes	Yes	Yes
Flagella	Cellular locomotion	Some	Some	No, except for some plant sperm cells.
Cilia	Cellular locomotion, movement of particles along extracellular surface of plasma membrane, and filtration	Some	Some	No
Plasma membrane	Separates cell from external environment; controls passage of organic molecules, ions, water, oxygen, and wastes into and out of cell	Yes	Yes	Yes

Table 1. Components of Prokaryotic and Eukaryotic Cells				
Cell Component	Function	Present in Prokaryotes?	Present in Animal Cells?	Present in Plant Cells?
Cell wall	Protection, structural support and maintenance of cell shape	Yes, primarily peptidoglycan	No	Yes, primarily cellulose

Inherited Diseases

Let's look back to our earlier disease list:

- **Pompe Disease**: characterized by excess accumulation of glycogen in muscle cells
- **Leigh Disease**: progressive disorder of lesions (dead or dying cells) in the brain
- **Emery-Dreifuss muscular dystrophy**: wasting and weakness in muscles of the shoulders, upper arms, and calf muscles

The following organelles are what cause the each disorder. As you explore the different websites that highlight each disease, think about how the function of these organelles directly relates to the disease symptoms.

- Pompe disease: lysosomes
- Leigh's Disease: mitochondria
- Emery-Dreifuss muscular dystrophy: nuclear envelope

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- Nuclei, membranes, ribosomes, eukaryotes and prokaryotes. **Authored by**: Khan Academy. **Located at**: https://youtu.be/jVSUQmtTk7Y. **License**: *CC BY: Attribution*. **License Terms**: Standard YouTube License

MODULE 5: CELL MEMBRANES

WHY IT MATTERS: CELL MEMBRANES

Why learn about cell membranes?

Cystic fibrosis (CF) is a genetic disorder that primarily affects the lungs, as well as the pancreas, liver, and intestine. CF is characterized by abnormal transport of chloride and sodium across body tissues, leading to thick, viscous secretions. The hallmark symptoms of cystic fibrosis are salty tasting skin, poor growth, poor weight gain despite a normal food intake, accumulation of thick, sticky mucus, frequent chest infections, and coughing or shortness of breath. Symptoms often appear in infancy and childhood, such as bowel obstruction in newborn babies.

The most serious symptoms of CF are difficulty breathing and frequent lung infections. Often, lung transplantation is ultimately necessary as CF worsens. Other symptoms, including sinus infections, poor growth, and infertility affect other parts of the body.

Cystic fibrosis results from the malfunctioning of a single membrane transporter. How could this error in membrane transport result in such a disease?

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INTRODUCTION TO STRUCTURE OF THE MEMBRANE

What you'll learn to do: Describe the structure and function of membranes, especially the phospholipid bilayer

In this outcome, we'll learn about the structure of membranes:

Watch this video online: https://youtu.be/NQwpbuUa5bk

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- Cell membrane 1- Intro. **Authored by**: Wendy Riggs. **Located at**: https://youtu.be/NQwpbuUa5bk. **License**: *CC BY: Attribution*

STRUCTURE OF THE CELL MEMBRANE

Learning Outcomes

- Describe the structure of cell membranes
- Identify components of the cell membrane, including phospholipids, cholesterol, proteins, and carbohydrates

Cell Membranes are Fluid

A cell's plasma membrane defines the boundary of the cell and determines the nature of its contact with the environment. Cells exclude some substances, take in others, and excrete still others, all in controlled quantities. Plasma membranes enclose the borders of cells, but rather than being a static bag, they are dynamic and constantly in flux. The plasma membrane must be sufficiently flexible to allow certain cells, such as red blood cells and white blood cells, to change shape as they pass through narrow capillaries. These are the more obvious functions of a plasma membrane. In addition, the surface of the plasma membrane carries markers that allow cells to recognize one another, which is vital as tissues and organs form during early development, and which later plays a role in the "self" versus "non-self" distinction of the immune response.

The plasma membrane also carries receptors, which are attachment sites for specific substances that interact with the cell. Each receptor is structured to bind with a specific substance. For example, surface receptors of the membrane create changes in the interior, such as changes in enzymes of metabolic pathways. These metabolic pathways might be vital for providing the cell with energy, making specific substances for the cell, or breaking down cellular waste or toxins for disposal. Receptors on the plasma membrane's exterior surface interact with hormones or neurotransmitters, and allow their messages to be transmitted into the cell. Some recognition sites are used by viruses as attachment points. Although they are highly specific, pathogens like viruses may evolve to exploit receptors to gain entry to a cell by mimicking the specific substance that the receptor is meant to bind. This specificity helps to explain why human immunodeficiency virus (HIV) or any of the five types of hepatitis viruses invade only specific cells.

Cell Membranes are Mosaics

In 1972, S. J. Singer and Garth L. Nicolson proposed a new model of the plasma membrane that, compared to earlier understanding, better explained both microscopic observations and the function of the plasma membrane. This was called the **fluid mosaic model**. The model has evolved somewhat over time, but still best accounts for the structure and functions of the plasma membrane as we now understand them. The fluid mosaic model describes the structure of the plasma membrane as a mosaic of components—including phospholipids, cholesterol, proteins, and carbohydrates—in which the components are able to flow and change position, while maintaining the basic integrity of the membrane. Both phospholipid molecules and embedded proteins are able to diffuse rapidly and laterally in the membrane. The fluidity of the plasma membrane is necessary for the activities of certain enzymes and transport molecules within the membrane. Plasma membranes range from 5–10 nm thick. As a comparison, human red blood cells, visible via light microscopy, are approximately 8 μm thick, or approximately 1,000 times thicker than a plasma membrane. (Figure 1)

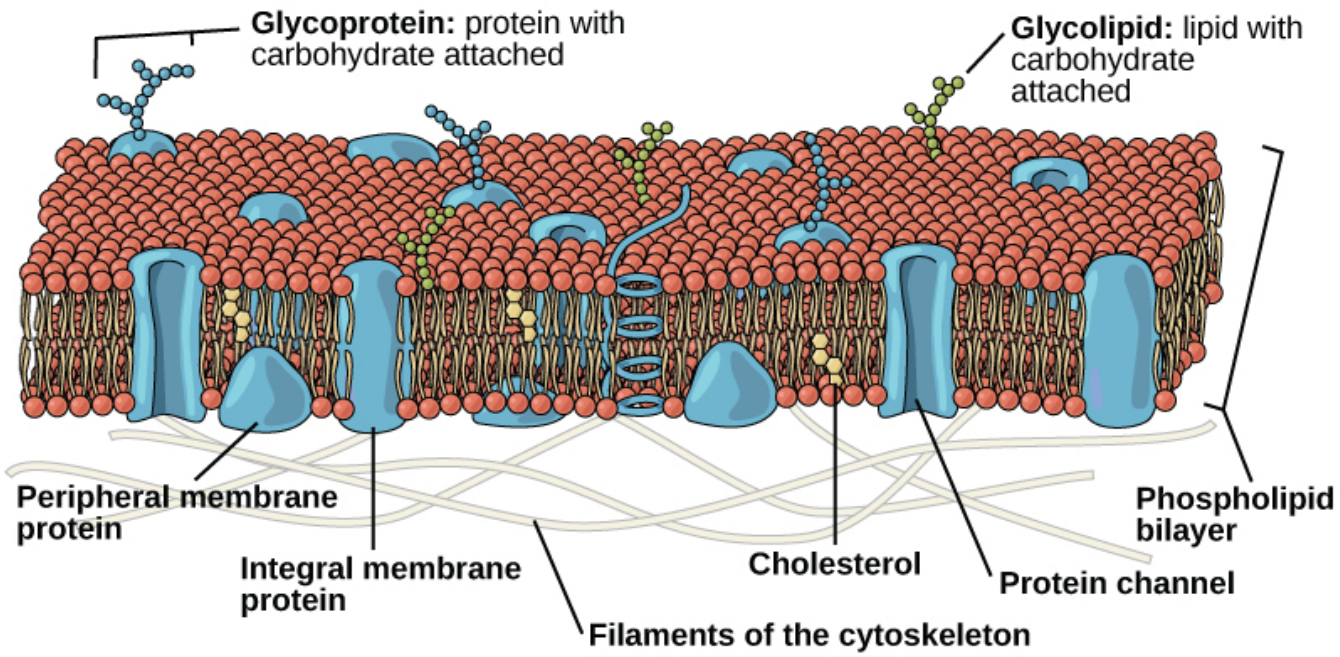

Figure 1. The fluid mosaic model of the plasma membrane structure describes the plasma membrane as a fluid combination of phospholipids, cholesterol, proteins, and carbohydrates.

The plasma membrane is made up primarily of a bilayer of phospholipids with embedded proteins, carbohydrates, glycolipids, and glycoproteins, and, in animal cells, cholesterol. The amount of cholesterol in animal plasma membranes regulates the fluidity of the membrane and changes based on the temperature of the cell's environment. In other words, cholesterol acts as antifreeze in the cell membrane and is more abundant in animals that live in cold climates.

The main fabric of the membrane is composed of two layers of phospholipid molecules, and the polar ends of these molecules (which look like a collection of balls in an artist's rendition of the model) (Figure 1) are in contact with aqueous fluid both inside and outside the cell. Thus, both surfaces of the plasma membrane are hydrophilic. In contrast, the interior of the membrane, between its two surfaces, is a hydrophobic or nonpolar region because of the fatty acid tails. This region has no attraction for water or other polar molecules.

Proteins make up the second major chemical component of plasma membranes. Integral proteins are embedded in the plasma membrane and may span all or part of the membrane. Integral proteins may serve as channels or pumps to move materials into or out of the cell. Peripheral proteins are found on the exterior or interior surfaces of membranes, attached either to integral proteins or to phospholipid molecules. Both integral and peripheral proteins may serve as enzymes, as structural attachments for the fibers of the cytoskeleton, or as part of the cell's recognition sites.

Carbohydrates are the third major component of plasma membranes. They are always found on the exterior surface of cells and are bound either to proteins (forming glycoproteins) or to lipids (forming glycolipids). These carbohydrate chains may consist of 2–60 monosaccharide units and may be either straight or branched. Along with peripheral proteins, carbohydrates form specialized sites on the cell surface that allow cells to recognize each other.

How Viruses Infect Specific Organs

Specific glycoprotein molecules exposed on the surface of the cell membranes of host cells are exploited by many viruses to infect specific organs. For example, HIV is able to penetrate the plasma membranes of specific kinds of white blood cells called T-helper cells and monocytes, as well as some cells of the central nervous system. The hepatitis virus attacks only liver cells.

These viruses are able to invade these cells, because the cells have binding sites on their surfaces that the viruses have exploited with equally specific glycoproteins in their coats. (Figure 2). The cell is tricked by the mimicry of the virus coat molecules, and the virus is able to enter the cell. Other recognition sites on the virus's surface interact with the human immune system, prompting the body to produce antibodies. Antibodies are made in response to the antigens (or proteins associated with invasive pathogens). These same sites serve as places for antibodies to attach, and either destroy or inhibit the activity of the virus. Unfortunately, these sites on HIV are encoded by genes that change quickly, making the production of an effective vaccine against the virus very difficult. The virus population within an infected individual quickly evolves through mutation into different populations, or variants, distinguished by differences in these recognition sites. This rapid change of viral surface markers decreases the effectiveness of the person's immune system in attacking the virus, because the antibodies will not recognize the new variations of the surface patterns.

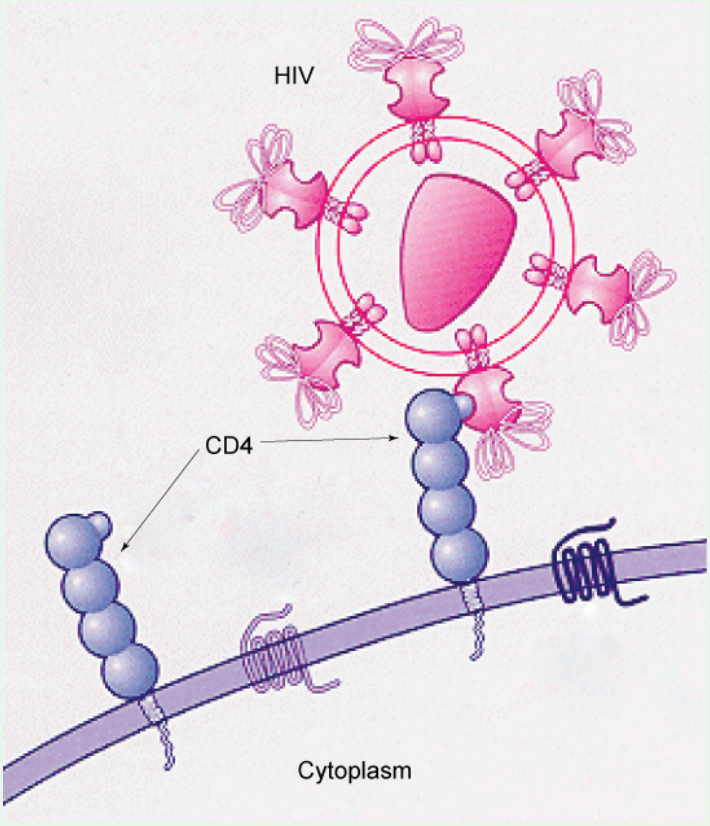

Figure 2. HIV docks at and binds to the CD4 receptor, a glycoprotein on the surface of T cells, before entering, or infecting, the cell. (credit: modification of work by US National Institutes of Health/National Institute of Allergy and Infectious Diseases)

In Summary: Structure of the Cell Membrane

The modern understanding of the plasma membrane is referred to as the fluid mosaic model. The plasma membrane is composed of a bilayer of phospholipids, with their hydrophobic, fatty acid tails in contact with each other. The landscape of the membrane is studded with proteins, some of which span the membrane. Some of these proteins serve to transport materials into or out of the cell. Carbohydrates are attached to some of the proteins and lipids on the outward-facing surface of the membrane. These form complexes that function to identify the cell to other cells. The fluid nature of the membrane owes itself to the configuration of the fatty acid tails, the presence of cholesterol embedded in the membrane (in animal cells), and the mosaic nature of the proteins and protein-carbohydrate complexes, which are not firmly fixed in place. Plasma membranes enclose the borders of cells, but rather than being a static bag, they are dynamic and constantly in flux.

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INTRODUCTION TO KINDS OF TRANSPORT

What you'll learn to do: Explain how substances are directly transported across a membrane

Plasma membranes must allow certain substances to enter and leave a cell, and prevent some harmful materials from entering and some essential materials from leaving. In other words, plasma membranes are **selectively permeable**—they allow some substances to pass through, but not others. If they were to lose this selectivity, the cell would no longer be able to sustain itself, and it would be destroyed. Some cells require larger amounts of specific substances than do other cells; they must have a way of obtaining these materials from extracellular fluids. This may happen passively, as certain materials move back and forth, or the cell may have special mechanisms that facilitate transport. Some materials are so important to a cell that it spends some of its energy, hydrolyzing adenosine triphosphate (ATP), to obtain these materials. For example, ed blood cells use some of their energy doing just that. All cells spend the majority of their energy to maintain an imbalance of sodium and potassium ions between the interior and exterior of the cell.

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PASSIVE TRANSPORT

Learning Outcomes

Explain how substances are directly transported across a membrane

The most direct forms of membrane transport are passive. **Passive transport** is a naturally occurring phenomenon and does not require the cell to exert any of its energy to accomplish the movement. In passive transport, substances move from an area of higher concentration to an area of lower concentration. A physical space in which there is a range of concentrations of a single substance is said to have a **concentration gradient**.

Selective Permeability

Plasma membranes are asymmetric: the interior of the membrane is not identical to the exterior of the membrane. In fact, there is a considerable difference between the array of phospholipids and proteins between the two leaflets that form a membrane. On the interior of the membrane, some proteins serve to anchor the membrane to fibers of the cytoskeleton. There are peripheral proteins on the exterior of the membrane that bind elements of the extracellular matrix. Carbohydrates, attached to lipids or proteins, are also found on the exterior surface of the plasma membrane. These carbohydrate complexes help the cell bind substances that the cell needs in the extracellular fluid. This adds considerably to the selective nature of plasma membranes (Figure 1).

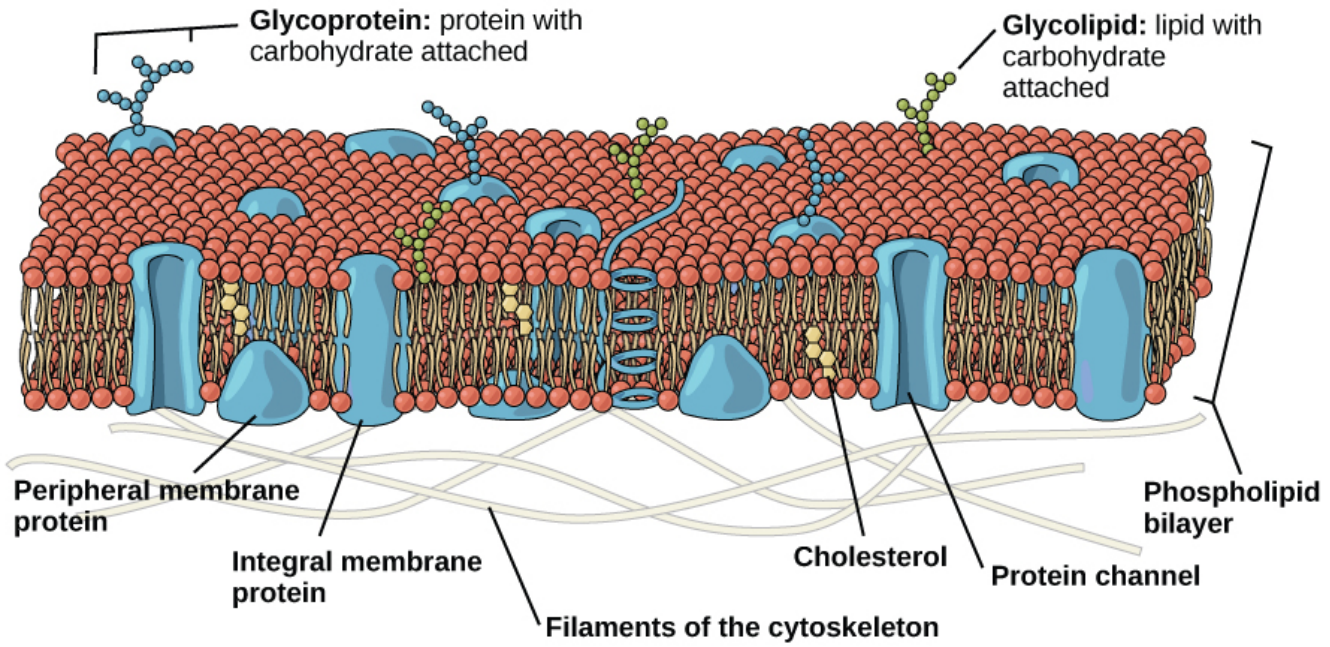

Figure 1. The eukaryotic plasma membrane is a phospholipid bilayer with proteins and cholesterol embedded in it.

Recall that plasma membranes are amphiphilic: they have hydrophilic and hydrophobic regions. This characteristic helps the movement of some materials through the membrane and hinders the movement of others. Lipid-soluble material with a low molecular weight can easily slip through the hydrophobic lipid core of the membrane. Substances such as the fat-soluble vitamins A, D, E, and K readily pass through the plasma membranes in the digestive tract and other tissues. Fat-soluble drugs and hormones also gain easy entry into cells and are readily transported into the body's tissues and organs. Molecules of oxygen and carbon dioxide have no charge and so pass through membranes by simple diffusion.

Polar substances present problems for the membrane. While some polar molecules connect easily with the outside of a cell, they cannot readily pass through the lipid core of the plasma membrane. Additionally, while small ions could easily slip through the spaces in the mosaic of the membrane, their charge prevents them from doing so. Ions such as sodium, potassium, calcium, and chloride must have special means of penetrating plasma membranes. Simple sugars and amino acids also need help with transport across plasma membranes, achieved by various transmembrane proteins (channels).

Diffusion

Diffusion is a passive process of transport (see Figure 2). A single substance tends to move from an area of high concentration to an area of low concentration until the concentration is equal across a space. You are familiar with diffusion of substances through the air.

For example, think about someone opening a bottle of ammonia in a room filled with people. The ammonia gas is at its highest concentration in the bottle; its lowest concentration is at the edges of the room. The ammonia vapor will diffuse, or spread away, from the bottle, and gradually, more and more people will smell the ammonia as it spreads. Materials move within the cell's cytosol by diffusion, and certain materials move through the plasma membrane by diffusion (Figure 3). Diffusion expends no energy. On the contrary, concentration gradients are a form of potential energy, dissipated as the gradient is eliminated.

Figure 2. Dispersion

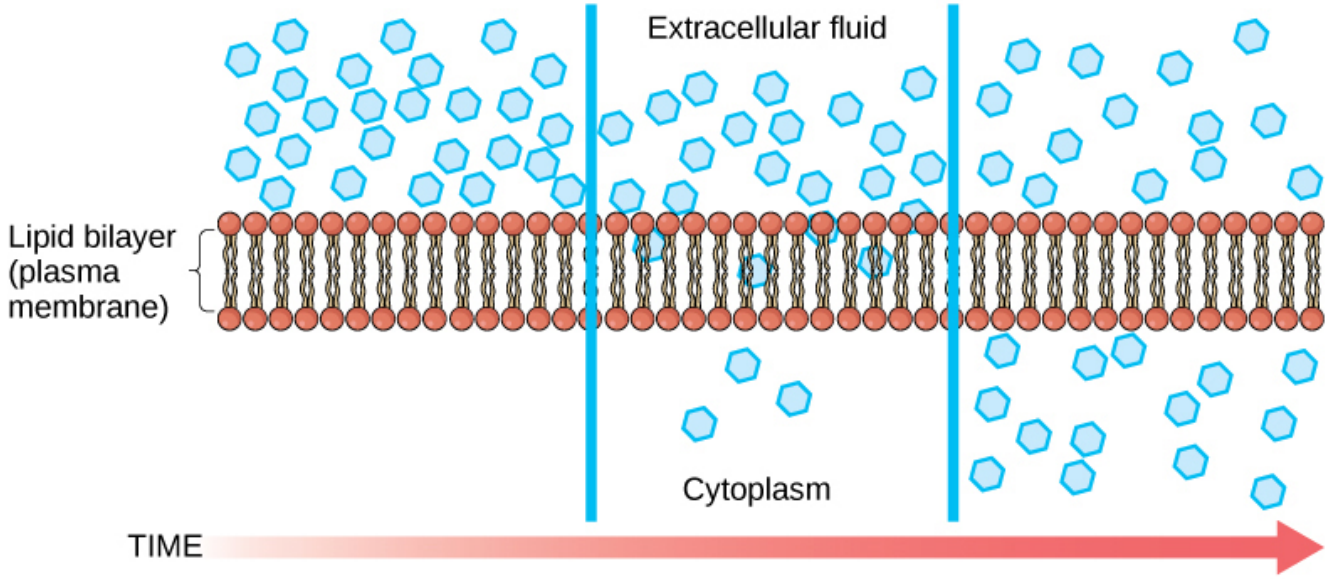

Figure 3. Diffusion through a permeable membrane moves a substance from an area of high concentration (extracellular fluid, in this case) down its concentration gradient (into the cytoplasm). (credit: modification of work by Mariana Ruiz Villareal)

Each separate substance in a medium, such as the extracellular fluid, has its own concentration gradient, independent of the concentration gradients of other materials. In addition, each substance will diffuse according to that gradient. Within a system, there will be different rates of diffusion of the different substances in the medium.

Facilitated Transport

In facilitated transport, also called facilitated diffusion, materials diffuse across the plasma membrane with the help of membrane proteins. A concentration gradient exists that would allow these materials to diffuse into the cell without expending cellular energy. However, these materials are ions or polar molecules that are repelled by the

hydrophobic parts of the cell membrane. Facilitated transport proteins shield these materials from the repulsive force of the membrane, allowing them to diffuse into the cell.

The material being transported is first attached to protein or glycoprotein receptors on the exterior surface of the plasma membrane. This allows the material that is needed by the cell to be removed from the extracellular fluid. The substances are then passed to specific integral proteins that facilitate their passage. Some of these integral proteins are collections of beta pleated sheets that form a pore or channel through the phospholipid bilayer. Others are carrier proteins which bind with the substance and aid its diffusion through the membrane.

Channels

The integral proteins involved in facilitated transport are collectively referred to as **transport proteins**, and they function as either channels for the material or carriers. In both cases, they are transmembrane proteins. Channels are specific for the substance that is being transported. **Channel proteins** have hydrophilic domains exposed to the intracellular and extracellular fluids; they additionally have a hydrophilic channel through their core that provides a hydrated opening through the membrane layers (Figure 4). Passage through the channel allows polar compounds to avoid the nonpolar central layer of the plasma membrane that would otherwise slow or prevent their entry into the cell. **Aquaporins** are channel proteins that allow water to pass through the membrane at a very high rate.

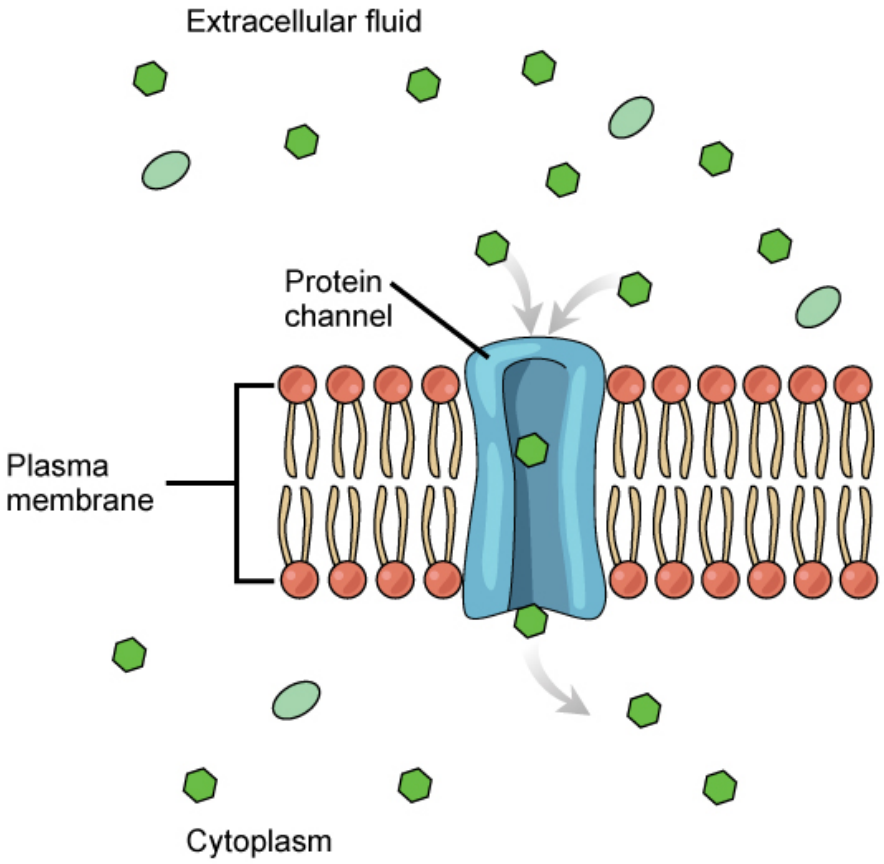

Figure 4. Facilitated transport moves substances down their concentration gradients. They may cross the plasma membrane with the aid of channel proteins. (credit: modification of work by Mariana Ruiz Villareal)

Channel proteins are either open at all times or they are “gated,” which controls the opening of the channel. The attachment of a particular ion to the channel protein may control the opening, or other mechanisms or substances may be involved. In some tissues, sodium and chloride ions pass freely through open channels, whereas in other tissues a gate must be opened to allow passage. An example of this occurs in the kidney, where both forms of channels are found in different parts of the renal tubules. Cells involved in the transmission of electrical impulses, such as nerve and muscle cells, have gated channels for sodium, potassium, and calcium in their membranes. Opening and closing of these channels changes the relative concentrations on opposing sides of the membrane of these ions, resulting in the facilitation of electrical transmission along membranes (in the case of nerve cells) or in muscle contraction (in the case of muscle cells).

Carrier Proteins

Another type of protein embedded in the plasma membrane is a **carrier protein**. This aptly named protein binds a substance and, in doing so, triggers a change of its own shape, moving the bound molecule from the outside of the cell to its interior (Figure 5); depending on the gradient, the material may move in the opposite direction. Carrier proteins are typically specific for a single substance. This selectivity adds to the overall selectivity of the

plasma membrane. The exact mechanism for the change of shape is poorly understood. Proteins can change shape when their hydrogen bonds are affected, but this may not fully explain this mechanism. Each carrier protein is specific to one substance, and there are a finite number of these proteins in any membrane. This can cause problems in transporting enough of the material for the cell to function properly. When all of the proteins are bound to their ligands, they are saturated and the rate of transport is at its maximum. Increasing the concentration gradient at this point will not result in an increased rate of transport.

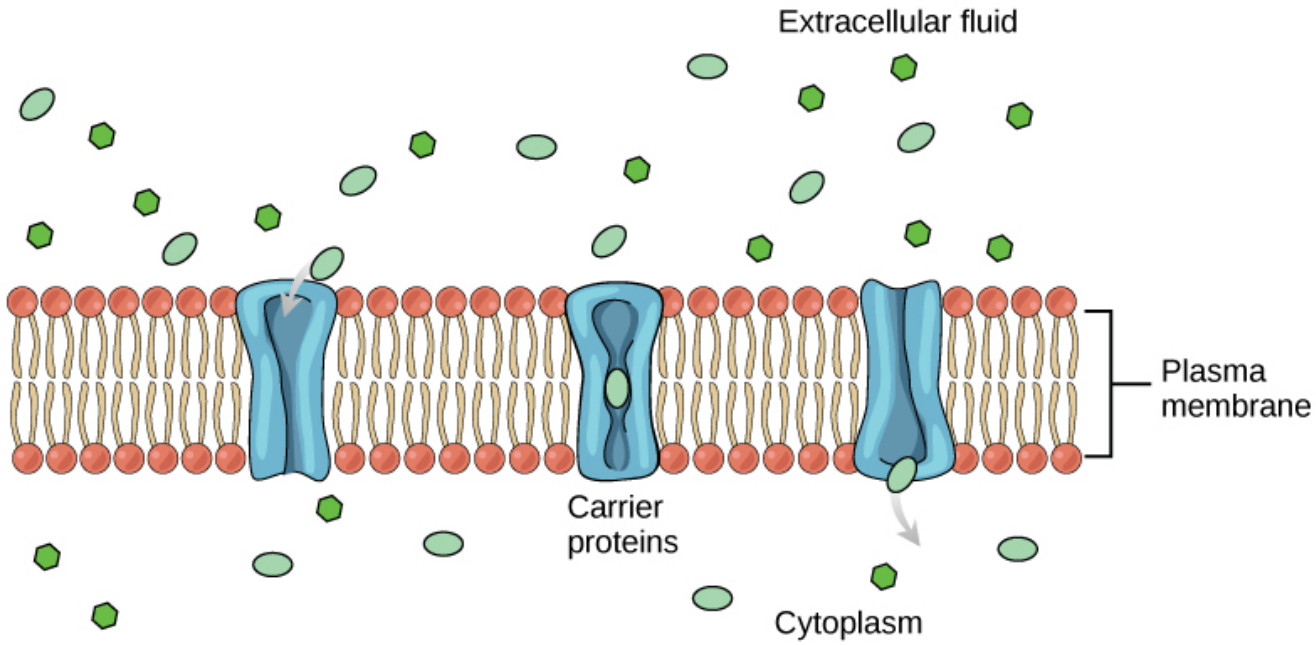

Figure 5. Some substances are able to move down their concentration gradient across the plasma membrane with the aid of carrier proteins. Carrier proteins change shape as they move molecules across the membrane. (credit: modification of work by Mariana Ruiz Villareal)

An example of this process occurs in the kidney. Glucose, water, salts, ions, and amino acids needed by the body are filtered in one part of the kidney. This filtrate, which includes glucose, is then reabsorbed in another part of the kidney. Because there are only a finite number of carrier proteins for glucose, if more glucose is present than the proteins can handle, the excess is not transported and it is excreted from the body in the urine. In a diabetic individual, this is described as “spilling glucose into the urine.” A different group of carrier proteins called glucose transport proteins, or GLUTs, are involved in transporting glucose and other hexose sugars through plasma membranes within the body.

Channel and carrier proteins transport material at different rates. Channel proteins transport much more quickly than do carrier proteins. Channel proteins facilitate diffusion at a rate of tens of millions of molecules per second, whereas carrier proteins work at a rate of a thousand to a million molecules per second.

Osmosis

Osmosis is the movement of water through a semipermeable membrane according to the concentration gradient of water across the membrane, which is inversely proportional to the concentration of solutes. While diffusion transports material across membranes and within cells, osmosis transports *only water* across a membrane and the membrane limits the diffusion of solutes in the water. Not surprisingly, the aquaporins that facilitate water movement play a large role in osmosis, most prominently in red blood cells and the membranes of kidney tubules.

Mechanism

Osmosis is a special case of diffusion. Water, like other substances, moves from an area of high concentration to one of low concentration. An obvious question is what makes water move at all? Imagine a beaker with a semipermeable membrane separating the two sides or halves (Figure 6). On both sides of the membrane the water level is the same, but there are different concentrations of a dissolved substance, or **solute**, that cannot cross the membrane (otherwise the concentrations on each side would be balanced by the solute crossing the membrane). If the volume of the solution on both sides of the membrane is the same, but the concentrations of solute are different, then there are different amounts of water, the solvent, on either side of the membrane.

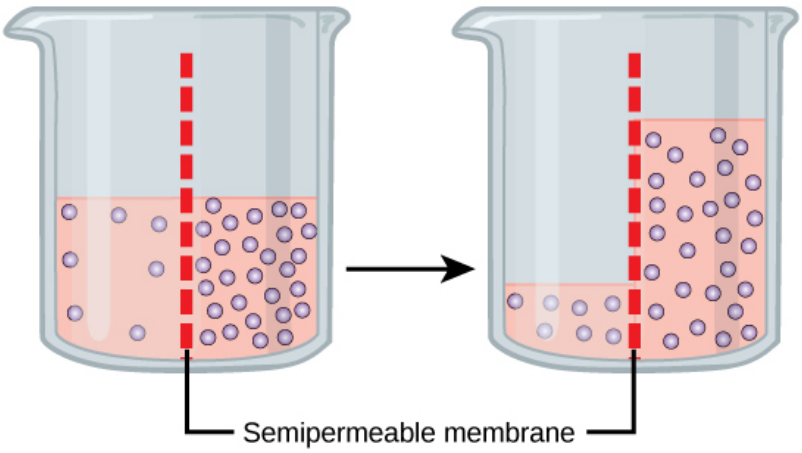

Figure 6. In osmosis, water always moves from an area of higher water concentration to one of lower concentration. In the diagram shown, the solute cannot pass through the selectively permeable membrane, but the water can.

To illustrate this, imagine two full glasses of water. One has a single teaspoon of sugar in it, whereas the second one contains one-quarter cup of sugar. If the total volume of the solutions in both cups is the same, which cup contains more water? Because the large amount of sugar in the second cup takes up much more space than the teaspoon of sugar in the first cup, the first cup has more water in it.

Returning to the beaker example, recall that it has a mixture of solutes on either side of the membrane. A principle of diffusion is that the molecules move around and will spread evenly throughout the medium if they can. However, only the material capable of getting through the membrane will diffuse through it. In this example, the solute cannot diffuse through the membrane, but the water can. Water has a concentration gradient in this system. Thus, water will diffuse down its concentration gradient, crossing the membrane to the side where it is less concentrated. This diffusion of water through the membrane—osmosis—will continue until the concentration gradient of water goes to zero or until the hydrostatic pressure of the water balances the osmotic pressure. Osmosis proceeds constantly in living systems.

Tonicity

Tonicity describes how an extracellular solution can change the volume of a cell by affecting osmosis. A solution's tonicity often directly correlates with the osmolarity of the solution. **Osmolarity** describes the total solute concentration of the solution. A solution with low osmolarity has a greater number of water molecules relative to the number of solute particles; a solution with high osmolarity has fewer water molecules with respect to solute particles. In a situation in which solutions of two different osmolarities are separated by a membrane permeable to water, though not to the solute, water will move from the side of the membrane with lower osmolarity (and more water) to the side with higher osmolarity (and less water). This effect makes sense if you remember that the solute cannot move across the membrane, and thus the only component in the system that can move—the water—moves along its own concentration gradient. An important distinction that concerns living systems is that osmolarity measures the number of particles (which may be molecules) in a solution. Therefore, a solution that is cloudy with cells may have a lower osmolarity than a solution that is clear, if the second solution contains more dissolved molecules than there are cells.

Hypotonic Solutions

Three terms—hypotonic, isotonic, and hypertonic—are used to relate the osmolarity of a cell to the osmolarity of the extracellular fluid that contains the cells. In a hypotonic situation, the extracellular fluid has lower osmolarity than the fluid inside the cell, and water enters the cell. (In living systems, the point of reference is always the

cytoplasm, so the prefix *hypo–* means that the extracellular fluid has a lower concentration of solutes, or a lower osmolarity, than the cell cytoplasm.) It also means that the extracellular fluid has a higher concentration of water in the solution than does the cell. In this situation, water will follow its concentration gradient and enter the cell.

Hypertonic Solutions

As for a hypertonic solution, the prefix *hyper–* refers to the extracellular fluid having a higher osmolarity than the cell's cytoplasm; therefore, the fluid contains less water than the cell does. Because the cell has a relatively higher concentration of water, water will leave the cell.

Isotonic Solutions

In an isotonic solution, the extracellular fluid has the same osmolarity as the cell. If the osmolarity of the cell matches that of the extracellular fluid, there will be no net movement of water into or out of the cell, although water will still move in and out. Blood cells and plant cells (Figure 7) in hypertonic, isotonic, and hypotonic solutions take on characteristic appearances.

Practice Question

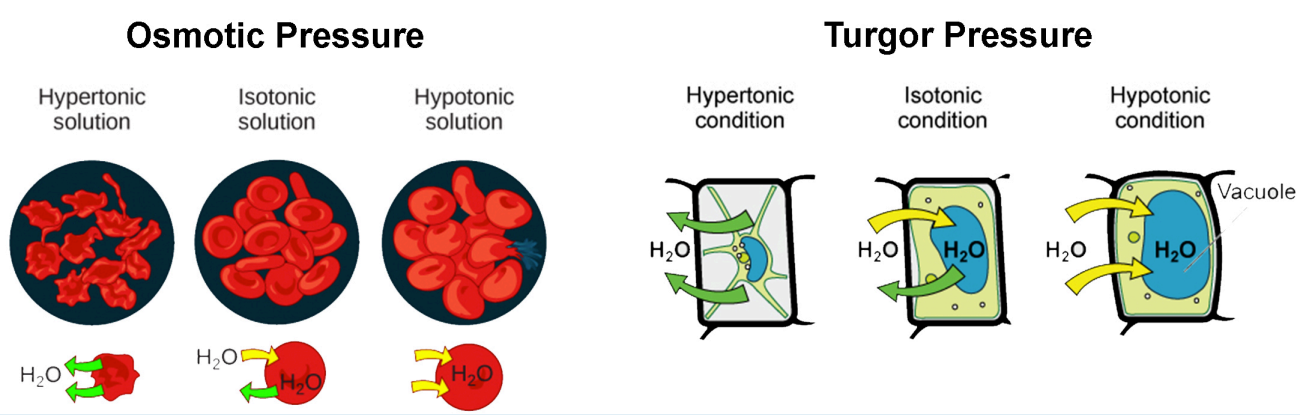

Figure 7. Osmotic pressure changes the shape of red blood cells in hypertonic, isotonic, and hypotonic solutions. The turgor pressure within a plant cell depends on the tonicity of the solution that it is bathed in. (credit: modification of work by Mariana Ruiz Villareal)

A doctor injects a patient with what the doctor thinks is an isotonic saline solution. The patient dies, and an autopsy reveals that many red blood cells have been destroyed. Do you think the solution the doctor injected was really isotonic?

Answer

No, it must have been hypotonic as a hypotonic solution would cause water to enter the cells, thereby making them burst.

Video Review

Watch this review of osmosis and diffusion

Watch this video online: https://youtu.be/aubZU0iWtgI

In Summary: Passive Transport

No energy is required. The "driving force" is a difference in the concentration of a substance on one side of the membrane compared that on the other side.

- Simple diffusion (O_2, CO_2, H_20. Water and nonpolar molecules).
  - Osmosis is a special kind of simple diffusion for water only.
- Familiarize yourself with the terms *hypotonic*, *isotonic*, and *hypertonic*, and be able to indicate what either a plant or an animal cell will look like if placed in a particular kind of solution.
  - Example: a plant cell has a cell wall and is full and happy when placed in water (a hypotonic solution). An animal cell does not have a cell wall and will swell and burst if placed in water. This is why a patient should never receive an IV injection of water: it will cause their red blood cells to burst.
- Facilitated diffusion (sugars, ions, amino acids, etc. Charged or polar molecules).
  - Carrier proteins
  - Channel proteins (such as ion channels or aquaporin)

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ACTIVE TRANSPORT

Learning Outcomes

Define and describe active transport

Active transport mechanisms require the use of the cell's energy, usually in the form of adenosine triphosphate (ATP). If a substance must move into the cell against its concentration gradient—that is, if the concentration of the substance inside the cell is greater than its concentration in the extracellular fluid (and vice versa)—the cell must use energy to move the substance. Some active transport mechanisms move small-molecular weight materials, such as ions, through the membrane. Other mechanisms transport much larger molecules.

Electrochemical Gradient

We have discussed simple concentration gradients—differential concentrations of a substance across a space or a membrane—but in living systems, gradients are more complex. Because ions move into and out of cells and because cells contain proteins that do not move across the membrane and are mostly negatively charged, there is also an electrical gradient, a difference of charge, across the plasma membrane. The interior of living cells is electrically negative with respect to the extracellular fluid in which they are bathed, and at the same time, cells have higher concentrations of potassium (K^+) and lower concentrations of sodium (Na^+) than does the extracellular fluid. So in a living cell, the concentration gradient of Na^+ tends to drive it into the cell, and the

electrical gradient of Na^+ (a positive ion) also tends to drive it inward to the negatively charged interior. The situation is more complex, however, for other elements such as potassium. The electrical gradient of K^+, a positive ion, also tends to drive it into the cell, but the concentration gradient of K^+ tends to drive K^+ *out* of the cell (Figure 1). The combined gradient of concentration and electrical charge that affects an ion is called its **electrochemical gradient**.

Practice Question

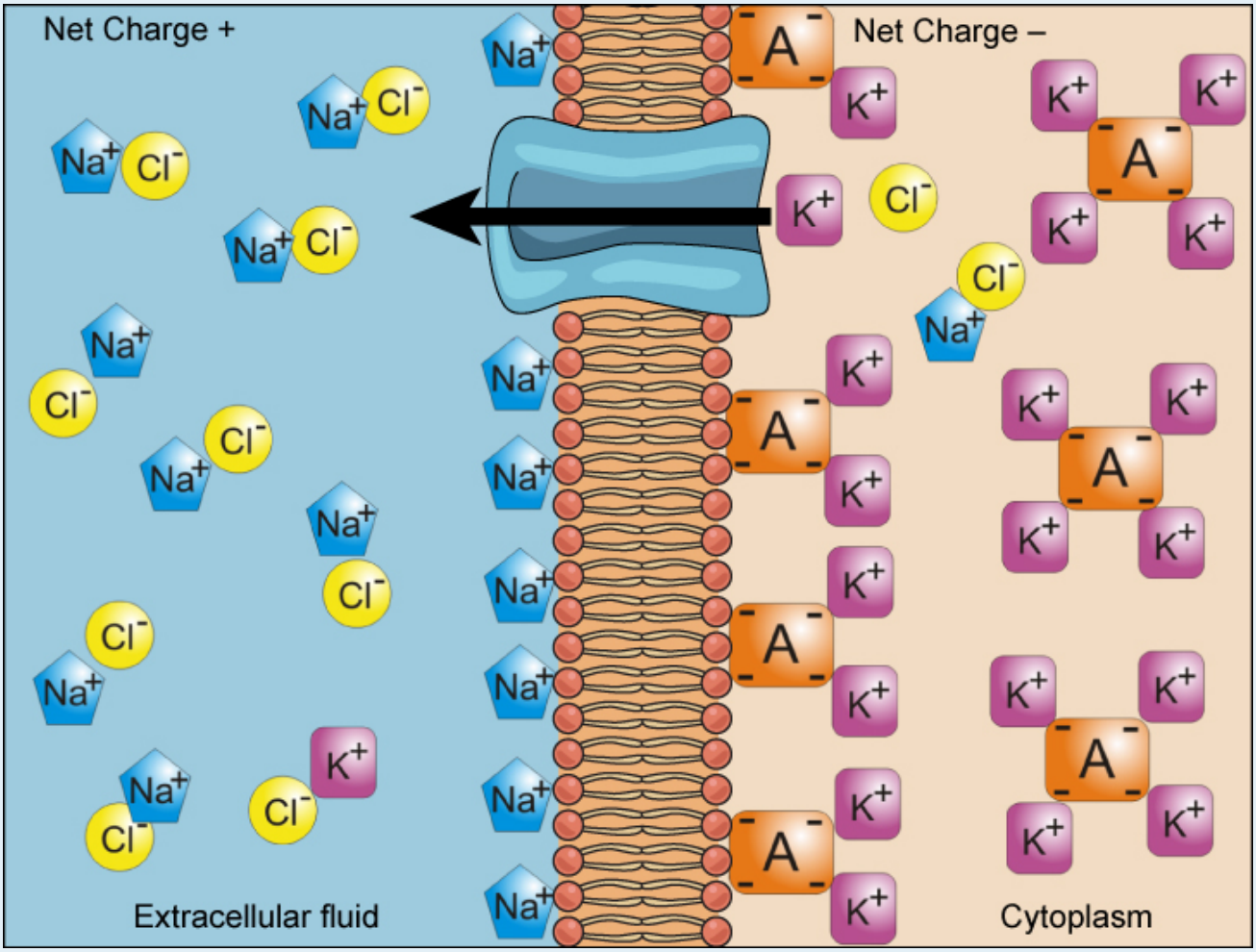

Figure 1. Electrochemical gradients arise from the combined effects of concentration gradients and electrical gradients. (credit: "Synaptitude"/Wikimedia Commons)

Injection of a potassium solution into a person's blood is lethal; this is used in capital punishment and euthanasia. Why do you think a potassium solution injection is lethal?

Answer

Cells typically have a high concentration of potassium in the cytoplasm and are bathed in a high concentration of sodium. Injection of potassium dissipates this electrochemical gradient. In heart muscle, the sodium/potassium potential is responsible for transmitting the signal that causes the muscle to contract. When this potential is dissipated, the signal can't be transmitted, and the heart stops beating. Potassium injections are also used to stop the heart from beating during surgery.

Moving Against a Gradient

To move substances against a concentration or electrochemical gradient, the cell must use energy. This energy is harvested from ATP generated through the cell's metabolism. Active transport mechanisms, collectively called **pumps**, work against electrochemical gradients. Small substances constantly pass through plasma membranes. Active transport maintains concentrations of ions and other substances needed by living cells in the face of these passive movements. Much of a cell's supply of metabolic energy may be spent maintaining these processes. (Most of a red blood cell's metabolic energy is used to maintain the imbalance between exterior and interior sodium and potassium levels required by the cell.) Because active transport mechanisms depend on a cell's metabolism for energy, they are sensitive to many metabolic poisons that interfere with the supply of ATP.

Two mechanisms exist for the transport of small-molecular weight material and small molecules. **Primary active transport** moves ions across a membrane and creates a difference in charge across that membrane, which is directly dependent on ATP. **Secondary active transport** describes the movement of material that is due to the electrochemical gradient established by primary active transport that does not directly require ATP.

Carrier Proteins for Active Transport

An important membrane adaption for active transport is the presence of specific carrier proteins or pumps to facilitate movement: there are three types of these proteins or **transporters** (Figure 2). A **uniporter** carries one specific ion or molecule. A **symporter** carries two different ions or molecules, both in the same direction. An **antiporter** also carries two different ions or molecules, but in different directions. All of these transporters can also transport small, uncharged organic molecules like glucose. These three types of carrier proteins are also found in facilitated diffusion, but they do not require ATP to work in that process. Some examples of pumps for active transport are $Na^{+-}K^{+}$ ATPase, which carries sodium and potassium ions, and $H^{+-}K^{+}$ ATPase, which carries hydrogen and potassium ions. Both of these are antiporter carrier proteins. Two other carrier proteins are $Ca2^{+}$ ATPase and H^{+} ATPase, which carry only calcium and only hydrogen ions, respectively. Both are pumps.

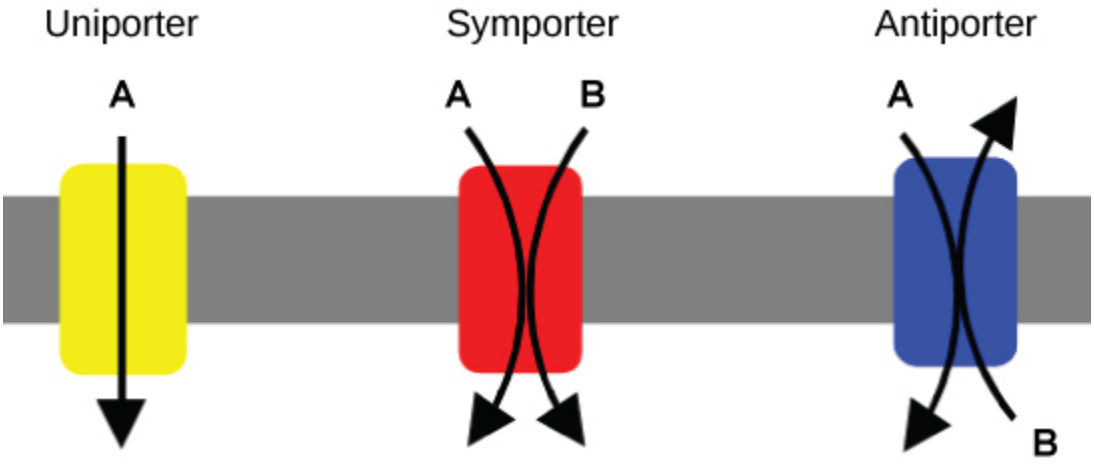

Figure 2. A uniporter carries one molecule or ion. A symporter carries two different molecules or ions, both in the same direction. An antiporter also carries two different molecules or ions, but in different directions. (credit: modification of work by "Lupask"/Wikimedia Commons)

Primary Active Transport

The primary active transport that functions with the active transport of sodium and potassium allows secondary active transport to occur. The second transport method is still considered active because it depends on the use of energy as does primary transport (Figure 3).

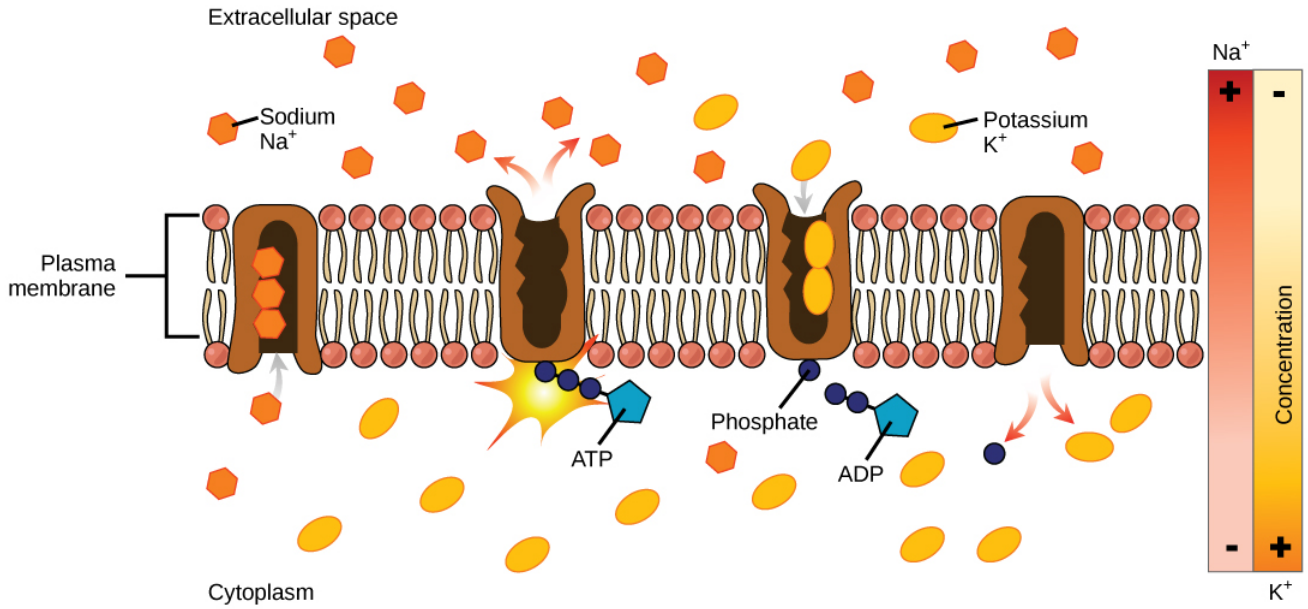

Figure 3. Primary active transport moves ions across a membrane, creating an electrochemical gradient (electrogenic transport). (credit: modification of work by Mariana Ruiz Villareal)

One of the most important pumps in animals cells is the sodium-potassium pump (Na^+-K^+ ATPase), which maintains the electrochemical gradient (and the correct concentrations of Na^+ and K^+) in living cells. The sodium-potassium pump moves K^+ into the cell while moving Na+ out at the same time, at a ratio of three Na^+ for every two K^+ ions moved in. Several things have happened as a result of this process. At this point, there are more sodium ions outside of the cell than inside and more potassium ions inside than out. For every three ions of sodium that move out, two ions of potassium move in. This results in the interior being slightly more negative relative to the exterior. This difference in charge is important in creating the conditions necessary for the secondary process. The sodium-potassium pump is, therefore, an electrogenic pump (a pump that creates a charge imbalance), creating an electrical imbalance across the membrane and contributing to the membrane potential.

Secondary Active Transport (Co-transport)

Secondary active transport brings sodium ions, and possibly other compounds, into the cell. As sodium ion concentrations build outside of the plasma membrane because of the action of the primary active transport process, an electrochemical gradient is created. If a channel protein exists and is open, the sodium ions will be pulled through the membrane. This movement is used to transport other substances that can attach themselves to the transport protein through the membrane (Figure 4). Many amino acids, as well as glucose, enter a cell this way. This secondary process is also used to store high-energy hydrogen ions in the mitochondria of plant and animal cells for the production of ATP. The potential energy that accumulates in the stored hydrogen ions is translated into kinetic energy as the ions surge through the channel protein ATP synthase, and that energy is used to convert ADP into ATP.

Practice Question

An electrochemical gradient, created by primary active transport, can move other substances against their concentration gradients, a process called co-transport or secondary active transport.

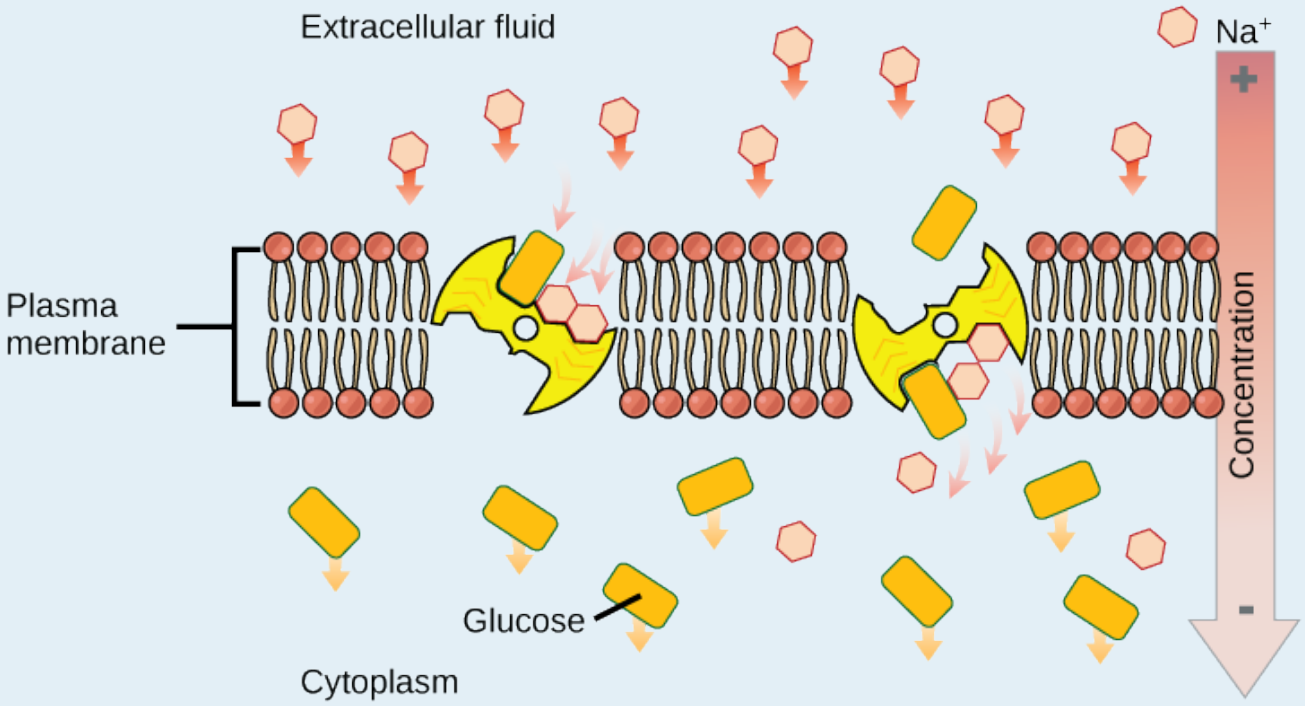

Figure 4. (credit: modification of work by Mariana Ruiz Villareal)

Answer

A decrease in pH means an increase in positively charged H^+ ions, and an increase in the electrical gradient across the membrane. The transport of amino acids into the cell will increase.

In Summary: Active Transport

Energy is required.

- Primary active transport (ATP is the “driving force”).
- Secondary active transport (the energy is provided by an electrochemical gradient).

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MEMBRANES AND TRANSPORT

Learning Outcomes

- Define and describe passive transport
- Define and describe active transport

Now that we've learned about active and passive transport separately, let's review both topics together. In addition to reviewing the ways transport works across membranes, this video will discuss the reasons cells must be selectively permeable.

Watch this video online: https://youtu.be/dPKvHrD1eS4

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INTRODUCTION TO ENDOCYTOSIS AND EXOCYTOSIS

What you'll learn to do: Describe the primary mechanisms by which cells import and export macromolecules

In addition to moving small ions and molecules through the membrane, cells also need to remove and take in larger molecules and particles. Some cells are even capable of engulfing entire unicellular microorganisms. You might have correctly hypothesized that the uptake and release of large particles by the cell requires energy. A large particle, however, cannot pass through the membrane, even with energy supplied by the cell. Instead, cells use one of two primary mechanisms that transport these large particles: endocytosis and exocytosis.

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ENDOCYTOSIS

Learning Outcomes

Describe endocytosis and identify different varieties of import, including phagocytosis, pinocytosis, and receptor-mediated endocytosis

Endocytosis is a type of active transport that moves particles, such as large molecules, parts of cells, and even whole cells, into a cell. There are different variations of endocytosis, but all share a common characteristic: the plasma membrane of the cell invaginates, forming a pocket around the target particle. The pocket pinches off, resulting in the particle being contained in a newly created intracellular vesicle formed from the plasma membrane.

Phagocytosis

Phagocytosis (the condition of "cell eating") is the process by which large particles, such as cells or relatively large particles, are taken in by a cell. For example, when microorganisms invade the human body, a type of white blood cell called a neutrophil will remove the invaders through this process, surrounding and engulfing the microorganism, which is then destroyed by the neutrophil (Figure 1).

In preparation for phagocytosis, a portion of the inward-facing surface of the plasma membrane becomes coated with a protein called *clathrin*, which stabilizes this section of the membrane. The coated portion of the membrane then extends from the body of the cell and surrounds the particle, eventually enclosing it. Once the vesicle containing the particle is enclosed within the cell, the clathrin disengages from the membrane and the vesicle merges with a lysosome for the breakdown of the material in the newly formed compartment (endosome). When accessible nutrients from the degradation of the vesicular contents have been extracted, the newly formed endosome merges with the plasma membrane and releases its contents into the extracellular fluid. The endosomal membrane again becomes part of the plasma membrane.

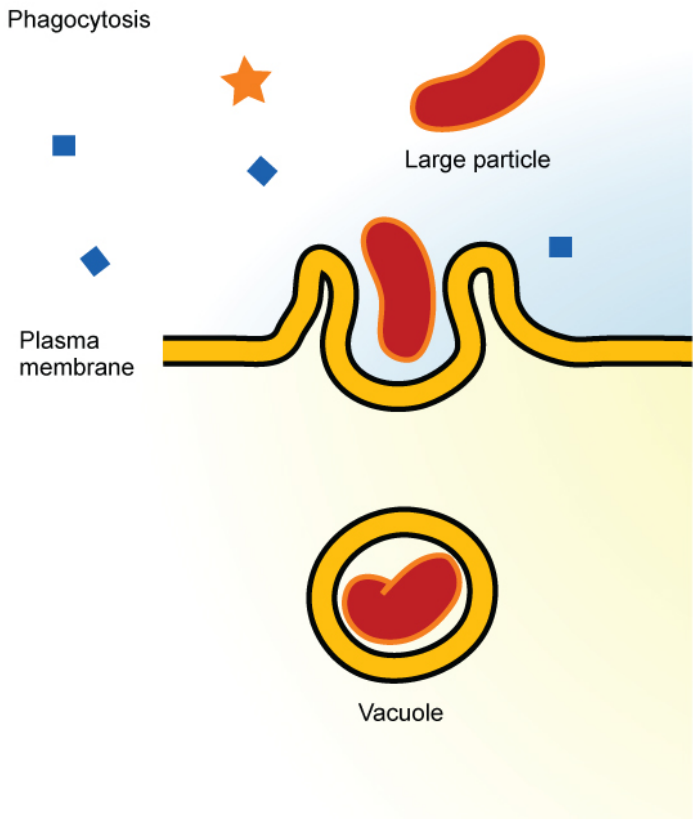

Figure 1. In phagocytosis, the cell membrane surrounds the particle and engulfs it. (credit: Mariana Ruiz Villareal)

Pinocytosis

A variation of endocytosis is called pinocytosis. This literally means “cell drinking” and was named at a time when the assumption was that the cell was purposefully taking in extracellular fluid. In reality, this is a process that takes in molecules, including water, which the cell needs from the extracellular fluid. Pinocytosis results in a much smaller vesicle than does phagocytosis, and the vesicle does not need to merge with a lysosome (Figure 2).

A variation of pinocytosis is called potocytosis. This process uses a coating protein, called caveolin, on the cytoplasmic side of the plasma membrane, which performs a similar function to clathrin. The cavities in the plasma membrane that form the vacuoles have membrane receptors and lipid rafts in addition to caveolin.

The vacuoles or vesicles formed in caveolae (singular caveola) are smaller than those in pinocytosis. Potocytosis is used to bring small molecules into the cell and to transport these molecules through the cell for their release on the other side of the cell, a process called transcytosis.

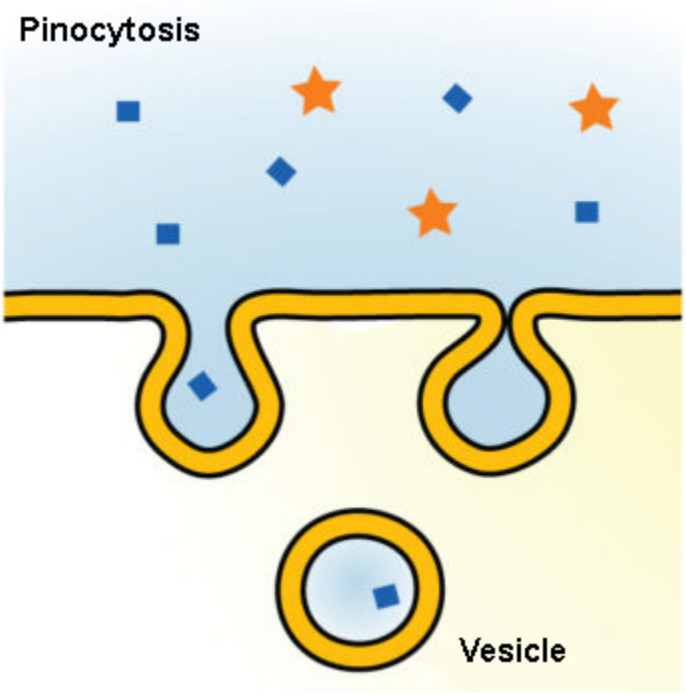

Figure 2. In pinocytosis, the cell membrane invaginates, surrounds a small volume of fluid, and pinches off. (credit: Mariana Ruiz Villareal)

Receptor-Mediated Endocytosis

A targeted variation of endocytosis employs receptor proteins in the plasma membrane that have a specific binding affinity for certain substances (Figure 3).

In receptor-mediated endocytosis, as in phagocytosis, clathrin is attached to the cytoplasmic side of the plasma membrane. If uptake of a compound is dependent on receptor-mediated endocytosis and the process is ineffective, the material will not be removed from the tissue fluids or blood. Instead, it will stay in those fluids and increase in concentration.

Some human diseases are caused by the failure of receptor-mediated endocytosis. For example, the form of cholesterol termed low-density lipoprotein or LDL (also referred to as “bad” cholesterol) is removed from the blood by receptor-mediated endocytosis. In the human genetic disease familial hypercholesterolemia, the LDL receptors are defective or missing entirely. People with this condition have life-threatening levels of cholesterol in their blood, because their cells cannot clear LDL particles from their blood.

Although receptor-mediated endocytosis is designed to bring specific substances that are normally found in the extracellular fluid into the cell, other substances may gain entry into the cell at the same site. Flu viruses, diphtheria, and cholera toxin all have sites that cross-react with normal receptor-binding sites and gain entry into cells.

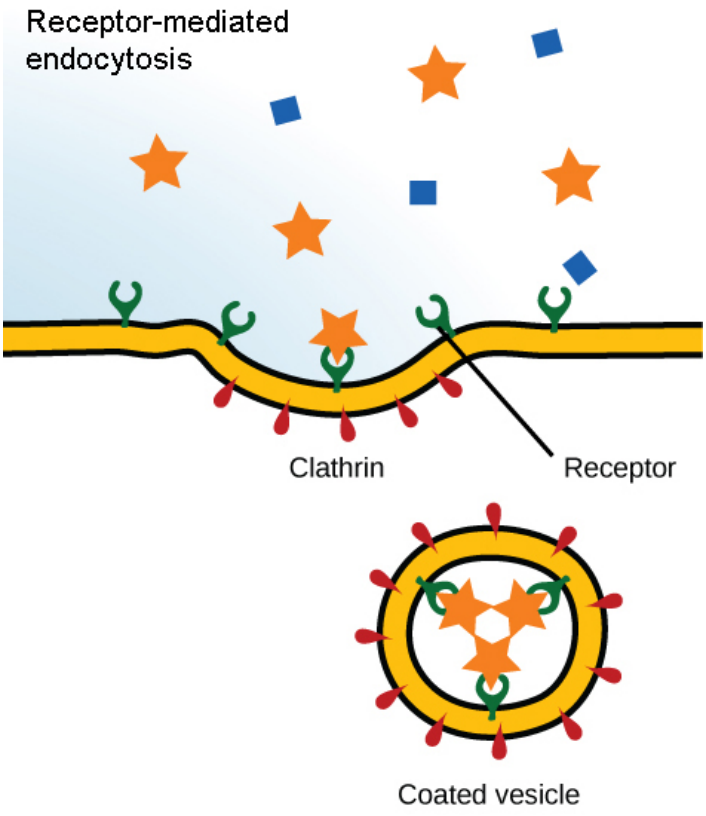

Figure 3. In receptor-mediated endocytosis, uptake of substances by the cell is targeted to a single type of substance that binds to the receptor on the external surface of the cell membrane. (credit: modification of work by Mariana Ruiz Villareal)

In Summary: Endocytosis

Cells perform three main types of endocytosis. Phagocytosis is the process by which cells ingest large particles, including other cells, by enclosing the particles in an extension of the cell membrane and budding off a new vacuole. During pinocytosis, cells take in molecules such as water from the extracellular fluid. Finally, receptor-mediated endocytosis is a targeted version of endocytosis where receptor proteins in the plasma membrane ensure only specific, targeted substances are brought into the cell.

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EXOCYTOSIS

Learning Outcomes

Identify the steps of exocytosis

The reverse process of moving material into a cell is the process of exocytosis. Exocytosis is the opposite of the processes discussed in the last section in that its purpose is to expel material from the cell into the extracellular fluid. Waste material is enveloped in a membrane and fuses with the interior of the plasma membrane. This fusion opens the membranous envelope on the exterior of the cell, and the waste material is expelled into the extracellular space (Figure 1). Other examples of cells releasing molecules via exocytosis include the secretion of proteins of the extracellular matrix and secretion of neurotransmitters into the synaptic cleft by synaptic vesicles.

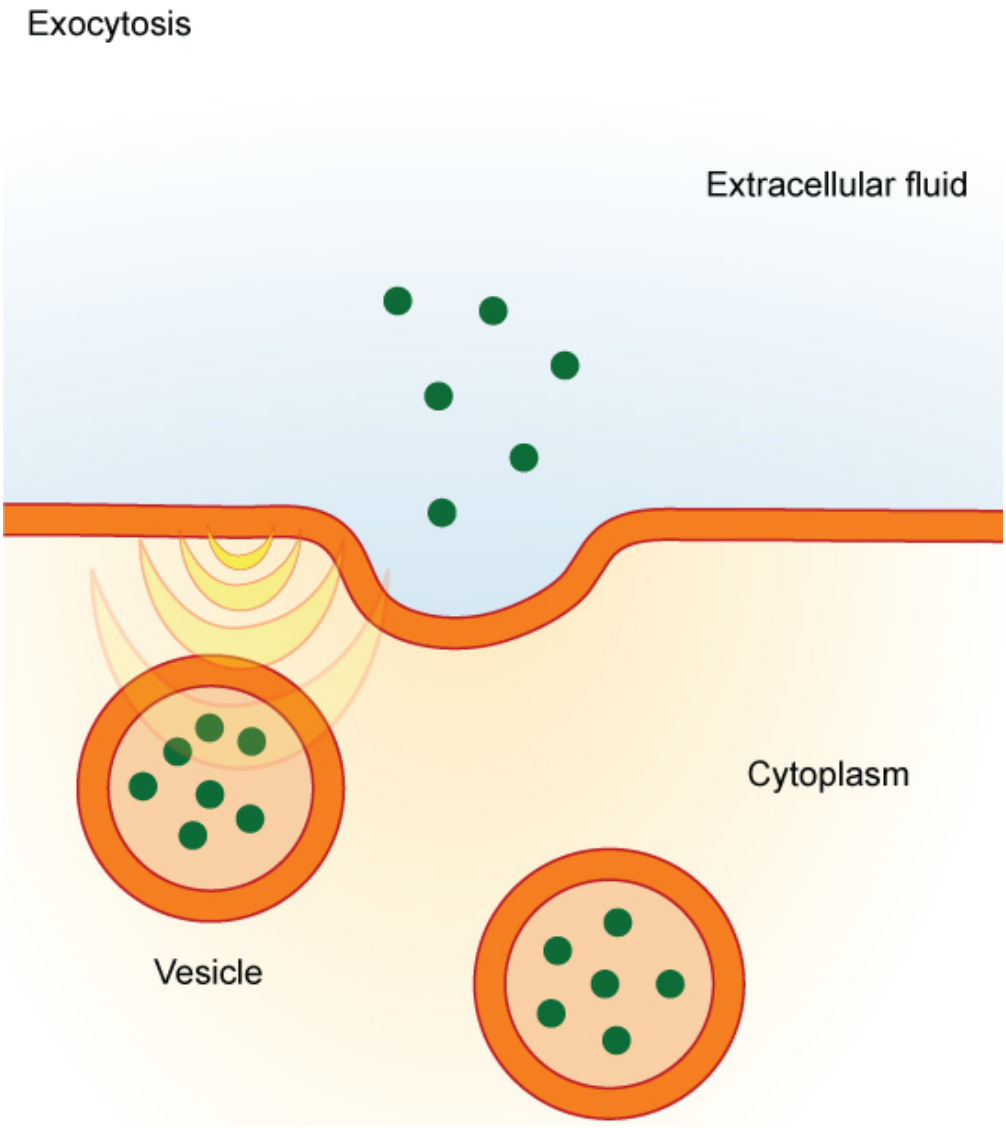

Figure 1. In exocytosis, vesicles containing substances fuse with the plasma membrane. The contents are then released to the exterior of the cell. (credit: modification of work by Mariana Ruiz Villareal)

A summary of the cellular transport methods discussed is contained in Table 1, which also includes the energy requirements and materials transported by each.

Table 1. Methods of Transport, Energy Requirements, and Types of Material Transported

Transport Method	Active/ Passive	Material Transported
Diffusion	Passive	Small-molecular weight material
Osmosis	Passive	Water
Facilitated transport/ diffusion	Passive	Sodium, potassium, calcium, glucose
Primary active transport	Active	Sodium, potassium, calcium
Secondary active transport	Active	Amino acids, lactose
Phagocytosis	Active	Large macromolecules, whole cells, or cellular structures
Pinocytosis	Active	Small molecules (liquids/water)
Receptor-mediated endocytosis	Active	Large quantities of macromolecules
Exocytosis	Active	Waste materials, proteins for the extracellular matrix, neurotransmitters

In Summary: Exocytosis

Exocytosis in many ways is the reverse process from endocytosis. Here cells expel material through the fusion of vesicles with the plasma membrane and subsequent dumping of their content into the extracellular fluid.

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PUTTING IT TOGETHER: CELL MEMBRANES

Let's return to our discussion of cystic fibrosis. Cystic fibrosis (CF) is caused by a defect in a single transmembrane protein: cystic fibrosis transmembrane conductance regulator (CFTR), as seen in Figure 1.

This regulator is a chloride ion channel that crosses through the plasma membrane. This channel is specifically active in epithelial tissues where it normally helps produce thin mucus via the movement of water. When mutated, the channel does not work properly and mucus becomes thick and viscous. This in turn leads directly to many of the symptoms of CF: thick, sticky mucus, frequent chest infections, and coughing or shortness of breath.

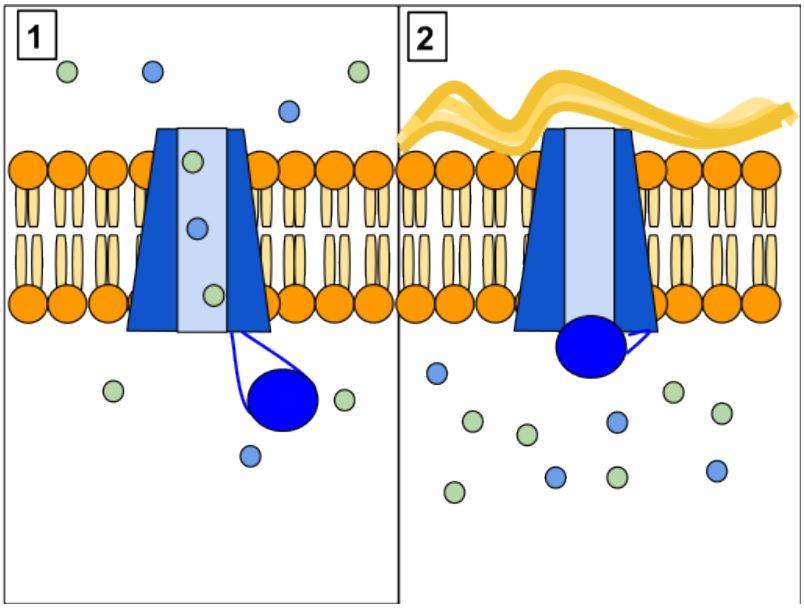

Figure 1. Panel 1 shows a properly working CFTR protein. Panel 2 shows a malfunctioning CFTR protein.

Treatment

Cystic fibrosis is a difficult disease to treat. As we mentioned at the beginning of the chapter, patients with CF often suffer lung infections and sometimes require lung transplants. In addition to this, many CF patients are on one or more antibiotics at all times—even when healthy—to suppress infection. Several mechanical techniques are used to dislodge sputum and encourage its expectoration. In the hospital setting, chest physiotherapy is utilized. As lung disease worsens, mechanical breathing support may become necessary. Bi-lateral lung transplantation often becomes necessary for individuals with cystic fibrosis as lung function and exercise tolerance declines.

Gene therapy has been explored as a potential cure for cystic fibrosis. Ideally, gene therapy attempts to place a normal copy of the CFTR gene into affected cells. Transferring the normal CFTR gene into the affected epithelium cells would result in the production of functional CFTR in all target cells, without adverse reactions or an inflammation response. Studies have shown that to prevent the lung manifestations of cystic fibrosis, only 5–10 percent the normal amount of CFTR gene expression is needed.

Finally, a number of small molecules that aim at compensating various mutations of the CFTR gene are under development. About 10 percent of CF cases result from a premature stop codon in the DNA, leading to early termination of protein synthesis and truncated proteins. One approach to combating a faulty receptor is to develop drugs that get the ribosome to overcome this premature stop codon and synthesize a full-length CFTR protein.

Learn More

- Cystic Fibrosis Foundation
- Cystic Fibrosis on the Mayo Clinic

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MODULE 6: METABOLIC PATHWAYS

WHY IT MATTERS: METABOLIC PATHWAYS

Why explain the metabolic pathways involved in the capture and release of energy in cells?

Every time you move—or even breathe—you're using energy. All living things are continually using energy; thus, they need a way to create or obtain new energy. Two of these ways are photosynthesis and cellular respiration.

Plants (and other autotrophs) undergo photosynthesis to create energy. Humans (and other heterotrophs) on the other hand must consume something that has energy (like plants or other animals)—we take this energy and convert it into a form our body can use. This process is known as cellular respiration.

Watch this 5 minute video for an overview of why even small changes in the global climate have the potential for big impacts on our daily lives through our food sources.

Watch this video online: https://youtu.be/cBqr43QbfBE

- What role does farming play in giving us energy to use every day?
- How do plants get energy to grow, and how do we then get our energy from them?

Ok, let's see where we get all the energy to stay awake during biology class!

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INTRODUCTION TO ENERGY AND METABOLISM

What you'll learn to do: Discuss energy and metabolism in living things

Scientists use the term bioenergetics to describe the concept of energy flow (Figure 1) through living systems, such as cells. Cellular processes such as the building and breaking down of complex molecules occur through stepwise chemical reactions. Some of these chemical reactions are spontaneous and release energy, whereas others require energy to proceed.

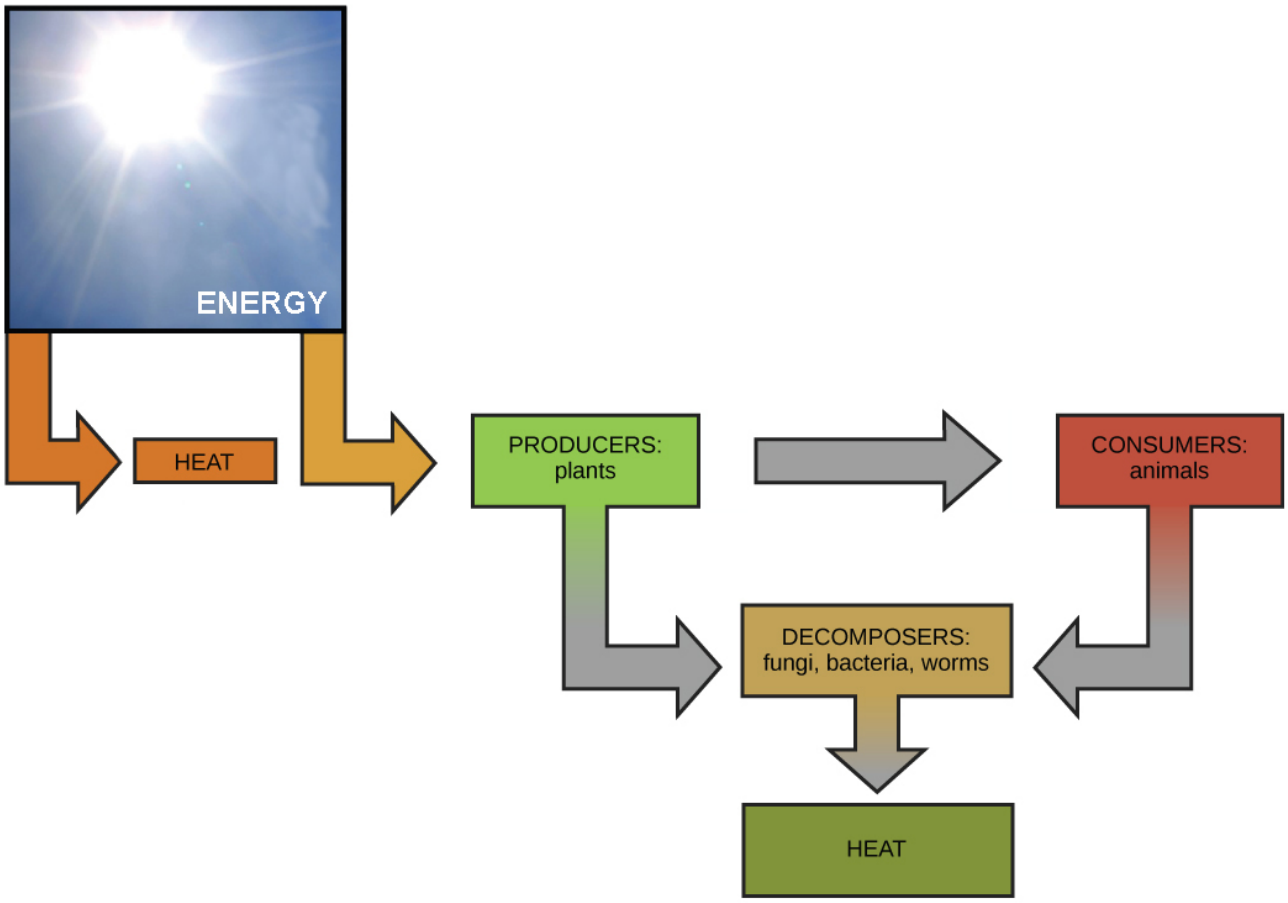

Figure 1. Ultimately, most life forms get their energy from the sun. Plants use photosynthesis to capture sunlight, and herbivores eat the plants to obtain energy. Carnivores eat the herbivores, and eventual decomposition of plant and animal material contributes to the nutrient pool.

Just as living things must continually consume food to replenish their energy supplies, cells must continually produce more energy to replenish that used by the many energy-requiring chemical reactions that constantly take place. Together, all of the chemical reactions that take place inside cells, including those that consume or generate energy, are referred to as the cell's metabolism.

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METABOLIC PATHWAYS

Learning Outcomes

Identify different types of metabolic pathways

Consider the metabolism of sugar. This is a classic example of one of the many cellular processes that use and produce energy. Living things consume sugars as a major energy source, because sugar molecules have a great

deal of energy stored within their bonds. For the most part, photosynthesizing organisms like plants produce these sugars. During photosynthesis, plants use energy (originally from sunlight) to convert carbon dioxide gas (CO_2) into sugar molecules (like glucose: $C_6H_{12}O_6$). They consume carbon dioxide and produce oxygen as a waste product. This reaction is summarized as:

$$6CO_2 + 6H_2O \rightarrow C_6H_{12}O_6 + 6O_2$$

Because this process involves synthesizing an energy-storing molecule, it requires energy input to proceed. During the light reactions of photosynthesis, energy is provided by a molecule called adenosine triphosphate (ATP), which is the primary energy currency of all cells. Just as the dollar is used as currency to buy goods, cells use molecules of ATP as energy currency to perform immediate work. In contrast, energy-storage molecules such as glucose are consumed only to be broken down to use their energy. The reaction that harvests the energy of a sugar molecule in cells requiring oxygen to survive can be summarized by the reverse reaction to photosynthesis. In this reaction, oxygen is consumed and carbon dioxide is released as a waste product. The reaction is summarized as:

$$C_6H_{12}O_6 \rightarrow 6H_2O + 6CO_2$$

Both of these reactions involve many steps.

The processes of making and breaking down sugar molecules illustrate two examples of metabolic pathways. A metabolic pathway is a series of chemical reactions that takes a starting molecule and modifies it, step-by-step, through a series of metabolic intermediates, eventually yielding a final product. In the example of sugar metabolism, the first metabolic pathway synthesized sugar from smaller molecules, and the other pathway broke sugar down into smaller molecules. These two opposite processes—the first requiring energy and the second producing energy—are referred to as anabolic pathways (building polymers) and catabolic pathways (breaking down polymers into their monomers), respectively. Consequently, metabolism is composed of synthesis (anabolism) and degradation (catabolism) (Figure 1).

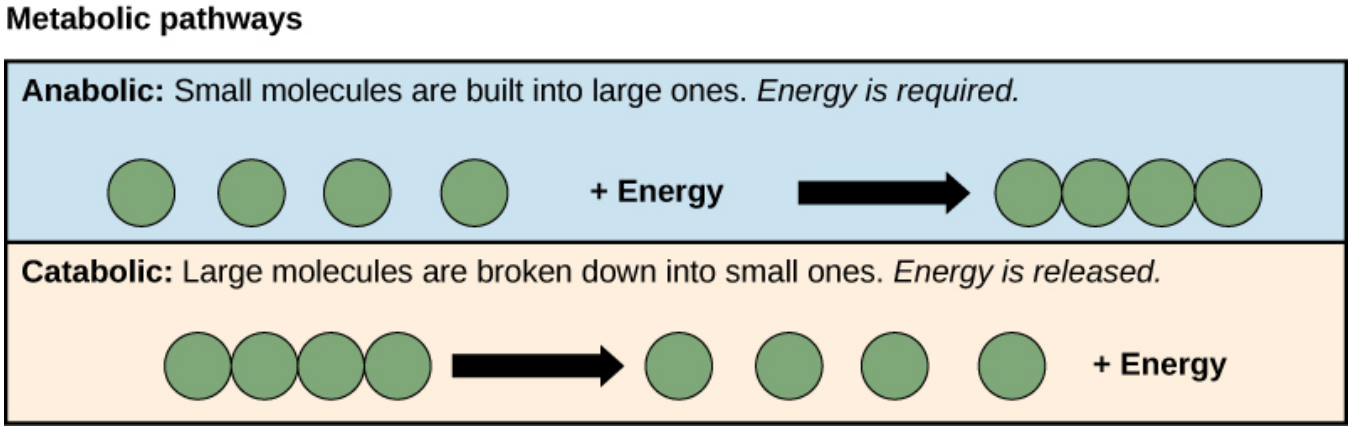

Figure 1. Catabolic pathways are those that generate energy by breaking down larger molecules. Anabolic pathways are those that require energy to synthesize larger molecules. Both types of pathways are required for maintaining the cell's energy balance.

It is important to know that the chemical reactions of metabolic pathways do not take place on their own. Each reaction step is facilitated, or catalyzed, by a protein called an enzyme. Enzymes are important for catalyzing all types of biological reactions—those that require energy as well as those that release energy.

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THERMODYNAMICS

Learning Outcomes

- Distinguish between an open and a closed system
- State the first law of thermodynamics
- State the second law of thermodynamics

Thermodynamics refers to the study of energy and energy transfer involving physical matter. The matter relevant to a particular case of energy transfer is called a system, and everything outside of that matter is called the surroundings. For instance, when heating a pot of water on the stove, the system includes the stove, the pot, and the water. Energy is transferred within the system (between the stove, pot, and water). There are two types of systems: open and closed. In an open system, energy can be exchanged with its surroundings. The stovetop system is open because heat can be lost to the air. A closed system cannot exchange energy with its surroundings.

Biological organisms are open systems. Energy is exchanged between them and their surroundings as they use energy from the sun to perform photosynthesis or consume energy-storing molecules and release energy to the environment by doing work and releasing heat. Like all things in the physical world, energy is subject to physical laws. The laws of thermodynamics govern the transfer of energy in and among all systems in the universe.

In general, energy is defined as the ability to do work, or to create some kind of change. Energy exists in different forms. For example, electrical energy, light energy, and heat energy are all different types of energy. To appreciate the way energy flows into and out of biological systems, it is important to understand two of the physical laws that govern energy.

The First Law of Thermodynamics

The first law of thermodynamics states that the total amount of energy in the universe is constant and conserved. In other words, there has always been, and always will be, exactly the same amount of energy in the universe. Energy exists in many different forms. According to the first law of thermodynamics, energy may be transferred from place to place or transformed into different forms, but it cannot be created or destroyed. The transfers and transformations of energy take place around us all the time. Light bulbs transform electrical energy into light and heat energy. Gas stoves transform chemical energy from natural gas into heat energy. Plants perform one of the most biologically useful energy transformations on earth: that of converting the energy of sunlight to chemical energy stored within organic molecules (Figure 1). Some examples of energy transformations are shown in (Figure 1).

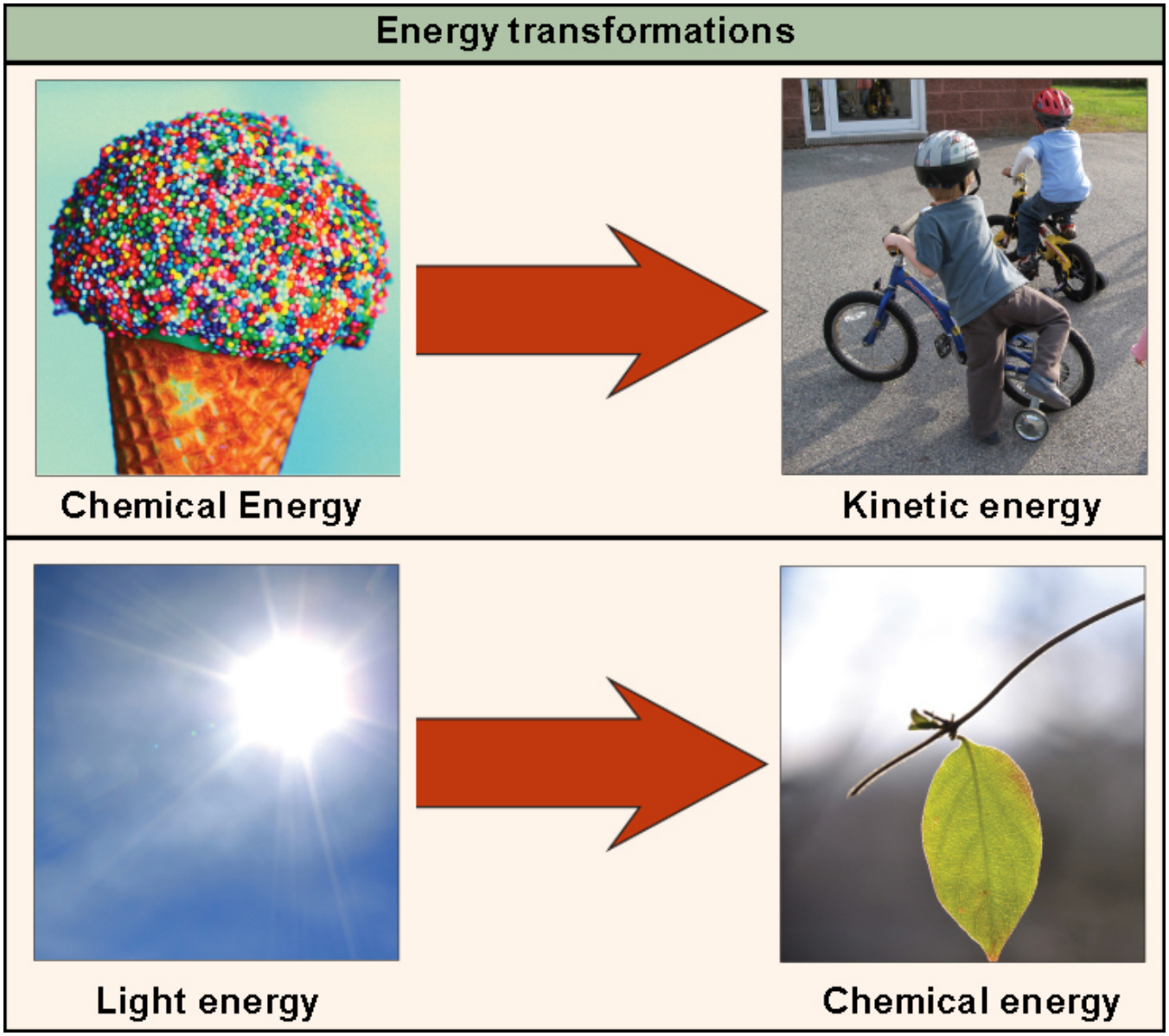

Figure 1. Shown are some examples of energy transferred and transformed from one system to another and from one form to another. The food we consume provides our cells with the energy required to carry out bodily functions, just as light energy provides plants with the means to create the chemical energy they need. (credit "ice cream": modification of work by D. Sharon Pruitt; credit "kids": modification of work by Max from Providence; credit "leaf": modification of work by Cory Zanker)

The challenge for all living organisms is to obtain energy from their surroundings in forms that they can transfer or transform into usable energy to do work. Living cells have evolved to meet this challenge very well. Chemical energy stored within organic molecules such as sugars and fats is transformed through a series of cellular chemical reactions into energy within molecules of ATP. Energy in ATP molecules is easily accessible to do work. Examples of the types of work that cells need to do include building complex molecules, transporting materials, powering the beating motion of cilia or flagella, contracting muscle fibers to create movement, and reproduction.

The Second Law of Thermodynamics

The challenge for all living organisms is to obtain energy from their surroundings in forms that they can transfer or transform into usable energy to do work. Living cells have evolved to meet this challenge. Chemical energy stored within organic molecules such as sugars and fats is transferred and transformed through a series of cellular chemical reactions into energy within molecules of ATP. Energy in ATP molecules is easily accessible to do work. Examples of the types of work that cells need to do include building complex molecules, transporting materials, powering the motion of cilia or flagella, and contracting muscle fibers to create movement.

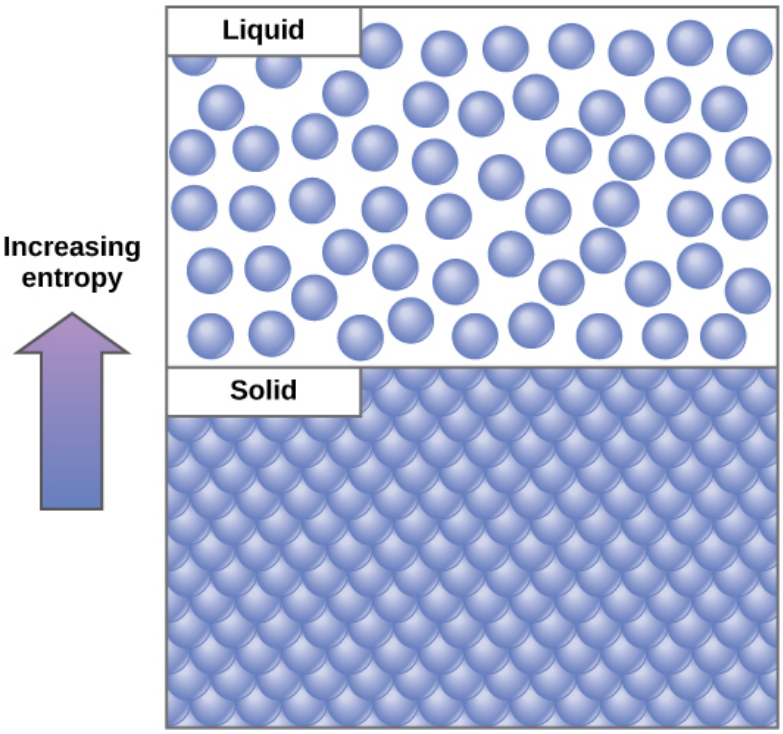

Figure 2. Entropy is a measure of randomness or disorder in a system. Gases have higher entropy than liquids, and liquids have higher entropy than solids.

A living cell's primary tasks of obtaining, transforming, and using energy to do work may seem simple. However, the second law of thermodynamics explains why these tasks are harder than they appear. All energy transfers and transformations are never completely efficient. In every energy transfer, some amount of energy is lost in a form that is unusable. In most cases, this form is heat energy. Thermodynamically, heat energy is defined as the energy transferred from one system to another that is not work. For example, when a light bulb is turned on, some of the energy being converted from electrical energy into light energy is lost as heat energy. Likewise, some energy is lost as heat energy during cellular metabolic reactions.

Try It Yourself

Set up a simple experiment to understand how energy is transferred and how a change in entropy results.

1. Take a block of ice. This is water in solid form, so it has a high structural order. This means that the molecules cannot move very much and are in a fixed position. The temperature of the ice is 0°C. As a result, the entropy of the system is low.
2. Allow the ice to melt at room temperature. What is the state of molecules in the liquid water now? How did the energy transfer take place? Is the entropy of the system higher or lower? Why?
3. Heat the water to its boiling point. What happens to the entropy of the system when the water is heated?

An important concept in physical systems is that of order and disorder. The more energy that is lost by a system to its surroundings, the less ordered and more random the system is. Scientists refer to the measure of randomness or disorder within a system as entropy. High entropy means high disorder and low energy. Molecules and chemical reactions have varying entropy as well. For example, entropy increases as molecules at a high concentration in one place diffuse and spread out. The second law of thermodynamics says that energy will always be lost as heat in energy transfers or transformations.

Living things are highly ordered, requiring constant energy input to be maintained in a state of low entropy.

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ENERGY

Learning outcomes

- Explain the difference between kinetic and potential energy
- Describe endergonic and exergonic reactions

Potential and Kinetic Energy

When an object is in motion, there is energy associated with that object. Think of a wrecking ball. Even a slow-moving wrecking ball can do a great deal of damage to other objects. Energy associated with objects in motion is called kinetic energy (Figure 1). A speeding bullet, a walking person, and the rapid movement of molecules in the air (which produces heat) all have kinetic energy.

Figure 1. Still water has potential energy; moving water, such as in a waterfall or a rapidly flowing river, has kinetic energy. (credit "dam": modification of work by "Pascal"/Flickr; credit "waterfall": modification of work by Frank Gualtieri)

Now what if that same motionless wrecking ball is lifted two stories above ground with a crane? If the suspended wrecking ball is unmoving, is there energy associated with it? The answer is yes. The energy that was required to lift the wrecking ball did not disappear, but is now stored in the wrecking ball by virtue of its position and the force of gravity acting on it. This type of energy is called potential energy (Figure 1). If the ball were to fall, the potential energy would be transformed into kinetic energy until all of the potential energy was exhausted when the ball rested on the ground. Wrecking balls also swing like a pendulum; through the swing, there is a constant change of potential energy (highest at the top of the swing) to kinetic energy (highest at the bottom of the swing). Other

examples of potential energy include the energy of water held behind a dam or a person about to skydive out of an airplane.

Potential energy is not only associated with the location of matter, but also with the structure of matter. Even a spring on the ground has potential energy if it is compressed; so does a rubber band that is pulled taut. On a molecular level, the bonds that hold the atoms of molecules together exist in a particular structure that has potential energy. Remember that anabolic cellular pathways require energy to synthesize complex molecules from simpler ones and catabolic pathways release energy when complex molecules are broken down. The fact that energy can be released by the breakdown of certain chemical bonds implies that those bonds have potential energy. In fact, there is potential energy stored within the bonds of all the food molecules we eat, which is eventually harnessed for use. This is because these bonds can release energy when broken. The type of potential energy that exists within chemical bonds, and is released when those bonds are broken, is called chemical energy. Chemical energy is responsible for providing living cells with energy from food. The release of energy occurs when the molecular bonds within food molecules are broken.

Free and Activation Energy

After learning that chemical reactions release energy when energy-storing bonds are broken, an important next question is the following: How is the energy associated with these chemical reactions quantified and expressed? How can the energy released from one reaction be compared to that of another reaction? A measurement of free energy is used to quantify these energy transfers. Recall that according to the second law of thermodynamics, all energy transfers involve the loss of some amount of energy in an unusable form such as heat. Free energy specifically refers to the energy associated with a chemical reaction that is available after the losses are accounted for. In other words, free energy is usable energy, or energy that is available to do work.

If energy is released during a chemical reaction, then the change in free energy, signified as ΔG (delta G) will be a negative number. A negative change in free energy also means that the products of the reaction have less free energy than the reactants, because they release some free energy during the reaction. Reactions that have a negative change in free energy and consequently release free energy are called exergonic reactions. Think: *ex*ergonic means energy is *ex*iting the system. These reactions are also referred to as spontaneous reactions, and their products have less stored energy than the reactants. An important distinction must be drawn between the term spontaneous and the idea of a chemical reaction occurring immediately. Contrary to the everyday use of the term, a spontaneous reaction is not one that suddenly or quickly occurs. The rusting of iron is an example of a spontaneous reaction that occurs slowly, little by little, over time.

If a chemical reaction absorbs energy rather than releases energy on balance, then the ΔG for that reaction will be a positive value. In this case, the products have more free energy than the reactants. Thus, the products of these reactions can be thought of as energy-storing molecules. These chemical reactions are called endergonic reactions and they are non-spontaneous. An endergonic reaction will not take place on its own without the addition of free energy.

There is another important concept that must be considered regarding endergonic and exergonic reactions. Exergonic reactions require a small amount of energy input to get going, before they can proceed with their energy-releasing steps. These reactions have a net release of energy, but still require some energy input in the beginning. This small amount of energy input necessary for all chemical reactions to occur is called the activation energy.

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ENZYMES

Learning Outcomes

Discuss how enzymes function as molecular catalysts

A substance that helps a chemical reaction to occur is called a catalyst, and the molecules that catalyze biochemical reactions are called enzymes. Most enzymes are proteins and perform the critical task of lowering the activation energies of chemical reactions inside the cell. Most of the reactions critical to a living cell happen too slowly at normal temperatures to be of any use to the cell. Without enzymes to speed up these reactions, life could not persist. Enzymes do this by binding to the reactant molecules and holding them in such a way as to make the chemical bond-breaking and -forming processes take place more easily. It is important to remember that enzymes do not change whether a reaction is exergonic (spontaneous) or endergonic. This is because they do not change the free energy of the reactants or products. They only reduce the activation energy required for the reaction to go forward (Figure 1). In addition, an enzyme itself is unchanged by the reaction it catalyzes. Once one reaction has been catalyzed, the enzyme is able to participate in other reactions.

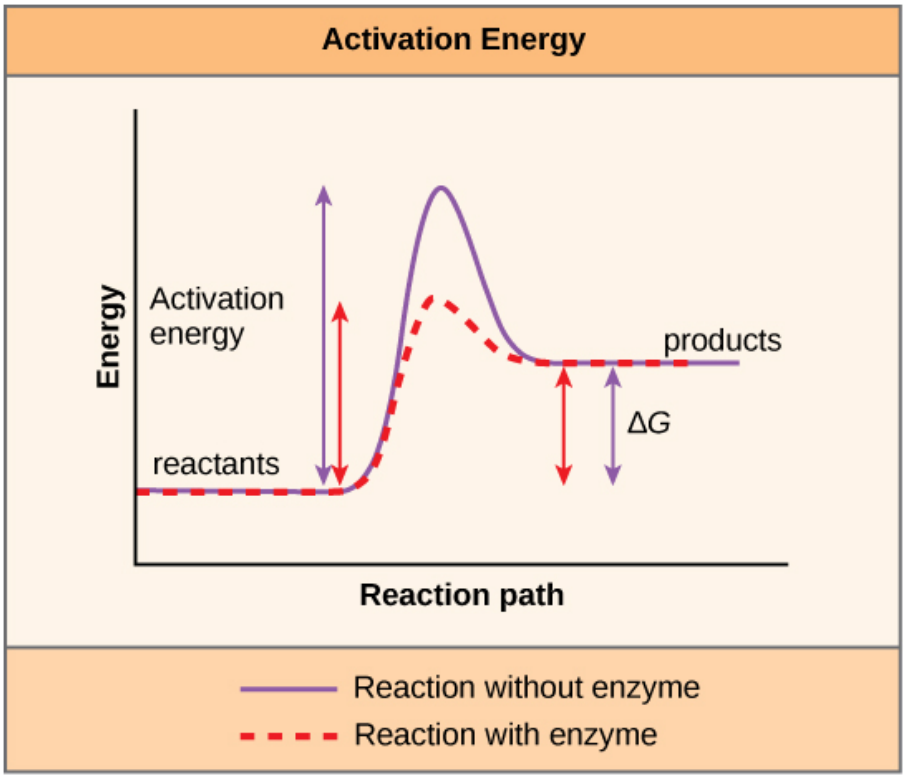

Figure 1. Enzymes lower the activation energy of the reaction but do not change the free energy of the reaction.

The chemical reactants to which an enzyme binds are called the enzyme's substrates. There may be one or more substrates, depending on the particular chemical reaction. In some reactions, a single reactant substrate is broken down into multiple products. In others, two substrates may come together to create one larger molecule. Two reactants might also enter a reaction and both become modified, but they leave the reaction as two products. The location within the enzyme where the substrate binds is called the enzyme's active site. The active site is where the "action" happens. Since enzymes are proteins, there is a unique combination of amino acid side chains within the active site. Each side chain is characterized by different properties. They can be large or small, weakly acidic or basic, hydrophilic or hydrophobic, positively or negatively charged, or neutral. The unique combination of side chains creates a very specific chemical environment within the active site. This specific environment is suited to bind to one specific chemical substrate (or substrates).

Active sites are subject to influences of the local environment. Increasing the environmental temperature generally increases reaction rates, enzyme-catalyzed or otherwise. However, temperatures outside of an optimal range reduce the rate at which an enzyme catalyzes a reaction. Hot temperatures will eventually cause enzymes to denature, an irreversible change in the three-dimensional shape and therefore the function of the enzyme.

Enzymes are also suited to function best within a certain pH and salt concentration range, and, as with temperature, extreme pH, and salt concentrations can cause enzymes to denature.

For many years, scientists thought that enzyme-substrate binding took place in a simple “lock and key” fashion. This model asserted that the enzyme and substrate fit together perfectly in one instantaneous step. However, current research supports a model called induced fit (Figure 2). The induced-fit model expands on the lock-and-key model by describing a more dynamic binding between enzyme and substrate. As the enzyme and substrate come together, their interaction causes a mild shift in the enzyme’s structure that forms an ideal binding arrangement between enzyme and substrate.

View this animation of induced fit.

When an enzyme binds its substrate, an enzyme-substrate complex is formed. This complex lowers the activation energy of the reaction and promotes its rapid progression in one of multiple possible ways. On a basic level, enzymes promote chemical reactions that involve more than one substrate by bringing the substrates together in an optimal orientation for reaction. Another way in which enzymes promote the reaction of their substrates is by creating an optimal environment within the active site for the reaction to occur.

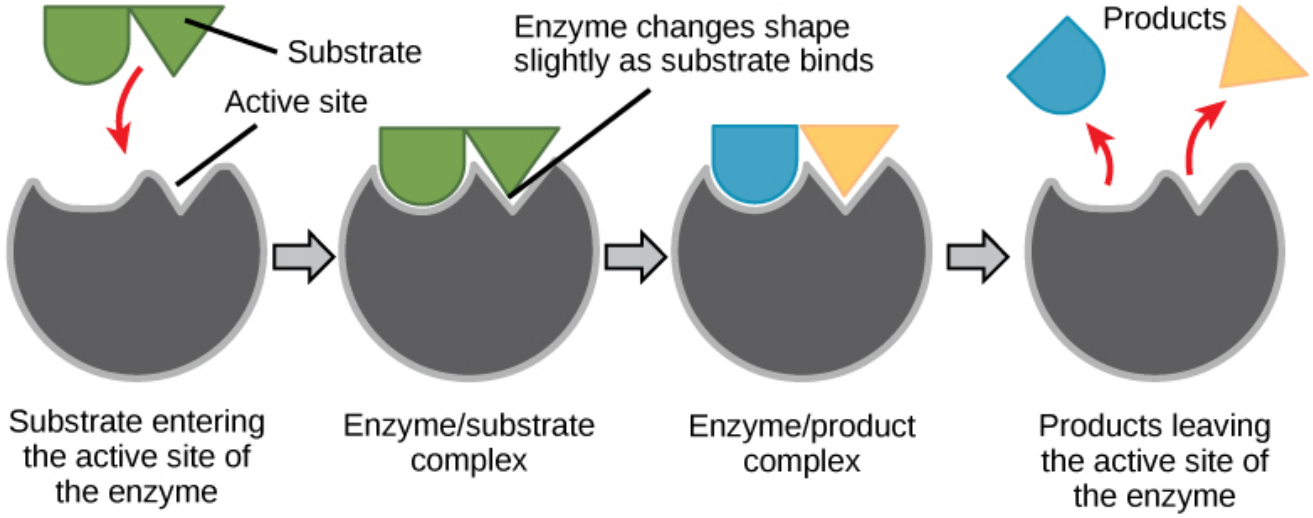

Figure 2. The induced-fit model is an adjustment to the lock-and-key model and explains how enzymes and substrates undergo dynamic modifications during the transition state to increase the affinity of the substrate for the active site.

Careers in Action: Pharmaceutical Drug Developer

Enzymes are key components of metabolic pathways. Understanding how enzymes work and how they can be regulated are key principles behind the development of many of the pharmaceutical drugs on the market today. Biologists working in this field collaborate with other scientists to design drugs.

Consider statins for example—statins is the name given to one class of drugs that can reduce cholesterol levels. These compounds are inhibitors of the enzyme HMG-CoA reductase, which is the enzyme that synthesizes cholesterol from lipids in the body. By inhibiting this enzyme, the level of cholesterol synthesized in the body can be reduced. Similarly, acetaminophen, popularly marketed under the brand name Tylenol, is an inhibitor of the enzyme cyclooxygenase. While it is used to provide relief from fever and inflammation (pain), its mechanism of action is still not completely understood.

Figure 3. Have you ever wondered how pharmaceutical drugs are developed? (credit: Deborah Austin)

How are drugs discovered? One of the biggest challenges in drug discovery is identifying a drug target. A drug target is a molecule that is literally the target of the drug. In the case of statins, HMG-CoA reductase is the drug target. Drug targets are identified through painstaking research in the laboratory. Identifying the target alone is not enough; scientists also need to know how the target acts inside the cell and which reactions go awry in the case of disease. Once the target and the pathway are identified, then the actual process of drug design begins. In this stage, chemists and biologists work together to design and synthesize molecules that can block or activate a particular reaction. However, this is only the beginning: If and when a drug prototype is successful in performing its function, then it is subjected to many tests from in vitro experiments to clinical trials before it can get approval from the U.S. Food and Drug Administration to be on the market.

Many enzymes do not work optimally, or even at all, unless bound to other specific non-protein helper molecules. They may bond either temporarily through ionic or hydrogen bonds, or permanently through stronger covalent bonds. Binding to these molecules promotes optimal shape and function of their respective enzymes. Two examples of these types of helper molecules are cofactors and coenzymes. Cofactors are inorganic ions such as ions of iron and magnesium. Coenzymes are organic helper molecules, those with a basic atomic structure made up of carbon and hydrogen. Like enzymes, these molecules participate in reactions without being changed themselves and are ultimately recycled and reused. Vitamins are the source of coenzymes. Some vitamins are the precursors of coenzymes and others act directly as coenzymes. Vitamin C is a direct coenzyme for multiple enzymes that take part in building the important connective tissue, collagen. Therefore, enzyme function is, in part, regulated by the abundance of various cofactors and coenzymes, which may be supplied by an organism's diet or, in some cases, produced by the organism.

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SUMMARY: ENERGY AND METABOLISM

Learning Outcomes

- Identify different types of metabolic pathways
- Distinguish between an open and a closed system
- State the first law of thermodynamics
- State the second law of thermodynamics
- Explain the difference between kinetic and potential energy
- Describe endergonic and exergonic reactions
- Discuss how enzymes function as molecular catalysts

Cells perform the functions of life through various chemical reactions. A cell's metabolism refers to the combination of chemical reactions that take place within it. Catabolic reactions break down complex chemicals into simpler ones and are associated with energy release. Anabolic processes build complex molecules out of simpler ones and require energy.

In studying energy, the term system refers to the matter and environment involved in energy transfers. Entropy is a measure of the disorder of a system. The physical laws that describe the transfer of energy are the laws of thermodynamics. The first law states that the total amount of energy in the universe is constant. The second law of thermodynamics states that every energy transfer involves some loss of energy in an unusable form, such as heat energy. Energy comes in different forms: kinetic, potential, and free. The change in free energy of a reaction can be negative (releases energy, exergonic) or positive (consumes energy, endergonic). All reactions require an initial input of energy to proceed, called the activation energy.

Enzymes are chemical catalysts that speed up chemical reactions by lowering their activation energy. Enzymes have an active site with a unique chemical environment that fits particular chemical reactants for that enzyme, called substrates. Enzymes and substrates are thought to bind according to an induced-fit model. Enzyme action is regulated to conserve resources and respond optimally to the environment.

Practice Questions

1. Look at each of the processes shown in Figure 1, and decide if it is endergonic or exergonic.

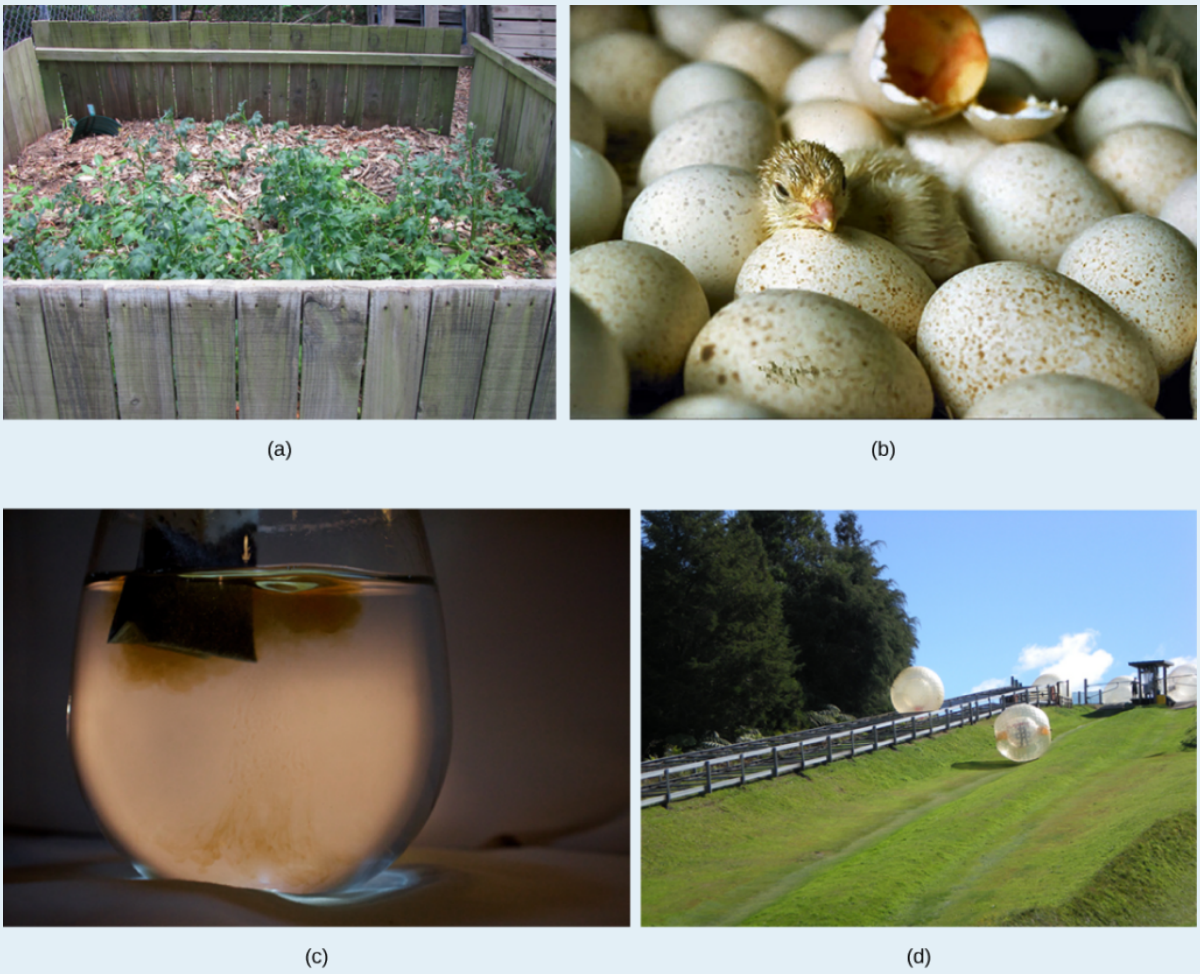

Figure 1. Shown are some examples of endergonic processes (ones that require energy) and exergonic processes (ones that release energy). (credit a: modification of work by Natalie Maynor; credit b: modification of work by USDA; credit c: modification of work by Cory Zanker; credit d: modification of work by Harry Malsch)

2. Does physical exercise to increase muscle mass involve anabolic and/or catabolic processes? Give evidence for your answer.
3. Explain in your own terms the difference between a spontaneous reaction and one that occurs instantaneously, and what causes this difference.
4. With regard to enzymes, why are vitamins and minerals necessary for good health? Give examples.

Answers

1. A compost pile decomposing is an exergonic process. A baby developing from a fertilized egg is an endergonic process. Tea dissolving into water is an exergonic process. A ball rolling downhill is an exergonic process.
2. Physical exercise involves both anabolic and catabolic processes. Body cells break down sugars to provide ATP to do the work necessary for exercise, such as muscle contractions. This is catabolism. Muscle cells also must repair muscle tissue damaged by exercise by building new muscle. This is anabolism.
3. A spontaneous reaction is one that has a negative ΔG and thus releases energy. However, a spontaneous reaction need not occur quickly or suddenly like an instantaneous reaction. It may occur over long periods of time due to a large energy of activation, which prevents the reaction from occurring quickly.

4. Most vitamins and minerals act as cofactors and coenzymes for enzyme action. Many enzymes require the binding of certain cofactors or coenzymes to be able to catalyze their reactions. Since enzymes catalyze many important reactions, it is critical to obtain sufficient vitamins and minerals from diet and supplements. Vitamin C (ascorbic acid) is a coenzyme necessary for the action of enzymes that build collagen.

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INTRODUCTION TO ATP IN LIVING SYSTEMS

What you'll learn to do: Describe how cells store and transfer free energy using ATP

All living things require energy to function. While different organisms acquire this energy in different ways, they store (and use it) in the same way. In this section, we'll learn about ATP—the energy of life. ATP is how cells store energy. These storage molecules are produced in the mitochondria, tiny organelles found in eukaryotic cells sometimes called the "powerhouse" of the cell.

Mitochondrial Disease Physician

What happens when the critical reactions of cellular respiration do not proceed correctly? Mitochondrial diseases are genetic disorders of metabolism. Mitochondrial disorders can arise from mutations in nuclear or mitochondrial DNA, and they result in the production of less energy than is normal in body cells.

In type 2 diabetes, for instance, the oxidation efficiency of NADH is reduced, impacting oxidative phosphorylation but not the other steps of respiration. Symptoms of mitochondrial diseases can include muscle weakness, lack of coordination, stroke-like episodes, and loss of vision and hearing. Most affected people are diagnosed in childhood, although there are some adult-onset diseases.

Identifying and treating mitochondrial disorders is a specialized medical field. The educational preparation for this profession requires a college education, followed by medical school with a specialization in medical genetics. Medical geneticists can be board certified by the American Board of Medical Genetics and go on to become associated with professional organizations devoted to the study of mitochondrial diseases, such as the Mitochondrial Medicine Society and the Society for Inherited Metabolic Disease.

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ATP IN LIVING SYSTEMS

Learning Outcomes

Describe how cells store and transfer free energy using ATP

A living cell cannot store significant amounts of free energy. Excess free energy would result in an increase of heat in the cell, which would result in excessive thermal motion that could damage and then destroy the cell. Rather, a cell must be able to handle that energy in a way that enables the cell to store energy safely and release it for use only as needed. Living cells accomplish this by using the compound adenosine triphosphate (ATP). ATP is often called the “energy currency” of the cell, and, like currency, this versatile compound can be used to fill any energy need of the cell. How? It functions similarly to a rechargeable battery.

Watch this video online: https://youtu.be/00jbG_cfGuQ

When ATP is broken down, usually by the removal of its terminal phosphate group, energy is released. The energy is used to do work by the cell, usually by the released phosphate binding to another molecule, activating it. For example, in the mechanical work of muscle contraction, ATP supplies the energy to move the contractile muscle proteins. Recall the active transport work of the sodium-potassium pump in cell membranes. ATP alters the structure of the integral protein that functions as the pump, changing its affinity for sodium and potassium. In this way, the cell performs work, pumping ions against their electrochemical gradients.

ATP Structure and Function

At the heart of ATP is a molecule of adenosine monophosphate (AMP), which is composed of an adenine molecule bonded to a ribose molecule and to a single phosphate group (Figure 1). Ribose is a five-carbon sugar found in RNA, and AMP is one of the nucleotides in RNA. The addition of a second phosphate group to this core molecule results in the formation of adenosine diphosphate (ADP); the addition of a third phosphate group forms adenosine triphosphate (ATP).

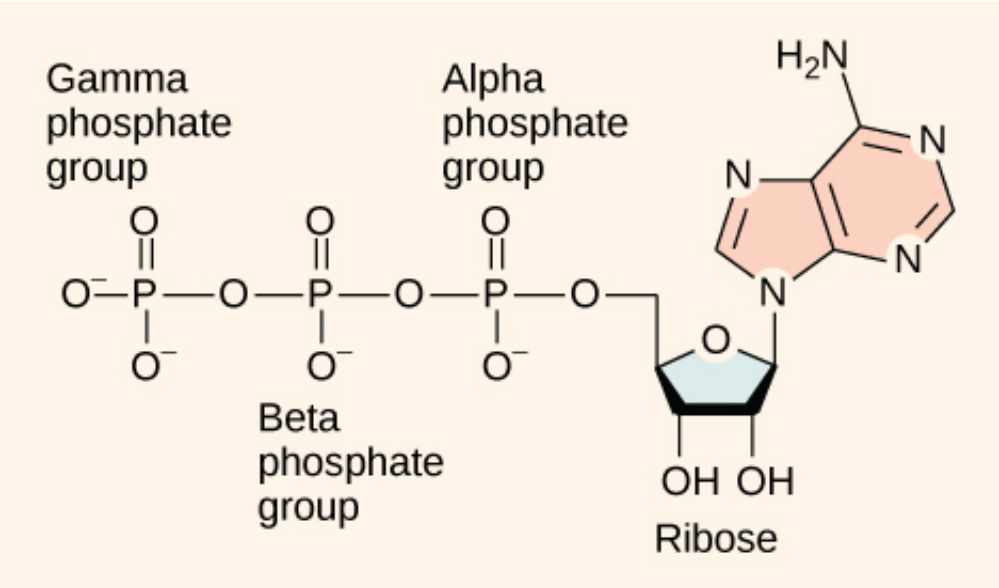

Figure 1. ATP (adenosine triphosphate) has three phosphate groups that can be removed by hydrolysis to form ADP (adenosine diphosphate) or AMP (adenosine monophosphate).The negative charges on the phosphate group naturally repel each other, requiring energy to bond them together and releasing energy when these bonds are broken.

The addition of a phosphate group to a molecule requires energy. Phosphate groups are negatively charged and thus repel one another when they are arranged in series, as they are in ADP and ATP. This repulsion makes the ADP and ATP molecules inherently unstable. The release of one or two phosphate groups from ATP, a process called **dephosphorylation**, releases energy.

Energy from ATP

Hydrolysis is the process of breaking complex macromolecules apart. During hydrolysis, water is split, or lysed, and the resulting hydrogen atom (H^+) and a hydroxyl group (OH^-) are added to the larger molecule. The hydrolysis of ATP produces ADP, together with an inorganic phosphate ion (P_i), and the release of free energy. To carry out life processes, ATP is continuously broken down into ADP, and like a rechargeable battery, ADP is continuously regenerated into ATP by the reattachment of a third phosphate group. Water, which was broken down into its hydrogen atom and hydroxyl group during ATP hydrolysis, is regenerated when a third phosphate is added to the ADP molecule, reforming ATP.

Obviously, energy must be infused into the system to regenerate ATP. Where does this energy come from? In nearly every living thing on earth, the energy comes from the metabolism of glucose. In this way, ATP is a direct link between the limited set of exergonic pathways of glucose catabolism and the multitude of endergonic pathways that power living cells.

Phosphorylation

Recall that, in some chemical reactions, enzymes may bind to several substrates that react with each other on the enzyme, forming an intermediate complex. An intermediate complex is a temporary structure, and it allows one of the substrates (such as ATP) and reactants to more readily react with each other; in reactions involving ATP, ATP is one of the substrates and ADP is a product. During an endergonic chemical reaction, ATP forms an intermediate complex with the substrate and enzyme in the reaction. This intermediate complex allows the ATP to transfer its third phosphate group, with its energy, to the substrate, a process called phosphorylation. **Phosphorylation** refers to the addition of the phosphate (~P). This is illustrated by the following generic reaction:

$$A + \text{enzyme} + ATP \rightarrow [A - \text{enzyme} - {\sim}P] \rightarrow B + \text{enzyme} + ADP + \text{phosphate ion}$$

When the intermediate complex breaks apart, the energy is used to modify the substrate and convert it into a product of the reaction. The ADP molecule and a free phosphate ion are released into the medium and are available for recycling through cell metabolism.

Substrate Phosphorylation

ATP is generated through two mechanisms during the breakdown of glucose. A few ATP molecules are generated (that is, regenerated from ADP) as a direct result of the chemical reactions that occur in the catabolic pathways. A phosphate group is removed from an intermediate reactant in the pathway, and the free energy of the reaction is used to add the third phosphate to an available ADP molecule, producing ATP (Figure 2). This very direct method of phosphorylation is called **substrate-level phosphorylation**.

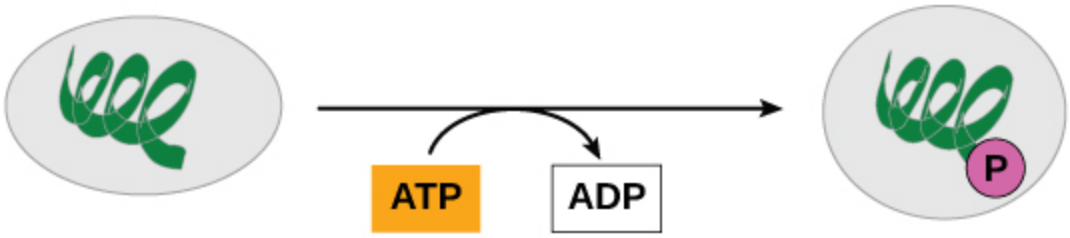

Figure 2. In phosphorylation reactions, the gamma phosphate of ATP is attached to a protein.

Oxidative Phosphorylation

Most of the ATP generated during glucose catabolism, however, is derived from a much more complex process, chemiosmosis, which takes place in mitochondria (Figure 3) within a eukaryotic cell or the plasma membrane of a prokaryotic cell.

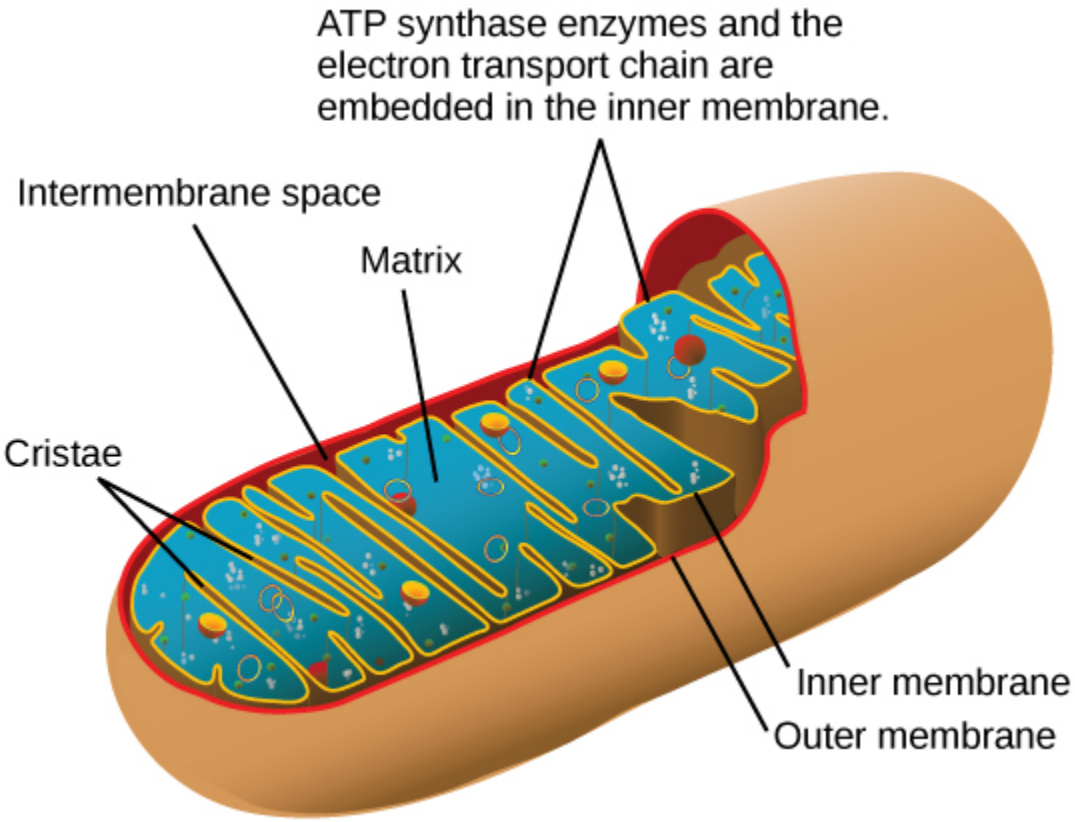

Figure 3. The mitochondria (Credit: modification of work by Mariana Ruiz Villareal)

Chemiosmosis, a process of ATP production in cellular metabolism, is used to generate 90 percent of the ATP made during glucose catabolism and is also the method used in the light reactions of photosynthesis to harness the energy of sunlight. The production of ATP using the process of chemiosmosis is called **oxidative phosphorylation** because of the involvement of oxygen in the process.

In Summary: ATP in Living Systems

ATP functions as the energy currency for cells. It allows the cell to store energy briefly and transport it within the cell to support endergonic chemical reactions. The structure of ATP is that of an RNA nucleotide with three phosphates attached. As ATP is used for energy, a phosphate group or two are detached, and either ADP or AMP is produced. Energy derived from glucose catabolism is used to convert ADP into ATP. When ATP is used in a reaction, the third phosphate is temporarily attached to a substrate in a process called phosphorylation. The two processes of ATP regeneration that are used in conjunction with glucose catabolism are substrate-level phosphorylation and oxidative phosphorylation through the process of chemiosmosis.

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INTRODUCTION TO CELLULAR RESPIRATION

What you'll learn to do: Identify the reactants and products of cellular respiration and where these reactions occur in a cell

Virtually every task performed by living organisms requires energy. Energy is needed to perform heavy labor and exercise, but humans also use energy while thinking, and even during sleep. In fact, the living cells of every organism constantly use energy.

Nutrients and other molecules are imported into the cell, metabolized (broken down) and possibly synthesized into new molecules, modified if needed, transported around the cell, and possibly distributed to the entire organism. For example, the large proteins that make up muscles are built from smaller molecules imported from dietary amino acids. Complex carbohydrates are broken down into simple sugars that the cell uses for energy.

Figure 1. A hummingbird needs energy to maintain prolonged flight. The bird obtains its energy from taking in food and transforming the energy contained in food molecules into forms of energy to power its flight through a series of biochemical reactions. (credit: modification of work by Cory Zanker)

Just as energy is required to both build and demolish a building, energy is required for the synthesis and breakdown of molecules as well as the transport of molecules into and out of cells. In addition, processes such as ingesting and breaking down pathogenic bacteria and viruses, exporting wastes and toxins, and movement of the cell require energy. From where, and in what form, does this energy come? How do living cells obtain energy, and how do they use it? This chapter will discuss different forms of energy and the physical laws that govern energy transfer. This chapter will also describe how cells use energy and replenish it, and how chemical reactions in the cell are performed with great efficiency.

In the process of photosynthesis, plants and other photosynthetic producers create glucose, which stores energy in its chemical bonds. You will actually study photosynthesis in more detail a bit later. But once photosynthesis

has created glucose to store energy, both plants and consumers, such as animals, undergo a series of metabolic pathways, collectively called cellular respiration, to use that energy. Cellular respiration extracts the energy from the bonds in glucose and converts it into a form that all living things can use. Now let's take a more detailed look at how all eukaryotes—which includes humans!—make use of this stored energy.

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GLYCOLYSIS

Learning Outcomes

Describe the process of glycolysis and identify its reactants and products

Even exergonic, energy-releasing reactions require a small amount of activation energy to proceed. However, consider endergonic reactions, which require much more energy input because their products have more free energy than their reactants. Within the cell, where does energy to power such reactions come from? The answer lies with an energy-supplying molecule called adenosine triphosphate, or **ATP**. ATP is a small, relatively simple molecule, but within its bonds contains the potential for a quick burst of energy that can be harnessed to perform cellular work. This molecule can be thought of as the primary energy currency of cells in the same way that money is the currency that people exchange for things they need. ATP is used to power the majority of energy-requiring cellular reactions.

ATP in Living Systems

A living cell cannot store significant amounts of free energy. Excess free energy would result in an increase of heat in the cell, which would denature enzymes and other proteins, and thus destroy the cell. Rather, a cell must be able to store energy safely and release it for use only as needed. Living cells accomplish this using ATP, which can be used to fill any energy need of the cell. How? It functions as a rechargeable battery.

When ATP is broken down, usually by the removal of its terminal phosphate group, energy is released. This energy is used to do work by the cell, usually by the binding of the released phosphate to another molecule, thus activating it. For example, in the mechanical work of muscle contraction, ATP supplies energy to move the contractile muscle proteins.

ATP Structure and Function

At the heart of ATP is a molecule of adenosine monophosphate (AMP), which is composed of an adenine molecule bonded to both a ribose molecule and a single phosphate group (Figure 1). Ribose is a five-carbon sugar found in RNA and AMP is one of the nucleotides in RNA. The addition of a second phosphate group to this core molecule results in adenosine diphosphate (ADP); the addition of a third phosphate group forms adenosine triphosphate (ATP).

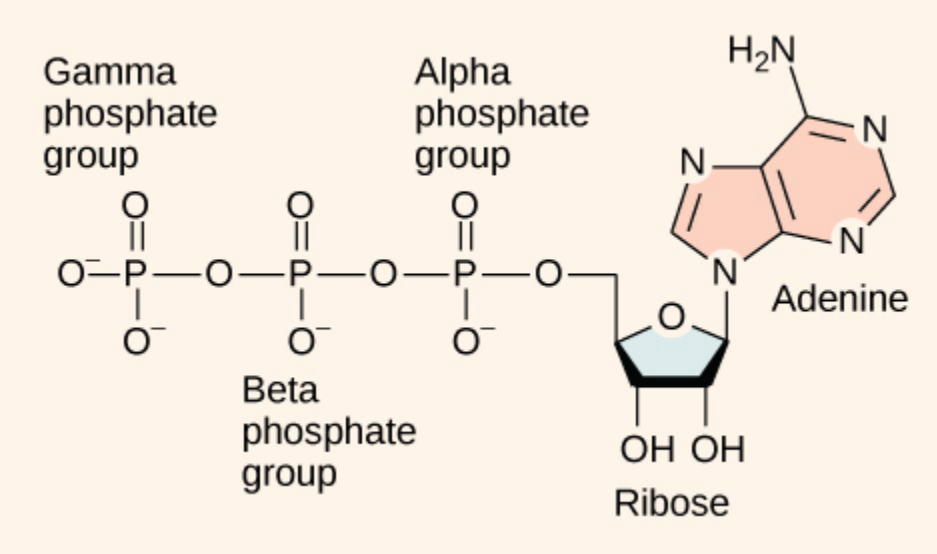

Figure 1. The structure of ATP shows the basic components of a two-ring adenine, five-carbon ribose, and three phosphate groups.

The addition of a phosphate group to a molecule requires a high amount of energy and results in a high-energy bond. Phosphate groups are negatively charged and thus repel one another when they are arranged in series, as they are in ADP and ATP. This repulsion makes the ADP and ATP molecules inherently unstable. The release of one or two phosphate groups from ATP, a process called hydrolysis, releases energy.

Glycolysis

You have read that nearly all of the energy used by living things comes to them in the bonds of the sugar, glucose. **Glycolysis** is the first step in the breakdown of glucose to extract energy for cell metabolism. Many living organisms carry out glycolysis as part of their metabolism. Glycolysis takes place in the cytoplasm of most prokaryotic and all eukaryotic cells.

Glycolysis begins with the six-carbon, ring-shaped structure of a single glucose molecule and ends with two molecules of a three-carbon sugar called pyruvate. Glycolysis consists of two distinct phases. In the first part of the glycolysis pathway, energy is used to make adjustments so that the six-carbon sugar molecule can be split evenly into two three-carbon pyruvate molecules. In the second part of glycolysis, ATP and nicotinamide-adenine dinucleotide (NADH) are produced (Figure 2).

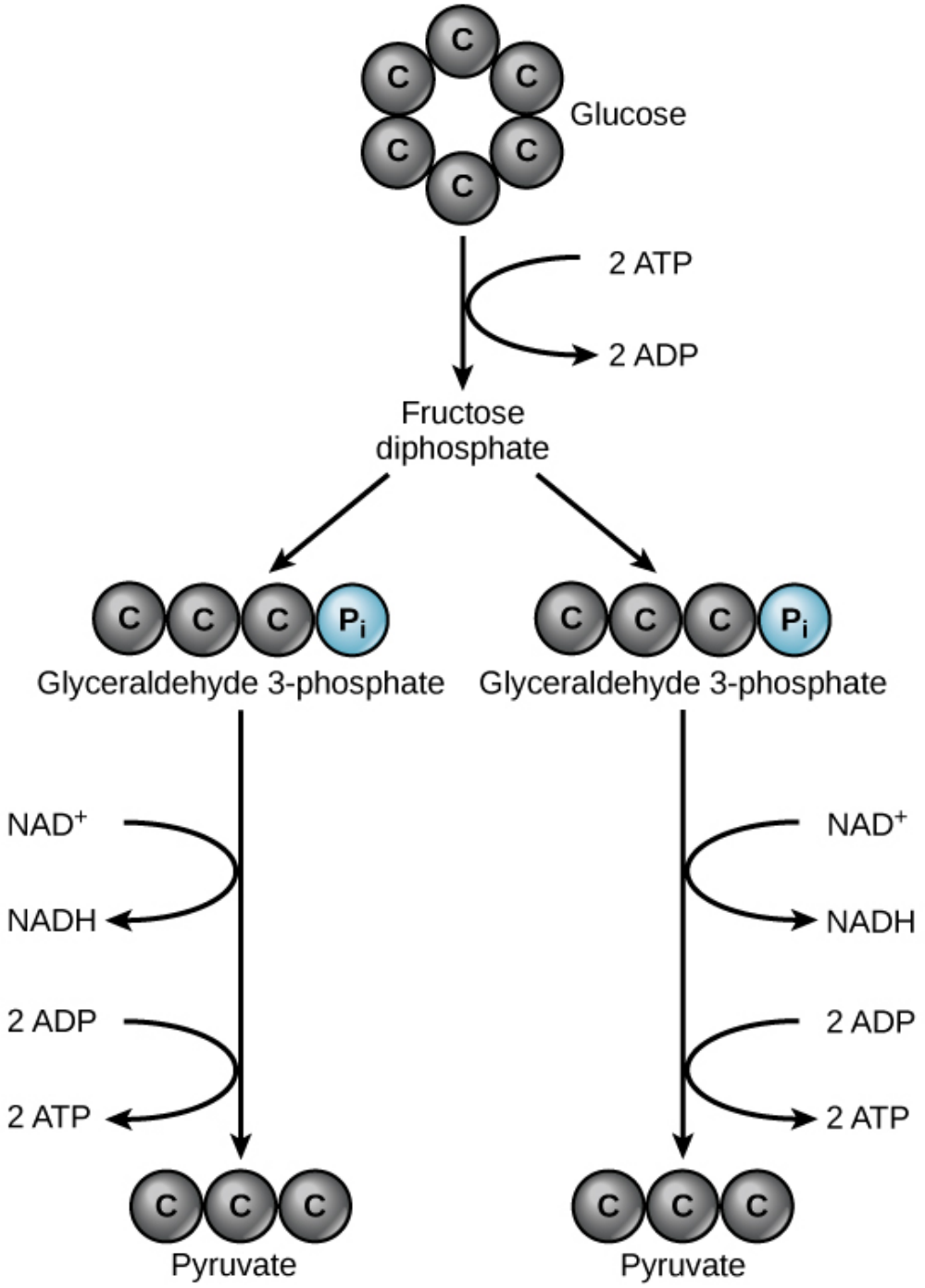

Figure 2. In glycolysis, a glucose molecule is converted into two pyruvate molecules.

If the cell cannot catabolize the pyruvate molecules further, it will harvest only two ATP molecules from one molecule of glucose. For example, mature mammalian red blood cells are only capable of glycolysis, which is their sole source of ATP. If glycolysis is interrupted, these cells would eventually die.

In Summary: Glycolysis

ATP functions as the energy currency for cells. It allows cells to store energy briefly and transport it within itself to support endergonic chemical reactions. The structure of ATP is that of an RNA nucleotide with three

phosphate groups attached. As ATP is used for energy, a phosphate group is detached, and ADP is produced. Energy derived from glucose catabolism is used to recharge ADP into ATP.

Glycolysis is the first pathway used in the breakdown of glucose to extract energy. Because it is used by nearly all organisms on earth, it must have evolved early in the history of life. Glycolysis consists of two parts: The first part prepares the six-carbon ring of glucose for separation into two three-carbon sugars. Energy from ATP is invested into the molecule during this step to energize the separation. The second half of glycolysis extracts ATP and high-energy electrons from hydrogen atoms and attaches them to NAD^+. Two ATP molecules are invested in the first half and four ATP molecules are formed during the second half. This produces a net gain of two ATP molecules per molecule of glucose for the cell.

Practice Question

Both prokaryotic and eukaryotic organisms carry out some form of glycolysis. How does that fact support or not support the assertion that glycolysis is one of the oldest metabolic pathways?

Answer

If glycolysis evolved relatively late, it likely would not be as universal in organisms as it is. It probably evolved in very primitive organisms and persisted, with the addition of other pathways of carbohydrate metabolism that evolved later.

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CITRIC ACID CYCLE AND OXIDATIVE PHOSPHORYLATION

Learning Outcomes

- Describe the process of the citric acid cycle (Krebs cycle) and identify its reactants and products
- Describe the overall outcome of the citric acid cycle and oxidative phosphorylation in terms of the products of each
- Describe the location of the citric acid cycle and oxidative phosphorylation in the cell

The Citric Acid Cycle

In eukaryotic cells, the pyruvate molecules produced at the end of glycolysis are transported into mitochondria, which are sites of cellular respiration. If oxygen is available, aerobic respiration will go forward. In mitochondria, pyruvate will be transformed into a two-carbon acetyl group (by removing a molecule of carbon dioxide) that will be picked up by a carrier compound called coenzyme A (CoA), which is made from vitamin B_5. The resulting compound is called acetyl CoA. (Figure 1). Acetyl CoA can be used in a variety of ways by the cell, but its major function is to deliver the acetyl group derived from pyruvate to the next pathway in glucose catabolism.

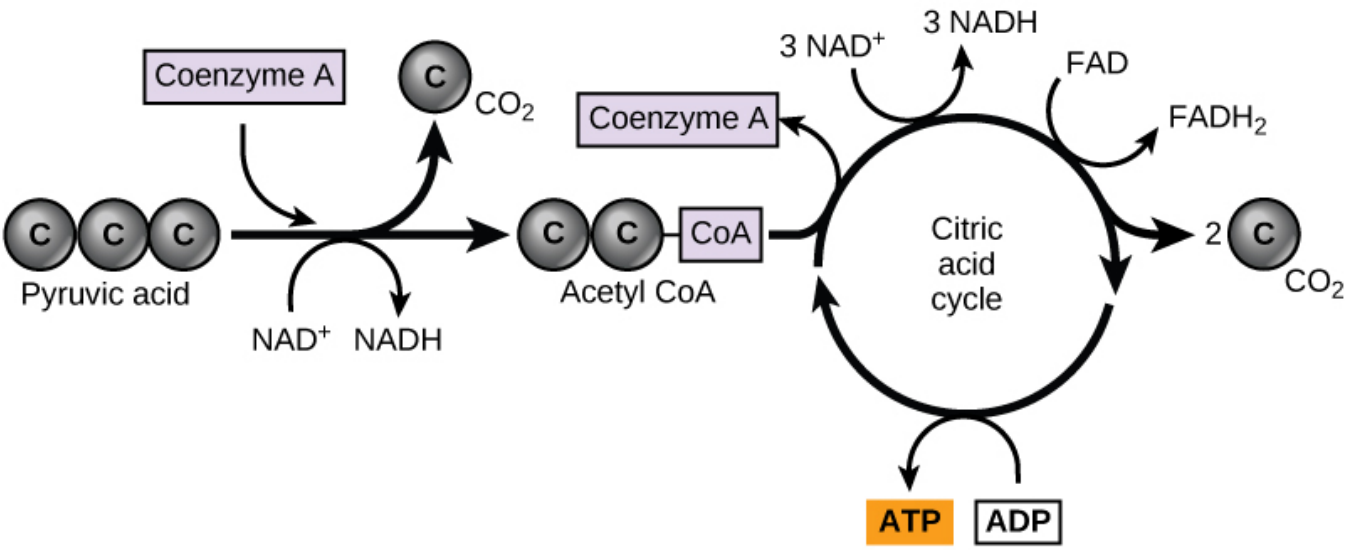

Figure 1. Pyruvate is converted into acetyl-CoA before entering the citric acid cycle.

Like the conversion of pyruvate to acetyl CoA, the citric acid cycle in eukaryotic cells takes place in the matrix of the mitochondria. Unlike glycolysis, the citric acid cycle is a closed loop: The last part of the pathway regenerates the compound used in the first step. The eight steps of the cycle are a series of chemical reactions that produces two carbon dioxide molecules, one ATP molecule (or an equivalent), and reduced forms (NADH and $FADH_2$) of NAD^+ and FAD^+, important coenzymes in the cell. Part of this is considered an aerobic pathway (oxygen-requiring) because the NADH and $FADH_2$ produced must transfer their electrons to the next pathway in the system, which will use oxygen. If oxygen is not present, this transfer does not occur.

Two carbon atoms come into the citric acid cycle from each acetyl group. Two carbon dioxide molecules are released on each turn of the cycle; however, these do not contain the same carbon atoms contributed by the acetyl group on that turn of the pathway. The two acetyl-carbon atoms will eventually be released on later turns of the cycle; in this way, all six carbon atoms from the original glucose molecule will be eventually released as carbon dioxide. It takes two turns of the cycle to process the equivalent of one glucose molecule. Each turn of the cycle forms three high-energy NADH molecules and one high-energy $FADH_2$ molecule. These high-energy carriers will connect with the last portion of aerobic respiration to produce ATP molecules. One ATP (or an equivalent) is also made in each cycle. Several of the intermediate compounds in the citric acid cycle can be used in synthesizing non-essential amino acids; therefore, the cycle is both anabolic and catabolic.

Oxidative Phosphorylation

You have just read about two pathways in glucose catabolism—glycolysis and the citric acid cycle—that generate ATP. Most of the ATP generated during the aerobic catabolism of glucose, however, is not generated directly from these pathways. Rather, it derives from a process that begins with passing electrons through a series of chemical reactions to a final electron acceptor, oxygen. These reactions take place in specialized protein complexes located in the inner membrane of the mitochondria of eukaryotic organisms and on the inner part of the cell membrane of prokaryotic organisms. The energy of the electrons is harvested and used to generate a electrochemical gradient across the inner mitochondrial membrane. The potential energy of this gradient is used to generate ATP. The entirety of this process is called oxidative phosphorylation.

The electron transport chain (Figure 2a) is the last component of aerobic respiration and is the only part of metabolism that uses atmospheric oxygen. Oxygen continuously diffuses into plants for this purpose. In animals, oxygen enters the body through the respiratory system. Electron transport is a series of chemical reactions that resembles a bucket brigade in that electrons are passed rapidly from one component to the next, to the endpoint of the chain where oxygen is the final electron acceptor and water is produced. There are four complexes composed of proteins, labeled I through IV in Figure 2c, and the aggregation of these four complexes, together with associated mobile, accessory electron carriers, is called the electron transport chain. The electron transport chain is present in multiple copies in the inner mitochondrial membrane of eukaryotes and in the plasma

membrane of prokaryotes. In each transfer of an electron through the electron transport chain, the electron loses energy, but with some transfers, the energy is stored as potential energy by using it to pump hydrogen ions across the inner mitochondrial membrane into the intermembrane space, creating an electrochemical gradient.

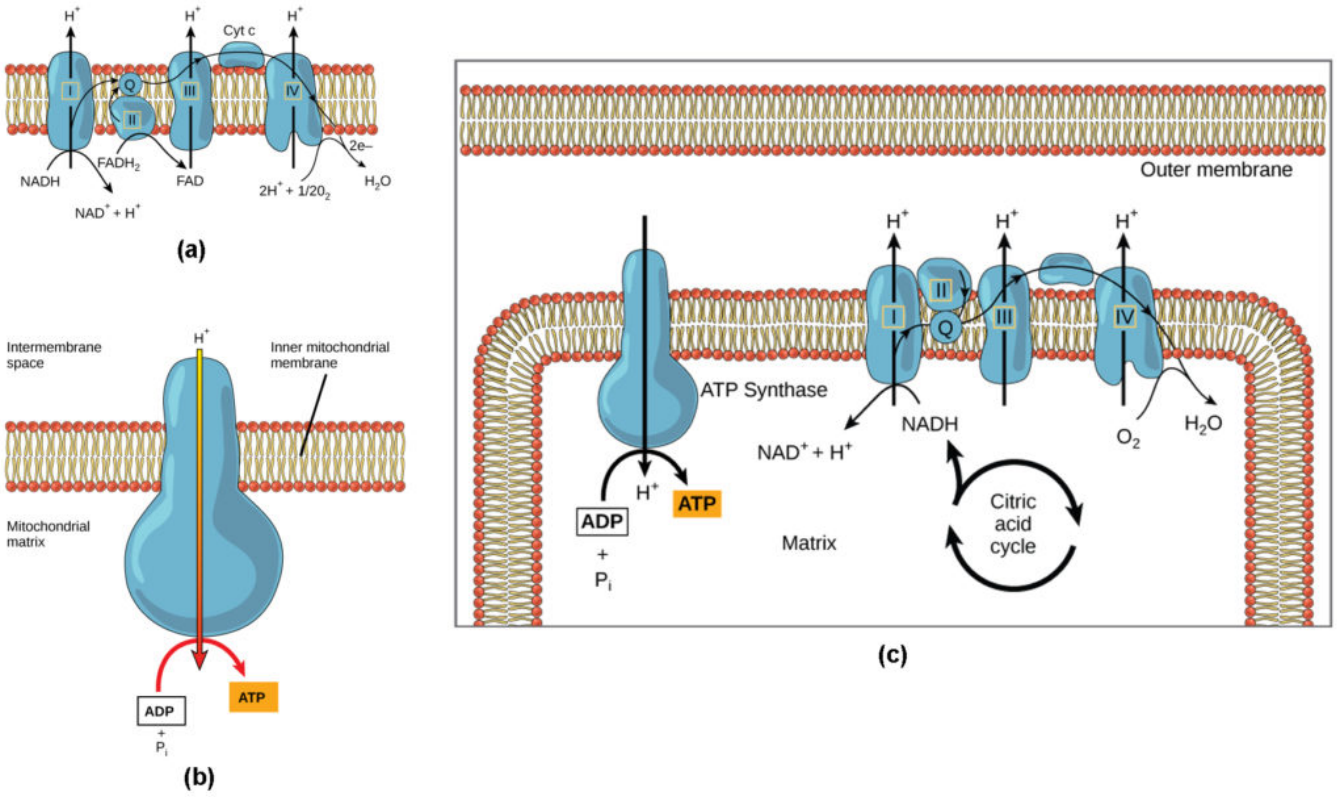

Figure 2. (a) The electron transport chain is a set of molecules that supports a series of oxidation-reduction reactions. (b) ATP synthase is a complex, molecular machine that uses an H+ gradient to regenerate ATP from ADP. (c) Chemiosmosis relies on the potential energy provided by the H+ gradient across the membrane.

Practice Question

Cyanide inhibits cytochrome c oxidase, a component of the electron transport chain. If cyanide poisoning occurs, would you expect the pH of the intermembrane space to increase or decrease? What affect would cyanide have on ATP synthesis?

Answer

After cyanide poisoning, the electron transport chain can no longer pump electrons into the intermembrane space. The pH of the intermembrane space would increase, and ATP synthesis would stop.

Electrons from NADH and $FADH_2$ are passed to protein complexes in the electron transport chain. As they are passed from one complex to another (there are a total of four), the electrons lose energy, and some of that energy is used to pump hydrogen ions from the mitochondrial matrix into the intermembrane space. In the fourth protein complex, the electrons are accepted by oxygen, the terminal acceptor. The oxygen with its extra electrons then combines with two hydrogen ions, further enhancing the electrochemical gradient, to form water. If there were no oxygen present in the mitochondrion, the electrons could not be removed from the system, and the entire electron transport chain would back up and stop. The mitochondria would be unable to generate new ATP in this way, and the cell would ultimately die from lack of energy. This is the reason we must breathe to draw in new oxygen.

In the electron transport chain, the free energy from the series of reactions just described is used to pump hydrogen ions across the membrane. The uneven distribution of H^+ ions across the membrane establishes an electrochemical gradient, owing to the H^+ ions' positive charge and their higher concentration on one side of the membrane.

Hydrogen ions diffuse through the inner membrane through an integral membrane protein called ATP synthase (Figure 2b). This complex protein acts as a tiny generator, turned by the force of the hydrogen ions diffusing through it, down their electrochemical gradient from the intermembrane space, where there are many mutually repelling hydrogen ions to the matrix, where there are few. The turning of the parts of this molecular machine regenerate ATP from ADP. This flow of hydrogen ions across the membrane through ATP synthase is called chemiosmosis.

Chemiosmosis (Figure 2c) is used to generate 90 percent of the ATP made during aerobic glucose catabolism. The result of the reactions is the production of ATP from the energy of the electrons removed from hydrogen atoms. These atoms were originally part of a glucose molecule. At the end of the electron transport system, the electrons are used to reduce an oxygen molecule to oxygen ions. The extra electrons on the oxygen ions attract hydrogen ions (protons) from the surrounding medium, and water is formed. The electron transport chain and the production of ATP through chemiosmosis are collectively called oxidative phosphorylation.

ATP Yield

The number of ATP molecules generated from the catabolism of glucose varies. For example, the number of hydrogen ions that the electron transport chain complexes can pump through the membrane varies between species. Another source of variance stems from the shuttle of electrons across the mitochondrial membrane. The NADH generated from glycolysis cannot easily enter mitochondria. Thus, electrons are picked up on the inside of the mitochondria by either NAD^+ or FAD^+. Fewer ATP molecules are generated when FAD^+ acts as a carrier. NAD^+ is used as the electron transporter in the liver and FAD^+ in the brain, so ATP yield depends on the tissue being considered.

Another factor that affects the yield of ATP molecules generated from glucose is that intermediate compounds in these pathways are used for other purposes. Glucose catabolism connects with the pathways that build or break down all other biochemical compounds in cells, and the result is somewhat messier than the ideal situations described thus far. For example, sugars other than glucose are fed into the glycolytic pathway for energy extraction. Other molecules that would otherwise be used to harvest energy in glycolysis or the citric acid cycle may be removed to form nucleic acids, amino acids, lipids, or other compounds. Overall, in living systems, these pathways of glucose catabolism extract about 34 percent of the energy contained in glucose.

In Summary: Citric Acid Cycle

The citric acid cycle is a series of chemical reactions that removes high-energy electrons and uses them in the electron transport chain to generate ATP. One molecule of ATP (or an equivalent) is produced per each turn of the cycle.

The electron transport chain is the portion of aerobic respiration that uses free oxygen as the final electron acceptor for electrons removed from the intermediate compounds in glucose catabolism. The electrons are passed through a series of chemical reactions, with a small amount of free energy used at three points to transport hydrogen ions across the membrane. This contributes to the gradient used in chemiosmosis. As the electrons are passed from NADH or $FADH_2$ down the electron transport chain, they lose energy. The products of the electron transport chain are water and ATP. A number of intermediate compounds can be diverted into the anabolism of other biochemical molecules, such as nucleic acids, non-essential amino acids, sugars, and lipids. These same molecules, except nucleic acids, can serve as energy sources for the glucose pathway.

Practice Questions

Cyanide inhibits cytochrome c oxidase, a component of the electron transport chain. If cyanide poisoning occurs, would you expect the pH of the intermembrane space to increase or decrease? What affect would cyanide have on ATP synthesis?

Answer

After cyanide poisoning, the electron transport chain can no longer pump electrons into the intermembrane space. The pH of the intermembrane space would increase, and ATP synthesis would stop.

We inhale oxygen when we breathe and exhale carbon dioxide. What is the oxygen used for and where does the carbon dioxide come from?

Answer

The oxygen we inhale is the final electron acceptor in the electron transport chain and allows aerobic respiration to proceed, which is the most efficient pathway for harvesting energy in the form of ATP from food molecules. The carbon dioxide we breathe out is formed during the citric acid cycle when the bonds in carbon compounds are broken.

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SUMMARY: CELLULAR RESPIRATION

Learning Outcomes

- Describe the process of glycolysis and identify its reactants and products
- Describe the process of the citric acid cycle (Krebs cycle) and identify its reactants and products
- Describe the overall outcome of the citric acid cycle and oxidative phosphorylation in terms of the products of each
- Describe the location of the citric acid cycle and oxidative phosphorylation in the cell

Cellular respiration is a process that all living things use to convert glucose into energy. Autotrophs (like plants) produce glucose during photosynthesis. Heterotrophs (like humans) ingest other living things to obtain glucose. While the process can seem complex, this page takes you through the key elements of each part of cellular respiration.

Let's Review

Cellular respiration is a collection of three unique metabolic pathways: glycolysis, the citric acid cycle, and the electron transport chain. Glycolysis is an anaerobic process, while the other two pathways are aerobic. In order to move from glycolysis to the citric acid cycle, pyruvate molecules (the output of glycolysis) must be oxidized in a process called pyruvate oxidation.

Glycolysis

Glycolysis is the first pathway in cellular respiration. This pathway is anaerobic and takes place in the cytoplasm of the cell. This pathway breaks down 1 glucose molecule and produces 2 pyruvate molecules. There are two halves of glycolysis, with five steps in each half. The first half is known as the "energy requiring" steps. This half splits glucose, and uses up 2 ATP. If the concentration of pyruvate kinase is high enough, the second half of glycolysis can proceed. In the second half, the "energy releasing: steps, 4 molecules of ATP and 2 NADH are released. Glycolysis has a **net gain** of 2 ATP molecules and 2 NADH.

Some cells (e.g., mature mammalian red blood cells) cannot undergo aerobic respiration, so glycolysis is their **only** source of ATP. However, most cells undergo pyruvate oxidation and continue to the other pathways of cellular respiration.

Pyruvate Oxidation

In eukaryotes, pyruvate oxidation takes place in the mitochondria. Pyruvate oxidation can only happen if oxygen is available. In this process, the pyruvate created by glycolysis is oxidized. In this oxidation process, a carboxyl group is removed from pyruvate, creating acetyl groups, which compound with coenzyme A (CoA) to form acetyl CoA. This process also releases CO_2.

Citric Acid Cycle

The citric acid cycle (also known as the Krebs cycle) is the second pathway in cellular respiration, and it also takes place in the mitochondria. The rate of the cycle is controlled by ATP concentration. When there is more ATP available, the rate slows down; when there is less ATP the rate increases. This pathway is a closed loop: the final step produces the compound needed for the first step.

The citric acid cycle is considered an aerobic pathway because the NADH and $FADH_2$ it produces act as temporary electron storage compounds, transferring their electrons to the next pathway (electron transport chain), which uses atmospheric oxygen. Each turn of the citric acid cycle provides a **net gain** of CO_2, 1 GTP or ATP, and 3 NADH and 1 $FADH_2$.

Electron Transport Chain

Most ATP from glucose is generated in the electron transport chain. It is the only part of cellular respiration that directly consumes oxygen; however, in some prokaryotes, this is an anaerobic pathway. In eukaryotes, this pathway takes place in the inner mitochondrial membrane. In prokaryotes it occurs in the plasma membrane.

The electron transport chain is made up of 4 proteins along the membrane and a proton pump. A cofactor shuttles electrons between proteins I–III. If NAD is depleted, skip I: $FADH_2$ starts on II. In chemiosmosis, a proton pump takes hydrogens from inside mitochondria to the outside; this spins the "motor" and the phosphate groups attach to that. The movement changes from ADP to ATP, creating 90% of ATP obtained from aerobic glucose catabolism.

Let's Practice

Now that you've reviewed cellular respiration, this practice activity will help you see how well you know cellular respiration:

Click here for a text-only version of the activity.

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INTRODUCTION TO FERMENTATION

What you'll learn to do: Illustrate the basic components and steps of fermentation.

The final metabolic pathway we'll discuss is fermentation. This is an anaerobic process (it occurs without oxygen).

You're most likely familiar with the idea that alcohol is created through a process called **fermentation**. However, you may not be familiar with just how this process works. Another type of fermentation—called lactic acid fermentation—takes place in the bodies of animals and some bacteria. Humans gain valuable products from both types of fermentation. Alcohol fermentation creates breads, beer, wine, and spirits for us. Lactic acid fermentation is used in making dairy based products such as yogurt.

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TYPES OF FERMENTATION

Learning Outcomes

- Describe the process of lactic acid fermentation
- Describe the process of alcohol fermentation

Lactic Acid Fermentation

The fermentation method used by animals and certain bacteria, like those in yogurt, is **lactic acid fermentation** (Figure 1). This type of fermentation is used routinely in mammalian red blood cells and in skeletal muscle that has an insufficient oxygen supply to allow aerobic respiration to continue (that is, in muscles used to the point of

fatigue). In muscles, lactic acid accumulation must be removed by the blood circulation and the lactate brought to the liver for further metabolism. The chemical reactions of lactic acid fermentation are the following:

$$\text{Pyruvic acid} + \text{NADH} \longleftrightarrow \text{lactic acid} + \text{NAD}^+$$

The enzyme used in this reaction is lactate dehydrogenase (LDH). The reaction can proceed in either direction, but the reaction from left to right is inhibited by acidic conditions. Such lactic acid accumulation was once believed to cause muscle stiffness, fatigue, and soreness, although more recent research disputes this hypothesis. Once the lactic acid has been removed from the muscle and circulated to the liver, it can be reconverted into pyruvic acid and further catabolized for energy.

Practice Question

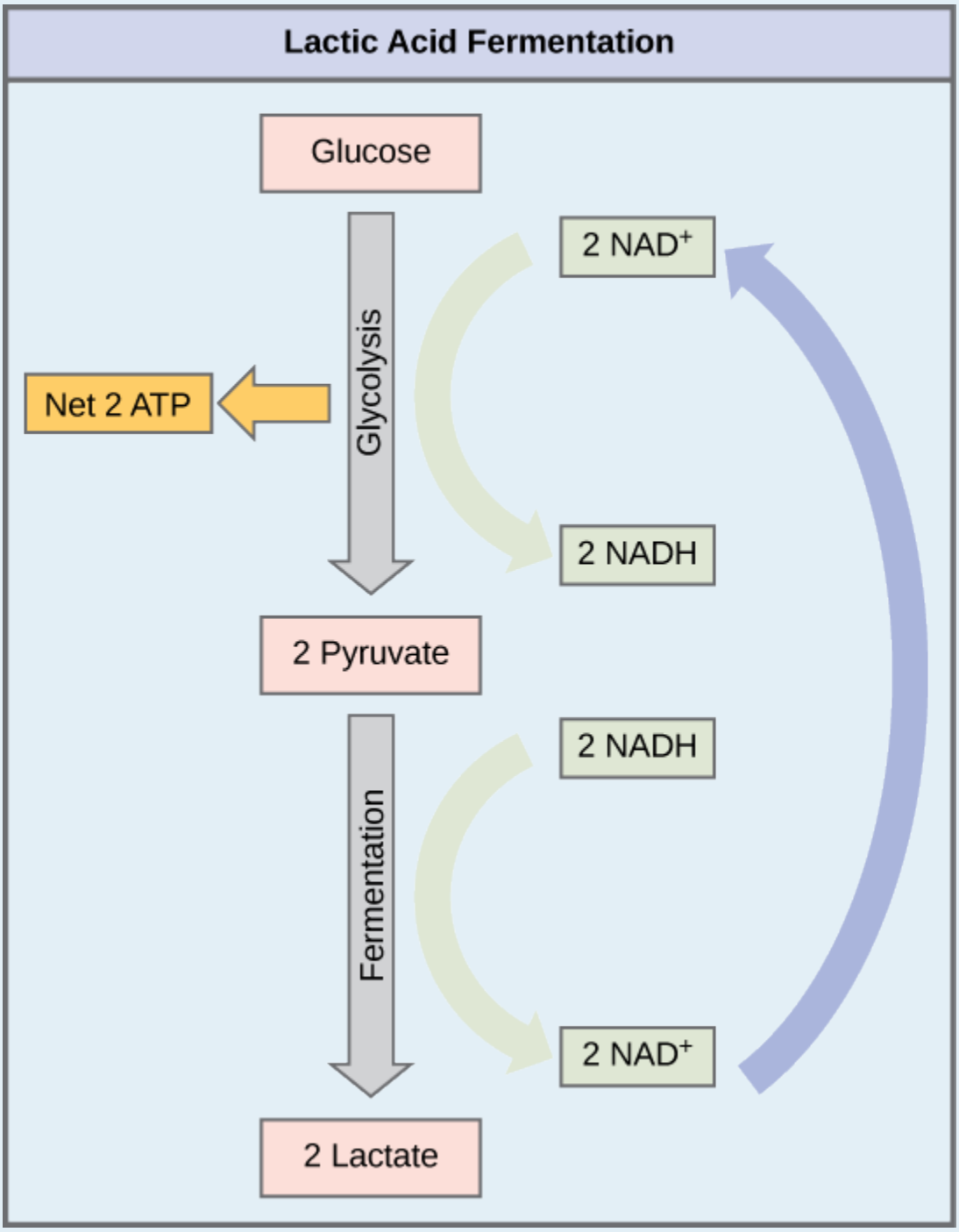

Figure 1. Lactic acid fermentation is common in muscle cells that have run out of oxygen.

Tremetol, a metabolic poison found in the white snake root plant, prevents the metabolism of lactate. When cows eat this plant, it is concentrated in the milk they produce. Humans who consume the milk become ill. Symptoms of this disease, which include vomiting, abdominal pain, and tremors, become worse after exercise. Why do you think this is the case?

Answer

The illness is caused by lactate accumulation. Lactate levels rise after exercise, making the symptoms worse. Milk sickness is rare today, but was common in the Midwestern United States in the early 1800s.

Alcohol Fermentation

Another familiar fermentation process is **alcohol fermentation** (Figure 3) that produces ethanol, an alcohol (because of this, this kind of fermentation is also sometimes known as **ethanol fermentation**). There are two main reactions in alcohol fermentation.

Figure 2. Fermentation of grape juice into wine produces CO_2 as a byproduct. Fermentation tanks have valves so that the pressure inside the tanks created by the carbon dioxide produced can be released.

The first reaction is catalyzed by pyruvate decarboxylase, a cytoplasmic enzyme, with a coenzyme of thiamine pyrophosphate (TPP, derived from vitamin B1 and also called thiamine). A carboxyl group is removed from pyruvic acid, releasing carbon dioxide as a gas. The loss of carbon dioxide reduces the size of the molecule by one carbon, making acetaldehyde.

The second reaction is catalyzed by alcohol dehydrogenase to oxidize NADH to NAD^+ and reduce acetaldehyde to ethanol. The fermentation of pyruvic acid by yeast produces the ethanol found in alcoholic beverages. Ethanol tolerance of yeast is variable, ranging from about 5 percent to 21 percent, depending on the yeast strain and environmental conditions.

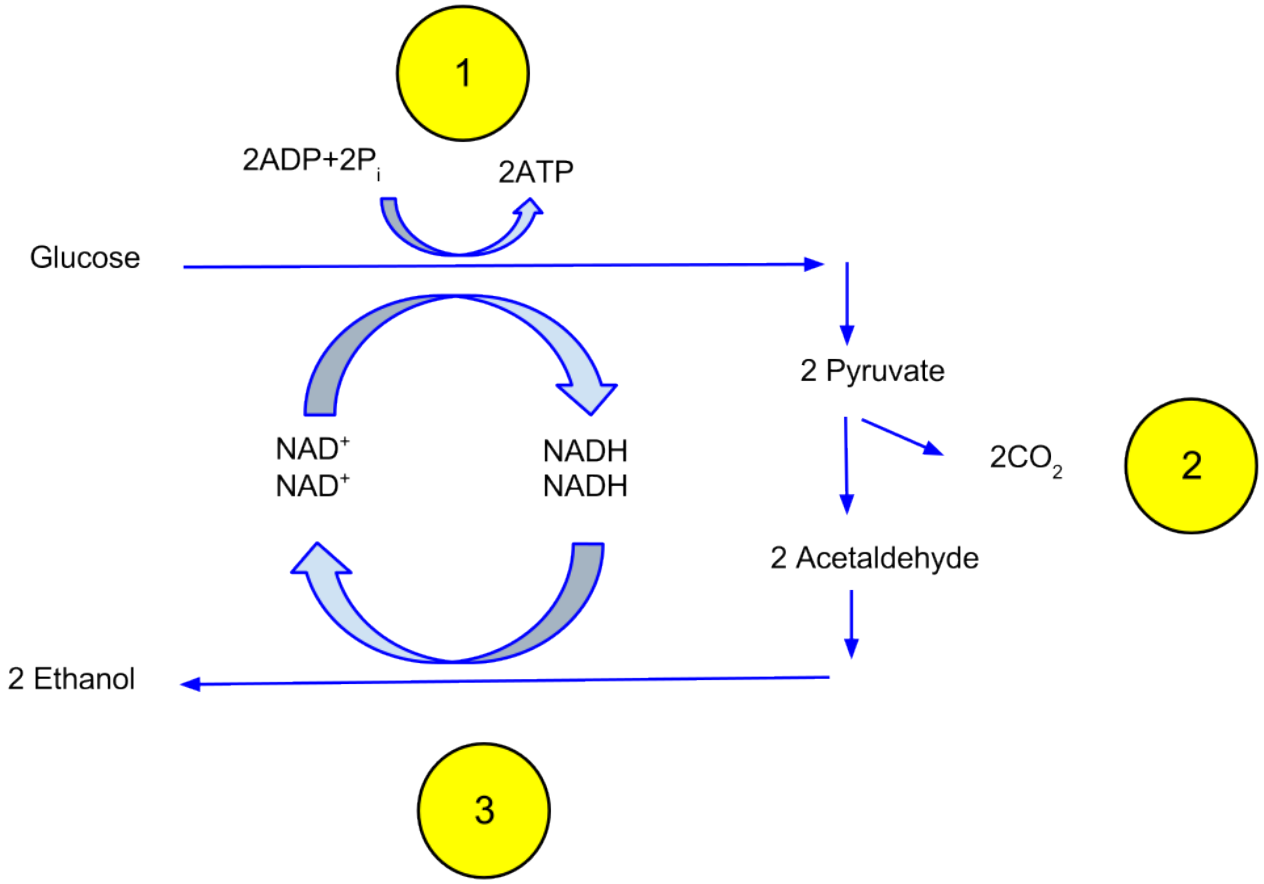

Figure 3. Diagram of alcohol fermentation

Other Types of Fermentation

Other fermentation methods occur in bacteria. Many prokaryotes are facultatively anaerobic. This means that they can switch between aerobic respiration and fermentation, depending on the availability of oxygen. Certain prokaryotes, like *Clostridia*, are obligate anaerobes. Obligate anaerobes live and grow in the absence of molecular oxygen. Oxygen is a poison to these microorganisms and kills them on exposure.

It should be noted that all forms of fermentation, except lactic acid fermentation, produce gas. The production of particular types of gas is used as an indicator of the fermentation of specific carbohydrates, which plays a role in the laboratory identification of the bacteria. Various methods of fermentation are used by assorted organisms to ensure an adequate supply of NAD^+ for the sixth step in glycolysis. Without these pathways, that step would not occur and no ATP would be harvested from the breakdown of glucose.

In Summary: Types of Fermentation

If NADH cannot be metabolized through aerobic respiration, another electron acceptor is used. Most organisms will use some form of fermentation to accomplish the regeneration of NAD^+, ensuring the continuation of glycolysis. The regeneration of NAD^+ in fermentation is not accompanied by ATP production; therefore, the potential for NADH to produce ATP using an electron transport chain is not utilized.

Practice Question

Tremetol, a metabolic poison found in white snake root plant, prevents the metabolism of lactate. When cows eat this plant, Tremetol is concentrated in the milk. Humans who consume the milk become ill. Symptoms of this disease, which include vomiting, abdominal pain, and tremors, become worse after exercise. Why do you think this is the case?

Answer

The illness is caused by lactic acid build-up. Lactic acid levels rise after exercise, making the symptoms worse. Milk sickness is rare today, but was common in the Midwestern United States in the early 1800s.

Practice Question

When muscle cells run out of oxygen, what happens to the potential for energy extraction from sugars and what pathways do the cell use?

Answer

Without oxygen, oxidative phosphorylation and the citric acid cycle stop, so ATP is no longer generated through this mechanism, which extracts the greatest amount of energy from a sugar molecule. In addition, NADH accumulates, preventing glycolysis from going forward because of an absence of NAD^+. Lactic acid fermentation uses the electrons in NADH to generate lactic acid from pyruvate, which allows glycolysis to continue and thus a smaller amount of ATP can be generated by the cell.

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INTRODUCTION TO PHOTOSYNTHESIS

What you'll learn to do: Identify the basic components and steps of photosynthesis

No matter how complex or advanced a machine, such as the latest cellular phone, the device cannot function without energy. Living things, similar to machines, have many complex components; they too cannot do anything without energy, which is why humans and all other organisms must "eat" in some form or another. That may be common knowledge, but how many people realize that every bite of every meal ingested depends on the process of photosynthesis?

Figure 1. This sage thrasher's diet, like that of almost all organisms, depends on photosynthesis. (credit: modification of work by Dave Menke, U.S. Fish and Wildlife Service)

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AN OVERVIEW OF PHOTOSYNTHESIS

Learning Outcomes

Summarize the process of photosynthesis

All living organisms on earth consist of one or more cells. Each cell runs on the chemical energy found mainly in carbohydrate molecules (food), and the majority of these molecules are produced by one process: photosynthesis. Through photosynthesis, certain organisms convert solar energy (sunlight) into chemical energy, which is then used to build carbohydrate molecules. The energy used to hold these molecules together is released when an organism breaks down food. Cells then use this energy to perform work, such as cellular respiration.

The energy that is harnessed from photosynthesis enters the ecosystems of our planet continuously and is transferred from one organism to another. Therefore, directly or indirectly, the process of photosynthesis provides most of the energy required by living things on earth.

Photosynthesis also results in the release of oxygen into the atmosphere. In short, to eat and breathe, humans depend almost entirely on the organisms that carry out photosynthesis.

Learn more about photosynthesis

Solar Dependence and Food Production

Some organisms can carry out photosynthesis, whereas others cannot. An autotroph is an organism that can produce its own food. The Greek roots of the word *autotroph* mean “self” (*auto*) “feeder” (*troph*). Plants are the best-known autotrophs, but others exist, including certain types of bacteria and algae (Figure 1). Oceanic algae contribute enormous quantities of food and oxygen to global food chains. Plants are also photoautotrophs, a type of autotroph that uses sunlight and carbon from carbon dioxide to synthesize chemical energy in the form of carbohydrates. All organisms carrying out photosynthesis require sunlight.

Figure 1. (a) Plants, (b) algae, and (c) certain bacteria, called cyanobacteria, are photoautotrophs that can carry out photosynthesis. Algae can grow over enormous areas in water, at times completely covering the surface. (credit a: Steve Hillebrand, U.S. Fish and Wildlife Service; credit b: “eutrophication&hypoxia”/Flickr; credit c: NASA; scale-bar data from Matt Russell)

Heterotrophs are organisms incapable of photosynthesis that must therefore obtain energy and carbon from food by consuming other organisms. The Greek roots of the word *heterotroph* mean “other” (*hetero*) “feeder” (*troph*), meaning that their food comes from other organisms. Even if the food organism is another animal, this food traces its origins back to autotrophs and the process of photosynthesis. Humans are heterotrophs, as are all animals. Heterotrophs depend on autotrophs, either directly or indirectly. Deer and wolves are heterotrophs. A deer obtains energy by eating plants. A wolf eating a deer obtains energy that originally came from the plants eaten by that deer. The energy in the plant came from photosynthesis, and therefore it is the only autotroph in this example (Figure 2). Using this reasoning, all food eaten by humans also links back to autotrophs that carry out photosynthesis.

Figure 2. The energy stored in carbohydrate molecules from photosynthesis passes through the food chain. The predator that eats these deer is getting energy that originated in the photosynthetic vegetation that the deer consumed. (credit: Steve VanRiper, U.S. Fish and Wildlife Service)

Photosynthesis is a multi-step process that requires sunlight, carbon dioxide (which is low in energy), and water as substrates (Figure 3). After the process is complete, it releases oxygen and produces glyceraldehyde-3-phosphate (GA3P), simple carbohydrate molecules (which are high in energy) that can subsequently be converted into glucose, sucrose, or any of dozens of other sugar molecules. These sugar molecules contain energy and the energized carbon that all living things need to survive.

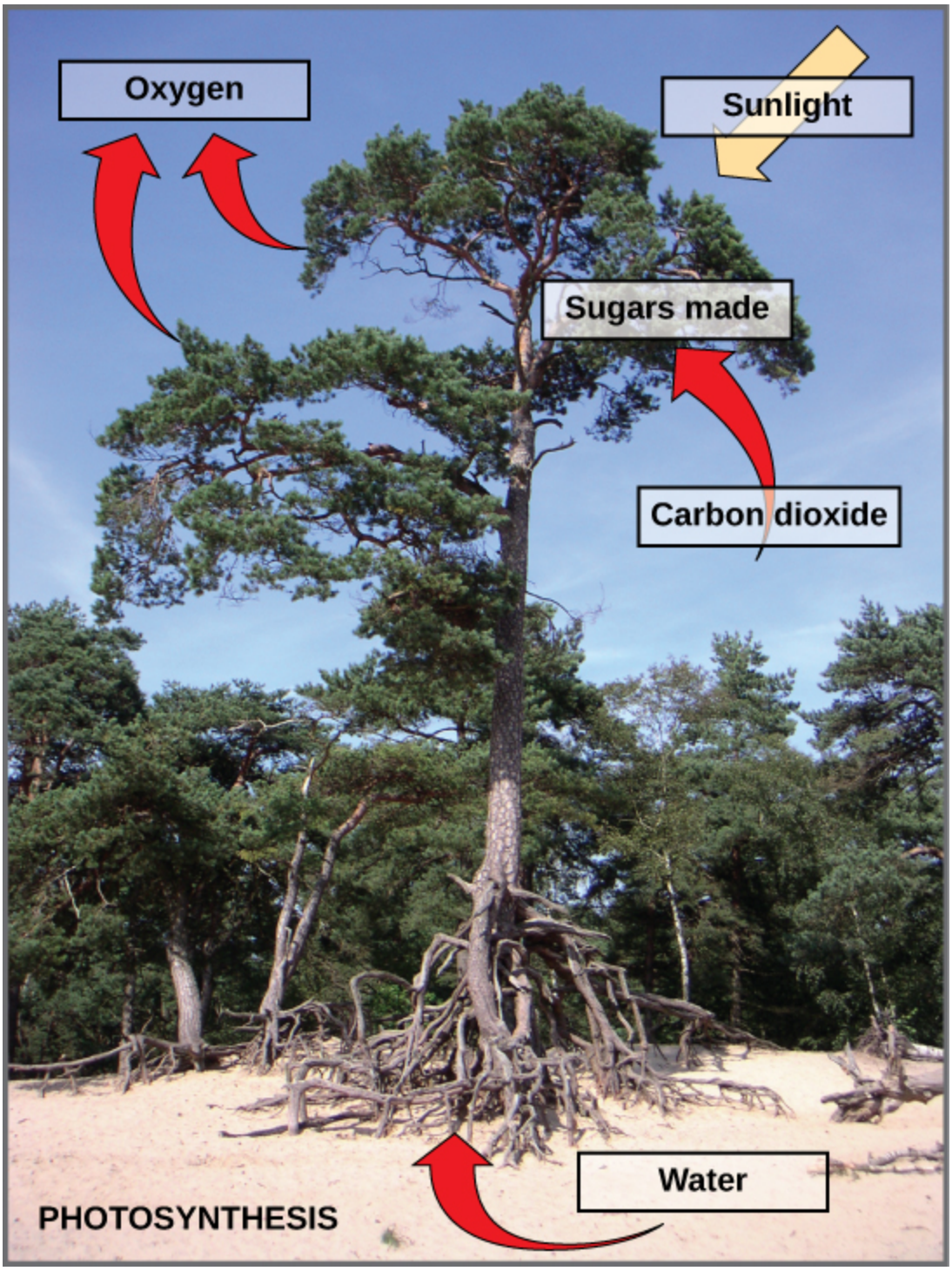

Figure 3. Photosynthesis uses solar energy, carbon dioxide, and water to produce energy-storing carbohydrates. Oxygen is generated as a waste product of photosynthesis.

The following is the chemical equation for photosynthesis (Figure 4):

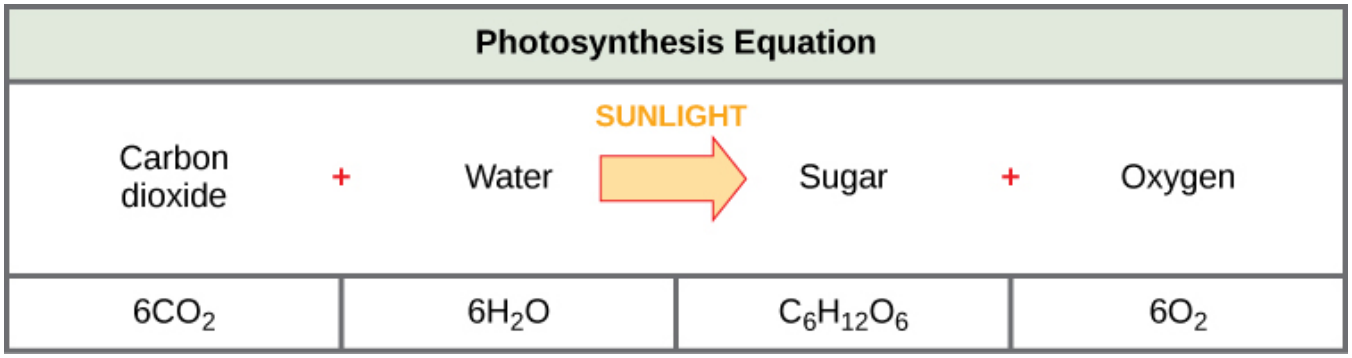

Figure 4. The basic equation for photosynthesis is deceptively simple. In reality, the process takes place in many steps involving intermediate reactants and products. Glucose, the primary energy source in cells, is made from two three-carbon GA3Ps.

Although the equation looks simple, the many steps that take place during photosynthesis are actually quite complex. Before learning the details of how photoautotrophs turn sunlight into food, it is important to become familiar with the structures involved.

In plants, photosynthesis generally takes place in leaves, which consist of several layers of cells. The process of photosynthesis occurs in a middle layer called the **mesophyll**. The gas exchange of carbon dioxide and oxygen occurs through small, regulated openings called **stomata** (singular: stoma), which also play roles in the regulation of gas exchange and water balance. The stomata are typically located on the underside of the leaf, which helps to minimize water loss. Each stoma is flanked by guard cells that regulate the opening and closing of the stomata by swelling or shrinking in response to osmotic changes.

In all autotrophic eukaryotes, photosynthesis takes place inside an organelle called a **chloroplast**. For plants, chloroplast-containing cells exist in the mesophyll. Chloroplasts have a double membrane envelope (composed of an outer membrane and an inner membrane). Within the chloroplast are stacked, disc-shaped structures called **thylakoids**. Embedded in the thylakoid membrane is chlorophyll, a **pigment** (molecule that absorbs light) responsible for the initial interaction between light and plant material, and numerous proteins that make up the electron transport chain. The thylakoid membrane encloses an internal space called the **thylakoid lumen**. As shown in Figure 5, a stack of thylakoids is called a **granum**, and the liquid-filled space surrounding the granum is called **stroma** or “bed” (not to be confused with stoma or “mouth,” an opening on the leaf epidermis).

Practice Question

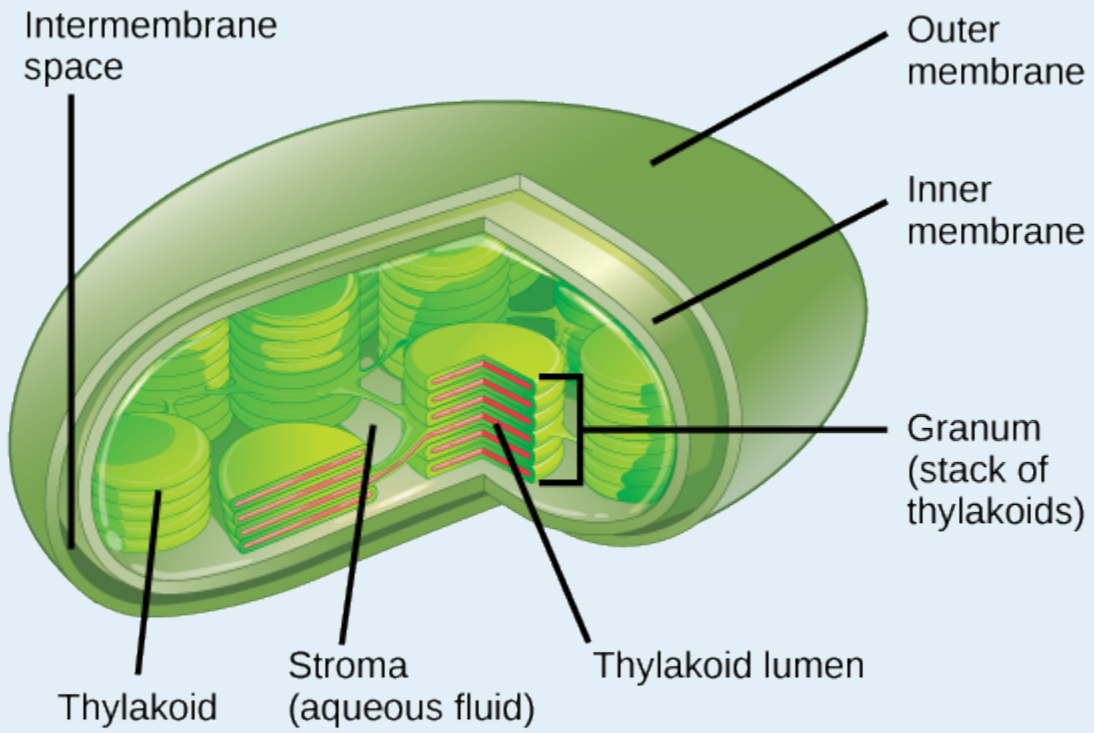

Figure 5. Photosynthesis takes place in chloroplasts, which have an outer membrane and an inner membrane. Stacks of thylakoids called grana form a third membrane layer.

On a hot, dry day, plants close their stomata to conserve water. What impact will this have on photosynthesis?

Answer

Levels of carbon dioxide (a necessary photosynthetic substrate) will immediately fall. As a result, the rate of photosynthesis will be inhibited.

The Two Parts of Photosynthesis

Photosynthesis takes place in two sequential stages: the light-dependent reactions and the light independent-reactions. In the **light-dependent reactions**, energy from sunlight is absorbed by chlorophyll and that energy is converted into stored chemical energy. In the **light-independent reactions**, the chemical energy harvested during the light-dependent reactions drive the assembly of sugar molecules from carbon dioxide. Therefore, although the light-independent reactions do not use light as a reactant, they require the products of the light-dependent reactions to function. In addition, several enzymes of the light-independent reactions are activated by light. The light-dependent reactions utilize certain molecules to temporarily store the energy: These are referred to as energy carriers. The energy carriers that move energy from light-dependent reactions to light-independent reactions can be thought of as “full” because they are rich in energy. After the energy is released, the “empty” energy carriers return to the light-dependent reaction to obtain more energy. Figure 6 illustrates the components inside the chloroplast where the light-dependent and light-independent reactions take place.

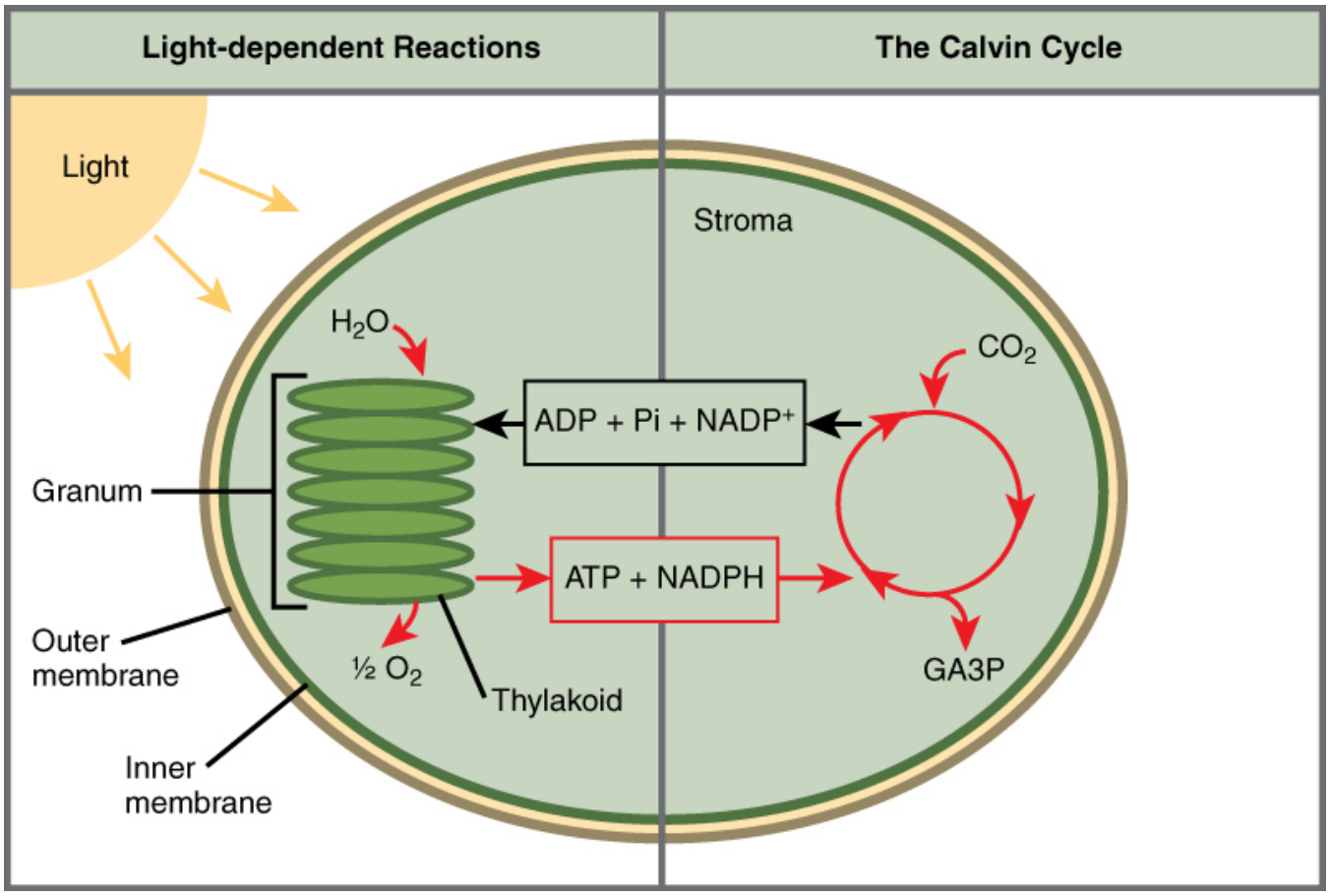

Figure 6. Photosynthesis takes place in two stages: light dependent reactions and the Calvin cycle. Light-dependent reactions, which take place in the thylakoid membrane, use light energy to make ATP and NADPH. The Calvin cycle, which takes place in the stroma, uses energy derived from these compounds to make GA3P from CO_2.

Click the link to learn more about photosynthesis.

Photosynthesis at the Grocery Store

Major grocery stores in the United States are organized into departments, such as dairy, meats, produce, bread, cereals, and so forth. Each aisle (Figure 7) contains hundreds, if not thousands, of different products for customers to buy and consume.

Although there is a large variety, each item links back to photosynthesis. Meats and dairy link because the animals were fed plant-based foods. The breads, cereals, and pastas come largely from starchy grains, which are the seeds of photosynthesis-dependent plants. What about desserts and drinks? All of these products contain

sugar—sucrose is a plant product, a disaccharide, a carbohydrate molecule, which is built directly from photosynthesis. Moreover, many items are less obviously derived from plants: for instance, paper goods are generally plant products, and many plastics (abundant as products and packaging) can be derived from algae or from oil, the fossilized remains of photosynthetic organisms. Virtually every spice and flavoring in the spice aisle was produced by a plant as a leaf, root, bark, flower, fruit, or stem. Ultimately, photosynthesis connects to every meal and every food a person consumes.

Figure 7. Foods that humans consume originate from photosynthesis. (credit: Associação Brasileira de Supermercados)

In Summary: An Overview of Photosynthesis

The process of photosynthesis transformed life on Earth. By harnessing energy from the sun, photosynthesis evolved to allow living things access to enormous amounts of energy. Because of photosynthesis, living things gained access to sufficient energy that allowed them to build new structures and achieve the biodiversity evident today.

Only certain organisms, called photoautotrophs, can perform photosynthesis; they require the presence of chlorophyll, a specialized pigment that absorbs certain portions of the visible spectrum and can capture energy from sunlight. Photosynthesis uses carbon dioxide and water to assemble carbohydrate molecules and release oxygen as a waste product into the atmosphere. Eukaryotic autotrophs, such as plants and algae, have organelles called chloroplasts in which photosynthesis takes place, and starch accumulates. In prokaryotes, such as cyanobacteria, the process is less localized and occurs within folded membranes, extensions of the plasma membrane, and in the cytoplasm.

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LIGHT ENERGY

Learning Outcomes

Describe how the wavelength of light affects its energy and color

How can light be used to make food? It is easy to think of light as something that exists and allows living organisms, such as humans, to see, but light is a form of energy. Like all energy, light can travel, change form, and be harnessed to do work. In the case of photosynthesis, light energy is transformed into chemical energy, which autotrophs use to build carbohydrate molecules. However, autotrophs only use a specific component of sunlight (Figure 1).

Figure 1. Autotrophs can capture light energy from the sun, converting it into chemical energy used to build food molecules. (credit: modification of work by Gerry Atwell, U.S. Fish and Wildlife Service)

What Is Light Energy?

The sun emits an enormous amount of electromagnetic radiation (solar energy). Humans can see only a fraction of this energy, which is referred to as “visible light.” The manner in which solar energy travels can be described and measured as waves. Scientists can determine the amount of energy of a wave by measuring its wavelength, the distance between two consecutive, similar points in a series of waves, such as from crest to crest or trough to trough (Figure 2).

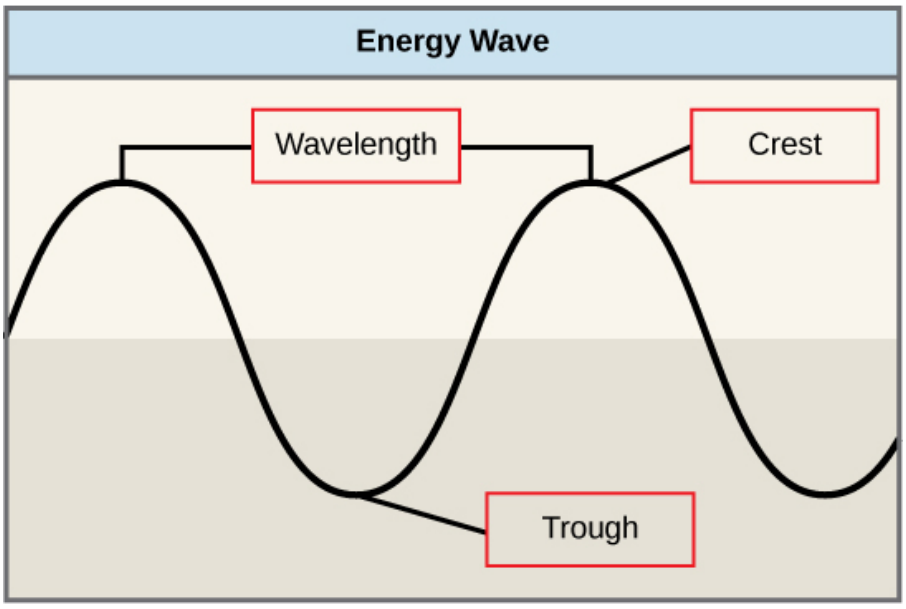

Figure 2. The wavelength of a single wave is the distance between two consecutive points along the wave.

Visible light constitutes only one of many types of electromagnetic radiation emitted from the sun. The electromagnetic spectrum is the range of all possible wavelengths of radiation (Figure 3). Each wavelength corresponds to a different amount of energy carried.

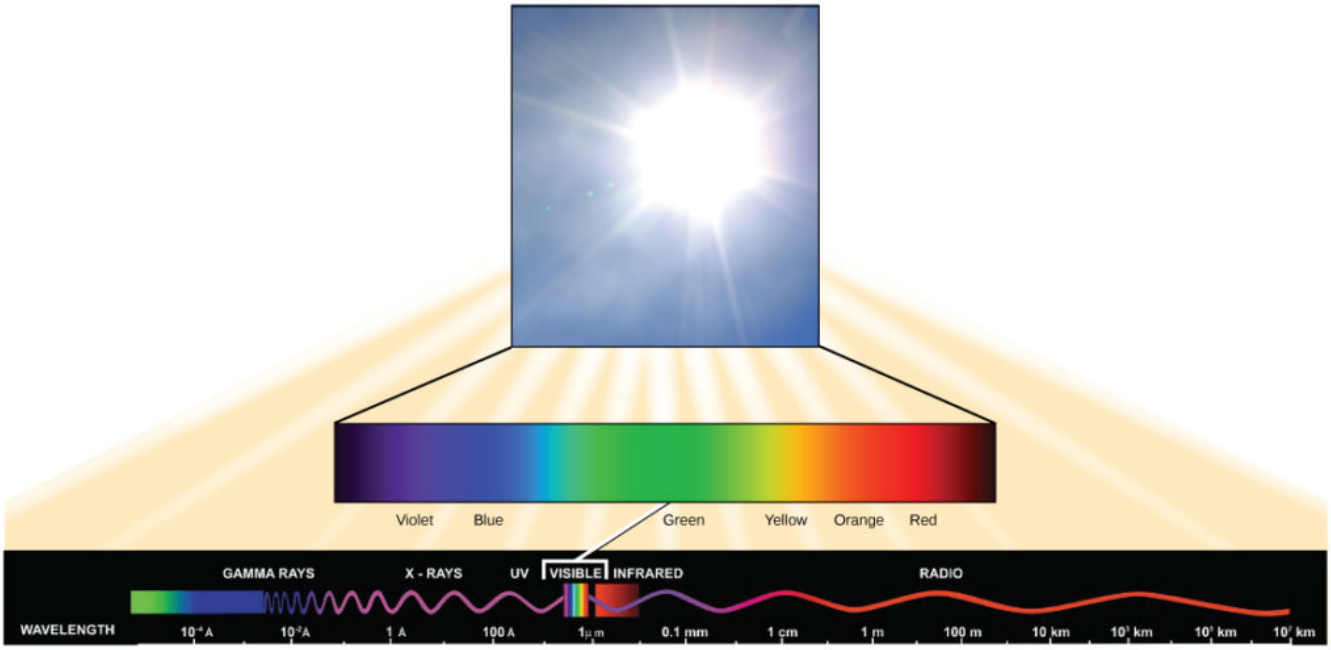

Figure 3. The sun emits energy in the form of electromagnetic radiation. This radiation exists in different wavelengths, each of which has its own characteristic energy. Visible light is one type of energy emitted from the sun.

Each type of electromagnetic radiation has a characteristic range of wavelengths. The longer the wavelength (or the more stretched out it appears), the less energy is carried. Short, tight waves carry the most energy. This may seem illogical, but think of it in terms of a piece of moving rope. It takes little effort by a person to move a rope in long, wide waves. To make a rope move in short, tight waves, a person would need to apply significantly more energy.

The sun emits (Figure 3) a broad range of electromagnetic radiation, including X-rays and ultraviolet (UV) rays. The higher-energy waves are dangerous to living things; for example, X-rays and UV rays can be harmful to humans.

Absorption of Light

Light energy enters the process of photosynthesis when pigments absorb the light. In plants, pigment molecules absorb only visible light for photosynthesis. The visible light seen by humans as white light actually exists in a rainbow of colors. Certain objects, such as a prism or a drop of water, disperse white light to reveal these colors to the human eye. The visible light portion of the electromagnetic spectrum is perceived by the human eye as a rainbow of colors, with violet and blue having shorter wavelengths and, therefore, higher energy. At the other end of the spectrum toward red, the wavelengths are longer and have lower energy.

Understanding Pigments

Different kinds of pigments exist, and each absorbs only certain wavelengths (colors) of visible light. Pigments reflect the color of the wavelengths that they cannot absorb.

All photosynthetic organisms contain a pigment called chlorophyll *a*, which humans see as the common green color associated with plants. Chlorophyll *a* absorbs wavelengths from either end of the visible spectrum (blue and red), but not from green. Because green is reflected, chlorophyll appears green.

Other pigment types include chlorophyll *b* (which absorbs blue and red-orange light) and the carotenoids. Each type of pigment can be identified by the specific pattern of wavelengths it absorbs from visible light, which is its absorption spectrum.

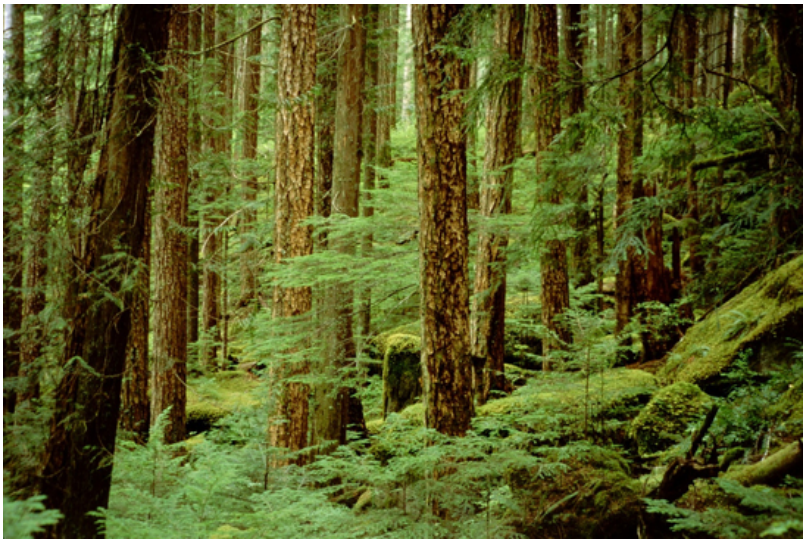

Figure 4. Plants that commonly grow in the shade benefit from having a variety of light-absorbing pigments. Each pigment can absorb different wavelengths of light, which allows the plant to absorb any light that passes through the taller trees. (credit: Jason Hollinger)

Many photosynthetic organisms have a mixture of pigments; between them, the organism can absorb energy from a wider range of visible-light wavelengths. Not all photosynthetic organisms have full access to sunlight. Some organisms grow underwater where light intensity decreases with depth, and certain wavelengths are absorbed by the water. Other organisms grow in competition for light. Plants on the rainforest floor must be able to absorb any bit of light that comes through, because the taller trees block most of the sunlight (Figure 4).

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THE LIGHT-DEPENDENT REACTIONS OF PHOTOSYNTHESIS

Learning Outcomes

Describe the light-dependent reactions that take place during photosynthesis

The overall purpose of the light-dependent reactions is to convert light energy into chemical energy. This chemical energy will be used by the Calvin cycle to fuel the assembly of sugar molecules.

The light-dependent reactions begin in a grouping of pigment molecules and proteins called a photosystem. Photosystems exist in the membranes of thylakoids. A pigment molecule in the photosystem absorbs one photon, a quantity or “packet” of light energy, at a time.

A photon of light energy travels until it reaches a molecule of chlorophyll. The photon causes an electron in the chlorophyll to become “excited.” The energy given to the electron allows it to break free from an atom of the chlorophyll molecule. Chlorophyll is therefore said to “donate” an electron (Figure 1).

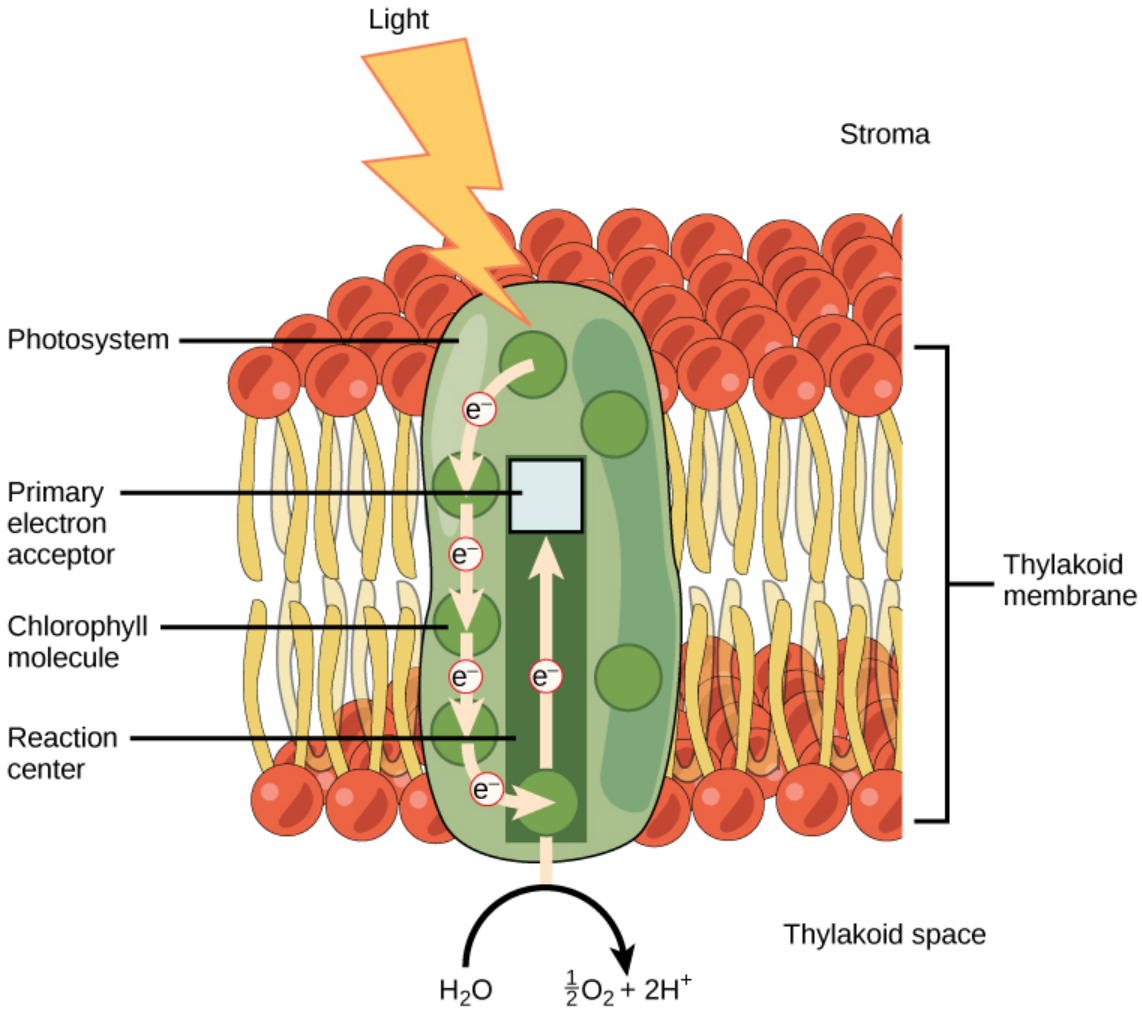

Figure 1. Light energy is absorbed by a chlorophyll molecule and is passed along a pathway to other chlorophyll molecules. The energy culminates in a molecule of chlorophyll found in the reaction center. The energy “excites” one of its electrons enough to leave the molecule and be transferred to a nearby primary electron acceptor. A molecule of water splits to release an electron, which is needed to replace the one donated. Oxygen and hydrogen ions are also formed from the splitting of water.

To replace the electron in the chlorophyll, a molecule of water is split. This splitting releases an electron and results in the formation of oxygen (O_2) and hydrogen ions (H^+) in the thylakoid space. Technically, each breaking of a water molecule releases a pair of electrons, and therefore can replace two donated electrons.

The replacing of the electron enables chlorophyll to respond to another photon. The oxygen molecules produced as byproducts find their way to the surrounding environment. The hydrogen ions play critical roles in the remainder of the light-dependent reactions.

Keep in mind that the purpose of the light-dependent reactions is to convert solar energy into chemical carriers that will be used in the Calvin cycle. In eukaryotes, two photosystems exist, the first is called photosystem II, which is named for the order of its discovery rather than for the order of function.

After the photon hits, photosystem II transfers the free electron to the first in a series of proteins inside the thylakoid membrane called the electron transport chain. As the electron passes along these proteins, energy from the electron fuels membrane pumps that actively move hydrogen ions against their concentration gradient from the stroma into the thylakoid space. This is quite analogous to the process that occurs in the mitochondrion in

which an electron transport chain pumps hydrogen ions from the mitochondrial stroma across the inner membrane and into the intermembrane space, creating an electrochemical gradient. After the energy is used, the electron is accepted by a pigment molecule in the next photosystem, which is called photosystem I (Figure 2).

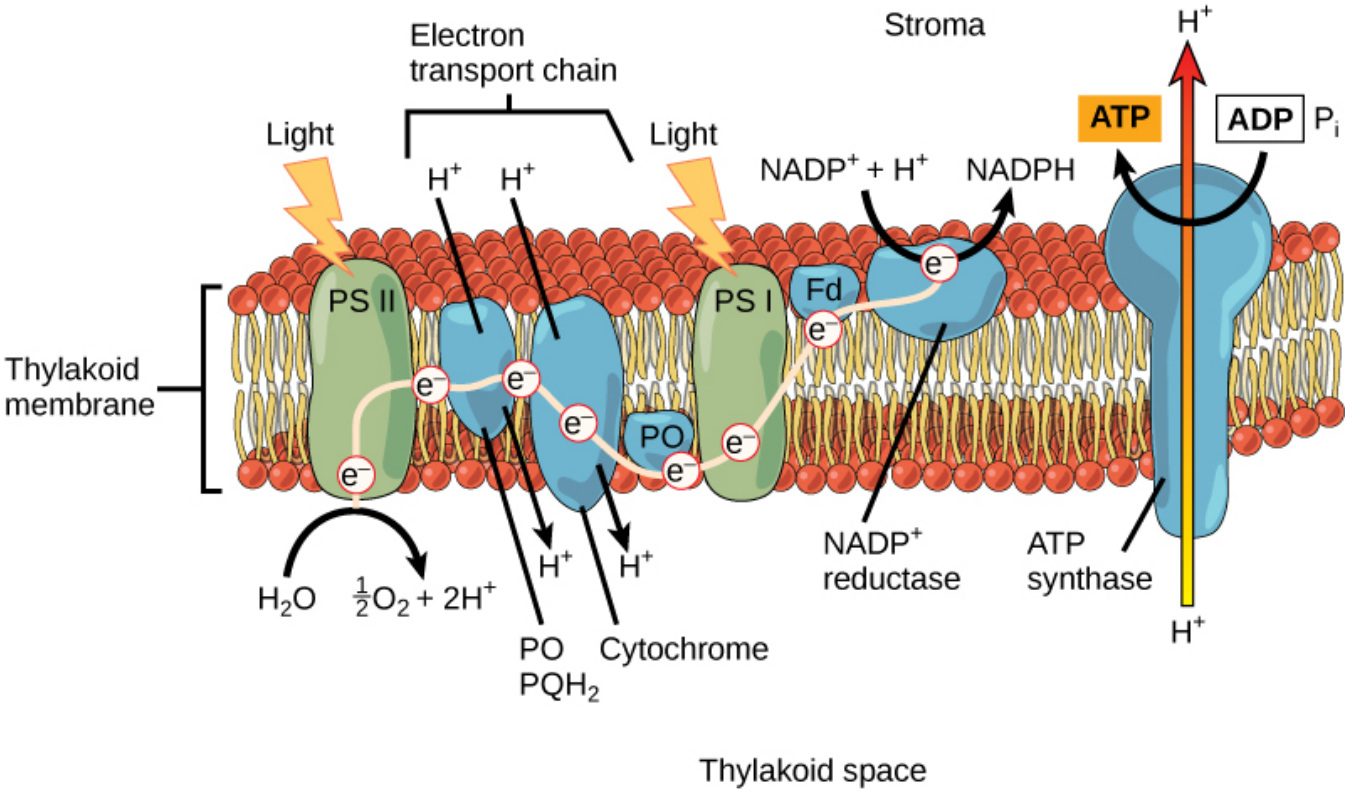

Figure 2. From photosystem II, the excited electron travels along a series of proteins. This electron transport system uses the energy from the electron to pump hydrogen ions into the interior of the thylakoid. A pigment molecule in photosystem I accepts the electron.

Generating an Energy Carrier: ATP

In the light-dependent reactions, energy absorbed by sunlight is stored by two types of energy-carrier molecules: ATP and NADPH. The energy that these molecules carry is stored in a bond that holds a single atom to the molecule. For ATP, it is a phosphate atom, and for NADPH, it is a hydrogen atom. NADH will be discussed further in relation to cellular respiration, which occurs in the mitochondrion, where it carries energy from the citric acid cycle to the electron transport chain. When these molecules release energy into the Calvin cycle, they each lose atoms to become the lower-energy molecules ADP and $NADP^+$.

The buildup of hydrogen ions in the thylakoid space forms an electrochemical gradient because of the difference in the concentration of protons (H^+) and the difference in the charge across the membrane that they create. This potential energy is harvested and stored as chemical energy in ATP through chemiosmosis, the movement of hydrogen ions down their electrochemical gradient through the transmembrane enzyme ATP synthase, just as in the mitochondrion.

The hydrogen ions are allowed to pass through the thylakoid membrane through an embedded protein complex called ATP synthase. This same protein generated ATP from ADP in the mitochondrion. The energy generated by the hydrogen ion stream allows ATP synthase to attach a third phosphate to ADP, which forms a molecule of ATP in a process called photophosphorylation. The flow of hydrogen ions through ATP synthase is called chemiosmosis, because the ions move from an area of high to low concentration through a semi-permeable structure.

Generating Another Energy Carrier: NADPH

The remaining function of the light-dependent reaction is to generate the other energy-carrier molecule, NADPH. As the electron from the electron transport chain arrives at photosystem I, it is re-energized with another photon captured by chlorophyll. The energy from this electron drives the formation of NADPH from $NADP^+$ and a hydrogen ion (H^+). Now that the solar energy is stored in energy carriers, it can be used to make a sugar molecule.

In Summary: The Light-Dependent Reactions of Photosynthesis

In the first part of photosynthesis, the light-dependent reaction, pigment molecules absorb energy from sunlight. The most common and abundant pigment is chlorophyll *a*. A photon strikes photosystem II to initiate photosynthesis. Energy travels through the electron transport chain, which pumps hydrogen ions into the thylakoid space. This forms an electrochemical gradient. The ions flow through ATP synthase from the thylakoid space into the stroma in a process called chemiosmosis to form molecules of ATP, which are used for the formation of sugar molecules in the second stage of photosynthesis. Photosystem I absorbs a second photon, which results in the formation of an NADPH molecule, another energy carrier for the Calvin cycle reactions.

Practice Question

Describe the pathway of energy in light-dependent reactions.

Answer

The energy is present initially as light. A photon of light hits chlorophyll, causing an electron to be energized. The free electron travels through the electron transport chain, and the energy of the electron is used to pump hydrogen ions into the thylakoid space, transferring the energy into the electrochemical gradient. The energy of the electrochemical gradient is used to power ATP synthase, and the energy is transferred into a bond in the ATP molecule. In addition, energy from another photon can be used to create a high-energy bond in the molecule NADPH.

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THE CALVIN CYCLE

Learning Outcomes

Describe the steps and processes in the Calvin Cycle

After the energy from the sun is converted and packaged into ATP and NADPH, the cell has the fuel needed to build food in the form of carbohydrate molecules. The carbohydrate molecules made will have a backbone of carbon atoms. Where does the carbon come from? The carbon atoms used to build carbohydrate molecules

comes from carbon dioxide, the gas that animals exhale with each breath. The Calvin cycle is the term used for the reactions of photosynthesis that use the energy stored by the light-dependent reactions to form glucose and other carbohydrate molecules. This process may also be called the light-independent reaction, as it does not directly require sunlight (but it does require the products produced from the light-dependent reactions).

The Innerworkings of the Calvin Cycle

In plants, carbon dioxide (CO_2) enters the chloroplast through the stomata and diffuses into the stroma of the chloroplast—the site of the Calvin cycle reactions where sugar is synthesized. The reactions are named after the scientist who discovered them, and reference the fact that the reactions function as a cycle. Others call it the Calvin-Benson cycle to include the name of another scientist involved in its discovery (Figure 1).

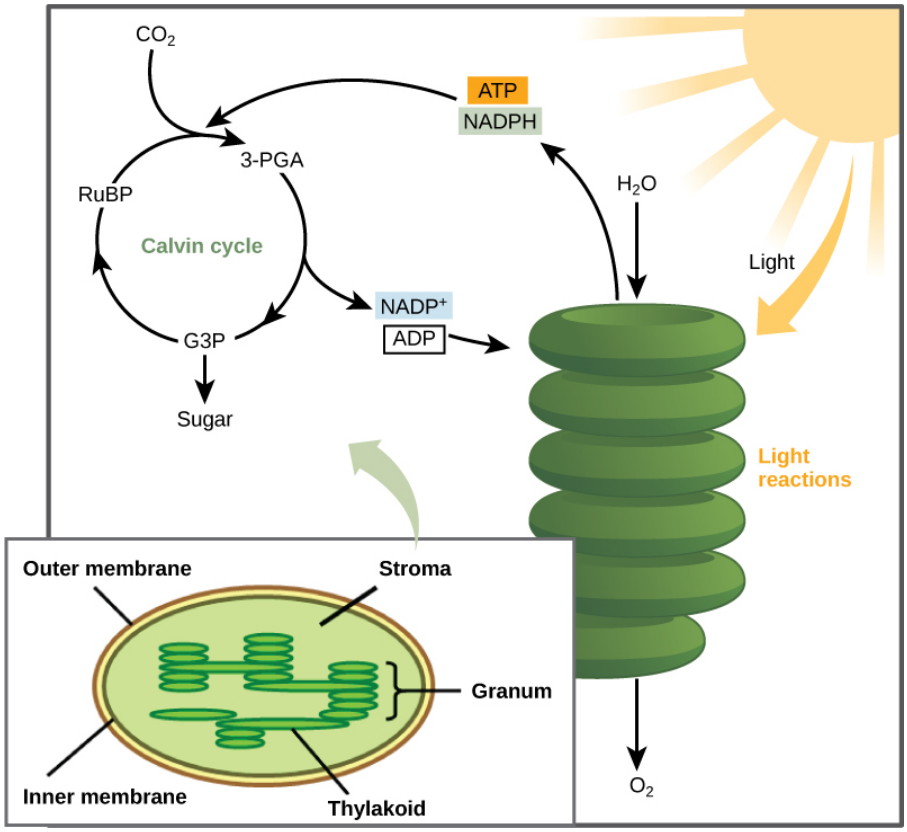

Figure 1. Light-dependent reactions harness energy from the sun to produce ATP and NADPH. These energy-carrying molecules travel into the stroma where the Calvin cycle reactions take place.

The Calvin cycle reactions (Figure 2) can be organized into three basic stages: fixation, reduction, and regeneration. In the stroma, in addition to CO_2, two other chemicals are present to initiate the Calvin cycle: an enzyme abbreviated RuBisCO, and the molecule ribulose bisphosphate (RuBP). RuBP has five atoms of carbon and a phosphate group on each end.

RuBisCO catalyzes a reaction between CO_2 and RuBP, which forms a six-carbon compound that is immediately converted into two three-carbon compounds. This process is called carbon fixation, because CO_2 is "fixed" from its inorganic form into organic molecules.

ATP and NADPH use their stored energy to convert the three-carbon compound, 3-PGA, into another three-carbon compound called G3P. This type of reaction is called a reduction reaction, because it involves the gain of electrons. A reduction is the gain of an electron by an atom or molecule. The molecules of ADP and NAD^+, resulting from the reduction reaction, return to the light-dependent reactions to be re-energized.

One of the G3P molecules leaves the Calvin cycle to contribute to the formation of the carbohydrate molecule, which is commonly glucose ($C_6H_{12}O_6$). Because the carbohydrate molecule has six carbon atoms, it takes six turns of the Calvin cycle to make one carbohydrate molecule (one for each carbon dioxide molecule fixed). The remaining G3P molecules regenerate RuBP, which enables the system to prepare for the carbon-fixation step. ATP is also used in the regeneration of RuBP.

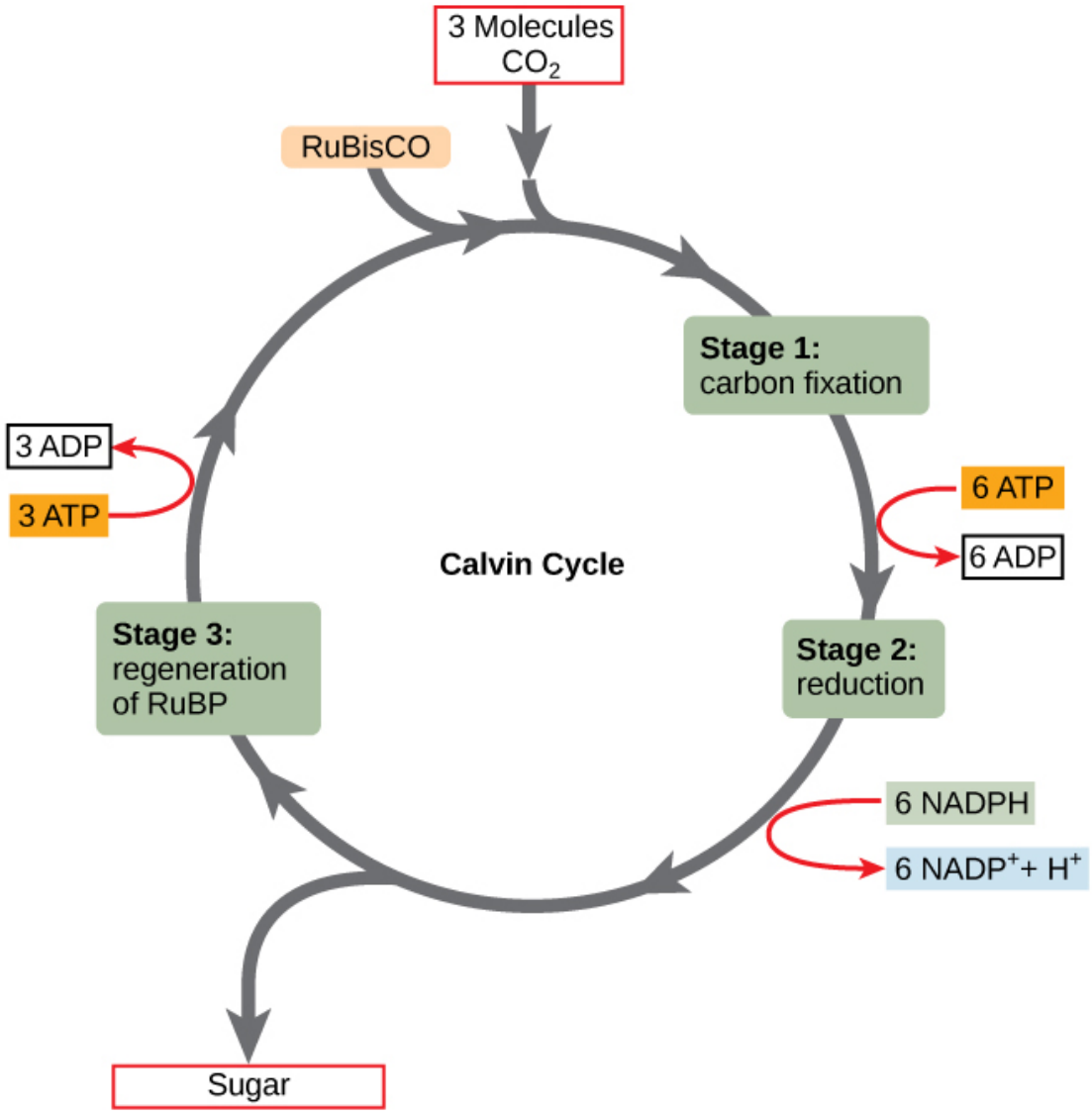

Figure 2. The Calvin cycle has three stages. In stage 1, the enzyme RuBisCO incorporates carbon dioxide into an organic molecule. In stage 2, the organic molecule is reduced. In stage 3, RuBP, the molecule that starts the cycle, is regenerated so that the cycle can continue.

In summary, it takes six turns of the Calvin cycle to fix six carbon atoms from CO_2. These six turns require energy input from 12 ATP molecules and 12 NADPH molecules in the reduction step and 6 ATP molecules in the regeneration step.

Check out this animation of the Calvin cycle. Click Stage 1, Stage 2, and then Stage 3 to see G3P and ATP regenerate to form RuBP.

Evolution in Action: Photosynthesis

The shared evolutionary history of all photosynthetic organisms is conspicuous, as the basic process has changed little over eras of time. Even between the giant tropical leaves in the rainforest and tiny cyanobacteria, the process and components of photosynthesis that use water as an electron donor remain largely the same. Photosystems function to absorb light and use electron transport chains to convert energy. The Calvin cycle reactions assemble carbohydrate molecules with this energy.

However, as with all biochemical pathways, a variety of conditions leads to varied adaptations that affect the basic pattern. Photosynthesis in dry-climate plants (Figure 3) has evolved with adaptations that conserve water. In the harsh dry heat, every drop of water and precious energy must be used to survive. Two adaptations have evolved in such plants. In one form, a more efficient use of CO_2 allows plants to photosynthesize even when CO_2 is in short supply, as when the stomata are closed on hot days. The other adaptation performs preliminary reactions of the Calvin cycle at night, because opening the stomata at this time conserves water due to cooler temperatures. In addition, this adaptation has allowed plants to carry out low levels of photosynthesis without opening stomata at all, an extreme mechanism to face extremely dry periods.

Figure 3. Living in the harsh conditions of the desert has led plants like this cactus to evolve variations in reactions outside the Calvin cycle. These variations increase efficiency and help conserve water and energy. (credit: Piotr Wojtkowski)

Photosynthesis in Prokaryotes

The two parts of photosynthesis—the light-dependent reactions and the Calvin cycle—have been described, as they take place in chloroplasts. However, prokaryotes, such as cyanobacteria, lack membrane-bound organelles. Prokaryotic photosynthetic autotrophic organisms have infoldings of the plasma membrane for chlorophyll attachment and photosynthesis (Figure 4). It is here that organisms like cyanobacteria can carry out photosynthesis.

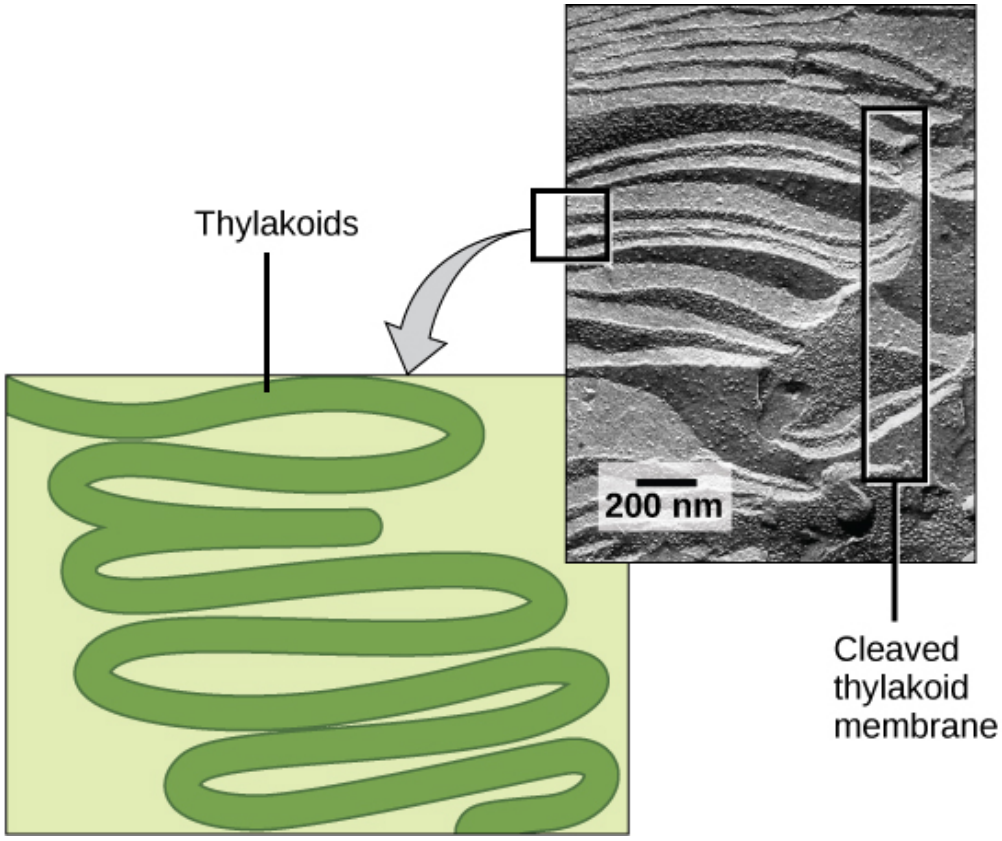

Figure 4. A photosynthetic prokaryote has infolded regions of the plasma membrane that function like thylakoids. Although these are not contained in an organelle, such as a chloroplast, all of the necessary components are present to carry out photosynthesis. (credit: scale-bar data from Matt Russell)

In Summary: The Calvin Cycle

Using the energy carriers formed in the first stage of photosynthesis, the Calvin cycle reactions fix CO_2 from the environment to build carbohydrate molecules. An enzyme, RuBisCO, catalyzes the fixation reaction, by combining CO_2 with RuBP. The resulting six-carbon compound is broken down into two three-carbon compounds, and the energy in ATP and NADPH is used to convert these molecules into G3P. One of the three-carbon molecules of G3P leaves the cycle to become a part of a carbohydrate molecule. The remaining G3P molecules stay in the cycle to be formed back into RuBP, which is ready to react with more CO_2. Photosynthesis forms a balanced energy cycle with the process of cellular respiration. Plants are capable of both photosynthesis and cellular respiration, since they contain both chloroplasts and mitochondria.

Practice Question

Which part of the Calvin cycle would be affected if a cell could not produce the enzyme RuBisCO?

Answer

None of the cycle could take place, because RuBisCO is essential in fixing carbon dioxide. Specifically, RuBisCO catalyzes the reaction between carbon dioxide and RuBP at the start of the cycle.

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SUMMARY: PHOTOSYNTHESIS

Learning Outcomes

- Summarize the process of photosynthesis
- Describe how the wavelength of light affects its energy and color
- Describe the light-dependent reactions that take place during photosynthesis
- Describe the steps and processes in the Calvin Cycle

Now that we've learned about the different pieces of photosynthesis, let's put it all together. This video walks you through the process of photosynthesis as a whole:

Watch this video online: https://youtu.be/sQK3Yr4Sc_k

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INTRODUCTION TO CONNECTIONS TO OTHER METABOLIC PATHWAYS

What you'll learn to do: Discuss the connections between metabolic pathways

You have learned about the catabolism of glucose, which provides energy to living cells. But living things consume more than just glucose for food. How does a turkey sandwich, which contains protein, provide energy to your cells? This happens because all of the catabolic pathways for carbohydrates, proteins, and lipids eventually connect into glycolysis and the citric acid cycle pathways. Metabolic pathways should be thought of as porous—that is, substances enter from other pathways, and other substances leave for other pathways. These pathways are not closed systems. Many of the products in a particular pathway are reactants in other pathways.

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CONNECTIONS TO OTHER METABOLIC PATHWAYS

Learning Outcomes

- Discuss the way in which carbohydrate metabolic pathways, glycolysis, and the citric acid cycle interrelate with protein and lipid metabolic pathways

Connections of Other Sugars to Glucose Metabolism

Glycogen, a polymer of glucose, is a short-term energy storage molecule in animals. When there is adequate ATP present, excess glucose is converted into glycogen for storage. Glycogen is made and stored in the liver and muscle. Glycogen will be taken out of storage if blood sugar levels drop. The presence of glycogen in muscle cells as a source of glucose allows ATP to be produced for a longer time during exercise.

Sucrose is a disaccharide made from glucose and fructose bonded together. Sucrose is broken down in the small intestine, and the glucose and fructose are absorbed separately. Fructose is one of the three dietary monosaccharides, along with glucose and galactose (which is part of milk sugar, the disaccharide lactose), that are absorbed directly into the bloodstream during digestion. The catabolism of both fructose and galactose produces the same number of ATP molecules as glucose.

Connections of Proteins to Glucose Metabolism

Proteins are broken down by a variety of enzymes in cells. Most of the time, amino acids are recycled into new proteins. If there are excess amino acids, however, or if the body is in a state of famine, some amino acids will be shunted into pathways of glucose catabolism. Each amino acid must have its amino group removed prior to entry into these pathways. The amino group is converted into ammonia. In mammals, the liver synthesizes urea from two ammonia molecules and a carbon dioxide molecule. Thus, urea is the principal waste product in mammals from the nitrogen originating in amino acids, and it leaves the body in urine.

Connections of Lipids to Glucose Metabolism

The lipids that are connected to the glucose pathways are cholesterol and triglycerides. Cholesterol is a lipid that contributes to cell membrane flexibility and is a precursor of steroid hormones. The synthesis of cholesterol starts with acetyl CoA and proceeds in only one direction. The process cannot be reversed, and ATP is not produced.

Triglycerides are a form of long-term energy storage in animals. Triglycerides store about twice as much energy as carbohydrates. Triglycerides are made of glycerol and three fatty acids. Animals can make most of the fatty acids they need. Triglycerides can be both made and broken down through parts of the glucose catabolism pathways. Glycerol can be phosphorylated and proceeds through glycolysis. Fatty acids are broken into two-carbon units that enter the citric acid cycle.

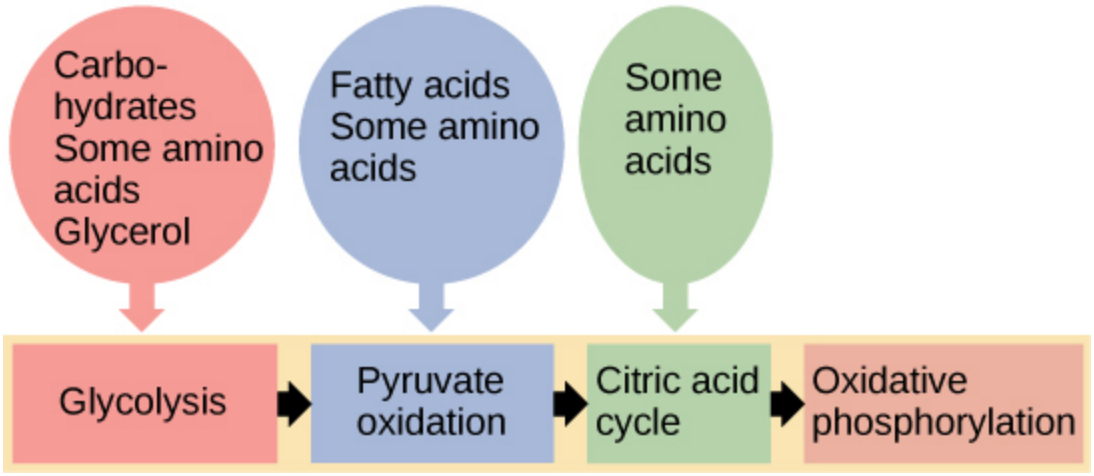

Figure 1. Glycogen from the liver and muscles, together with fats, can feed into the catabolic pathways for carbohydrates.

Pathways of Photosynthesis and Cellular Metabolism

Photosynthesis and cellular metabolism consist of several very complex pathways. It is generally thought that the first cells arose in an aqueous environment—a "soup" of nutrients. If these cells reproduced successfully and their numbers climbed steadily, it follows that the cells would begin to deplete the nutrients from the medium in which they lived, as they shifted the nutrients into their own cells. This hypothetical situation would have resulted in natural selection favoring those organisms that could exist by using the nutrients that remained in their environment and by manipulating these nutrients into materials that they could use to survive. Additionally, selection would favor those organisms that could extract maximal value from the available nutrients.

An early form of photosynthesis developed that harnessed the sun's energy using compounds other than water as a source of hydrogen atoms, but this pathway did not produce free oxygen. It is thought that glycolysis developed prior to this time and could take advantage of simple sugars being produced, but these reactions were not able to fully extract the energy stored in the carbohydrates. A later form of photosynthesis used water as a source of hydrogen ions and generated free oxygen. Over time, the atmosphere became oxygenated. Living things adapted to exploit this new atmosphere and allowed respiration as we know it to evolve. When the full process of photosynthesis as we know it developed and the atmosphere became oxygenated, cells were finally able to use the oxygen expelled by photosynthesis to extract more energy from the sugar molecules using the citric acid cycle.

In Summary: Connections to Other Metabolic Pathways

The breakdown and synthesis of carbohydrates, proteins, and lipids connect with the pathways of glucose catabolism. The carbohydrates that can also feed into glucose catabolism include galactose, fructose, and glycogen. These connect with glycolysis. The amino acids from proteins connect with glucose catabolism through pyruvate, acetyl CoA, and components of the citric acid cycle. Cholesterol synthesis starts with acetyl CoA, and the components of triglycerides are picked up by acetyl CoA and enter the citric acid cycle.

Additional Self Check Question

Would you describe metabolic pathways as inherently wasteful or inherently economical, and why?

Answer

They are very economical. The substrates, intermediates, and products move between pathways and do so in response to finely tuned feedback inhibition loops that keep metabolism overall on an even keel. Intermediates in one pathway may occur in another, and they can move from one pathway to another fluidly in response to the needs of the cell.

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THE ENERGY CYCLE

Learning Outcomes

Describe the energy cycle of all living organisms

Living things access energy by breaking down carbohydrate molecules. However, if plants make carbohydrate molecules, why would they need to break them down? Carbohydrates are storage molecules for energy in all living things. Although energy can be stored in molecules like ATP, carbohydrates are much more stable and efficient reservoirs for chemical energy. Photosynthetic organisms also carry out the reactions of respiration to harvest the energy that they have stored in carbohydrates, for example, plants have mitochondria in addition to chloroplasts.

You may have noticed that the overall reaction for photosynthesis:

$$6CO_2 + 6H_2O \rightarrow C_6H_{12}O_6 + 6O_2$$

is the reverse of the overall reaction for cellular respiration:

$$6O_2 + C_6H_{12}O_6 \rightarrow 6CO_2 + 6H_2O$$

Photosynthesis produces oxygen as a byproduct, and respiration produces carbon dioxide as a byproduct.

In nature, there is no such thing as waste. Every single atom of matter is conserved, recycling indefinitely. Substances change form or move from one type of molecule to another, but never disappear (Figure 1).

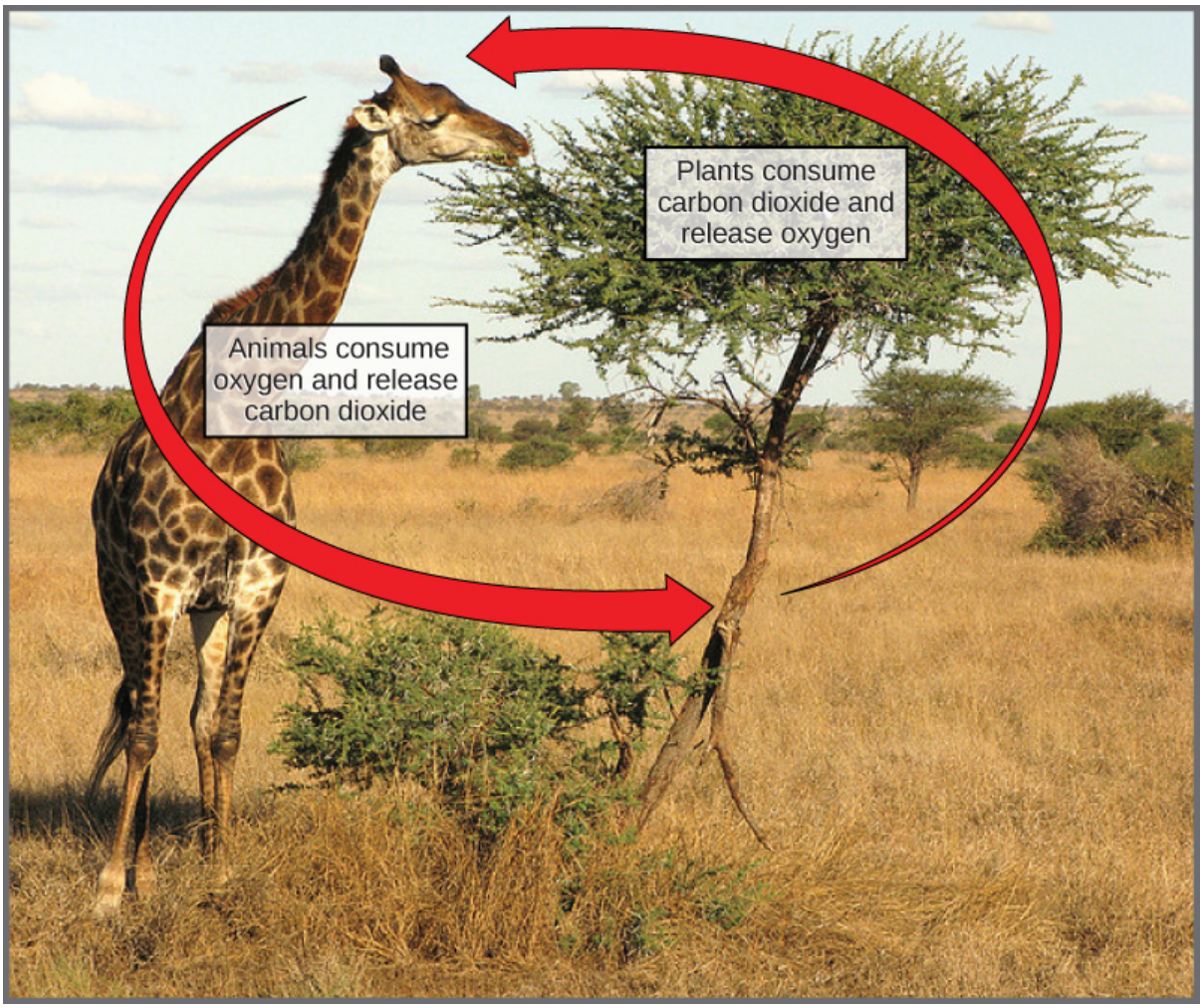

Figure 1. In the carbon cycle, the reactions of photosynthesis and cellular respiration share reciprocal reactants and products. (credit: modification of work by Stuart Bassil)

CO_2 is no more a form of waste produced by respiration than oxygen is a waste product of photosynthesis. Both are byproducts of reactions that move on to other reactions. Photosynthesis absorbs energy to build carbohydrates in chloroplasts, and aerobic cellular respiration releases energy by using oxygen to break down carbohydrates in mitochondria. Both organelles use electron transport chains to generate the energy necessary to drive other reactions. Photosynthesis and cellular respiration function in a biological cycle, allowing organisms to access life-sustaining energy that originates millions of miles away in a star.

Practice Question

Explain the reciprocal nature of the net chemical reactions for photosynthesis and respiration.

Answer

Photosynthesis takes the energy of sunlight and combines water and carbon dioxide to produce sugar and oxygen as a waste product. The reactions of respiration take sugar and consume oxygen to break it down into carbon dioxide and water, releasing energy. Thus, the reactants of photosynthesis are the products of respiration, and vice versa.

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PUTTING IT TOGETHER: METABOLIC PATHWAYS

Whether the organism is a bacterium, plant, or animal, all living things access energy by breaking down carbohydrate molecules. But if plants make carbohydrate molecules, why would they need to break them down, especially when it has been shown that the gas organisms release as a "waste product" (CO_2) acts as a substrate for the formation of more food in photosynthesis? Remember, living things need energy to perform life functions. In addition, an organism can either make its own food or eat another organism—either way, the food still needs to be converted to a form cells can actually use. Finally, in that process of conversion, called cellular respiration, organisms release needed energy and produce "waste" in the form of CO_2 gas.

Biofuels

Obviously its important for providing energy for living organisms to power themselves. But is that the only power that photosynthesis provides? What about biofuels? Watch this 14 minute video for an amazing discussion of a proposed biofuel source that doesn't use arable land, doesn't take away food crops, and utilizes wastewater from cities.

Watch this video online: https://youtu.be/X-HE4Hfa-OY

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MODULE 7: CELL DIVISION

WHY IT MATTERS: CELL DIVISION

Why learn about the various stages of cell division?

Cell division is key to life: from the moment we are first conceived, we are continually changing and growing. In order for our bodies to grow and develop, they must produce new cells—and allow for the death of old cells. Cell division is also an essential component of injury repair. If our cells couldn't divide and create new cells, our bodies could never produce new skin cells to heal road rash, or grow a fingernail back. However, when cell division goes awry, dramatic results may occur. Without sufficient cellular oversight, repeated rounds of unregulated cell division can lead to a minor condition like psoriasis or a life-threatening disease like cancer. Cell division takes occurs by a strict cycle, with multiple stages and checkpoints to ensure things *don't* go awry.

Perhaps most importantly, without cell division, no species would be able to reproduce—life would simply end (or would have ended a long time ago). Every human, as well as every sexually reproducing organism, begins life as a fertilized egg (embryo) or zygote. Trillions of cell divisions subsequently occur in a controlled manner to produce a complex, multicellular human. In other words, that original single cell is the ancestor of every other cell in the body. Single-celled organisms use cell division as their method of reproduction.

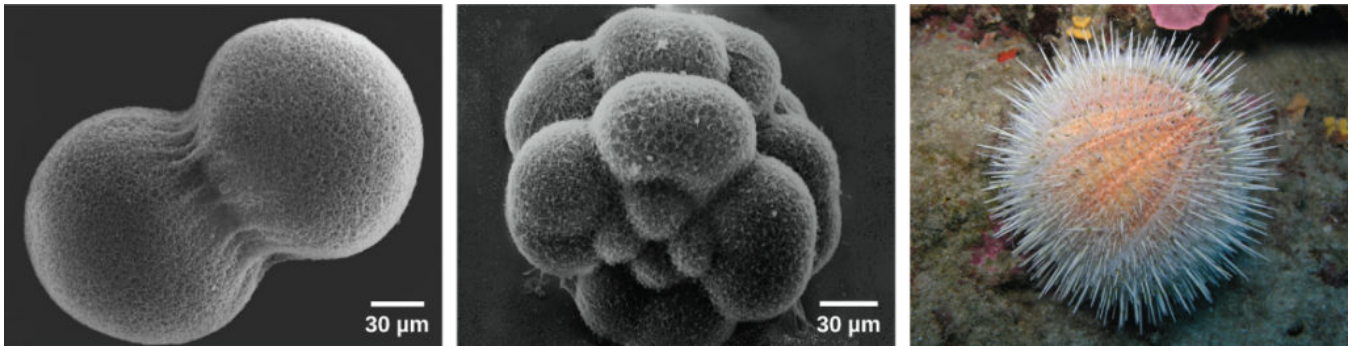

Figure 1. A sea urchin begins life as a single cell that (a) divides to form two cells, visible by scanning electron microscopy. After four rounds of cell division, (b) there are 16 cells, as seen in this SEM image. After many rounds of cell division, the individual develops into a complex, multicellular organism, as seen in this (c) mature sea urchin. (credit a: modification of work by Evelyn Spiegel, Louisa Howard; credit b: modification of work by Evelyn Spiegel, Louisa Howard; credit c: modification of work by Marco Busdraghi; scale-bar data from Matt Russell)

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INTRODUCTION TO CHROMOSOMES AND DNA PACKAGING

What you'll learn to do: Understand chromosome structure and organization in eukaryotic cells

When a cell divides, it is essential that the new cell (also known as the daughter cell) contains the same genetic information as the old cell (also known as the parent cell). This genetic information is our DNA, which is packaged into chromosomes. Each chromosome contains information about specific traits of an organism. These chromosomes can be sorted into two categories: autosomes and sex chromosomes. In this section will discuss these two types of chromosomes and the differences between the two as well as how cells package DNA. Watch the video below for an overview of chromosomes.

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DNA AND CHROMOSOMES

Learning Outcomes

Differentiate between two kinds of chromosomes: autosomes and sex chromosomes

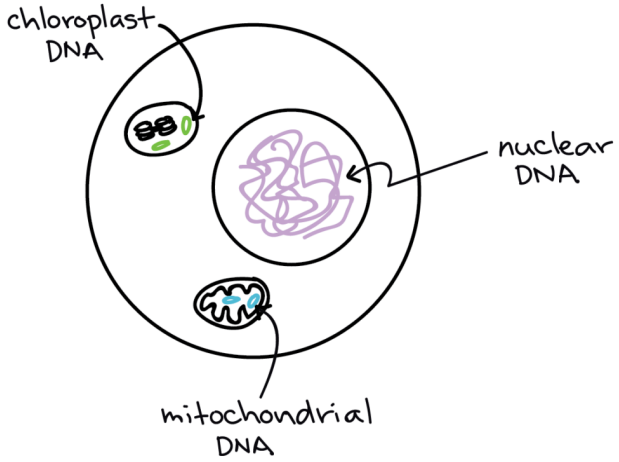
When a cell divides in two, one of its main jobs is to make sure that each of the two new cells gets a full, perfect copy of genetic material. Mistakes during copying, or unequal division of the genetic material between cells, can lead to cells that are unhealthy or nonfunctional (and even to diseases such as cancer). But what exactly is this genetic material, and how does it behave over the course of a cell division?

DNA and Genomes

DNA (deoxyribonucleic acid) is the genetic material of living organisms. In humans, DNA is found in almost all the cells of the body and provides the instructions they need to grow, function, and respond to their environment. When a cell of the body divides, it will pass on a copy of its DNA to each of its daughter cells. DNA is also passed on at the at the level of organisms, with the DNA in sperm and egg cells combining to form a new organism that has genetic material from both its parents. Physically speaking, DNA is a long string of paired chemical units (nucleotides) that come in four different types, and it carries information organized into units called genes. Genes typically provide instructions for making proteins, which give cells and organisms their functional characteristics.

In eukaryotes such as plants and animals, the great majority of DNA is found in the nucleus and is called **nuclear DNA**. In bacteria and other prokaryotes, most of the DNA is found in a central region of the cell called the **nucleoid**, which functions similarly to a nucleus but is not surrounded by a membrane.

A cell's set of DNA is called its **genome**. Since all of the cells in an organism (with a few exceptions) contain the same DNA, you can also say that an organism has its own genome, and since the members of a species typically have similar genomes, you can also describe the genome of a species. In general, when people refer to the human genome, or any other eukaryotic genome, they mean the set of DNA found in the nucleus (that is, the nuclear genome).



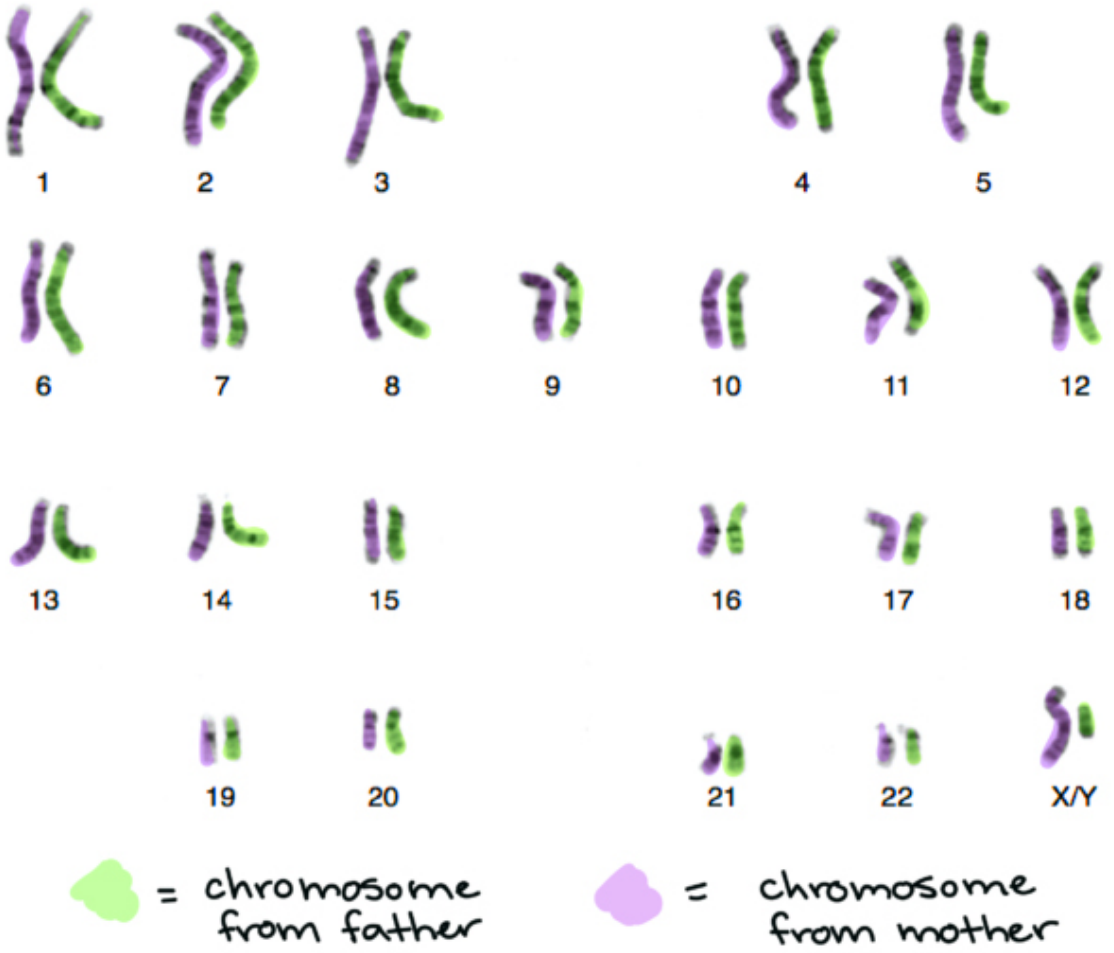
Chromosomes

Each species has its own characteristic number of chromosomes. Humans, for instance, have 46 chromosomes in a typical body cell, while dogs have 78. Like many species of animals and plants, humans are **diploid (2*n*)**, meaning that most of their chromosomes come in matched sets known as **homologous pairs**. Thus, the 46 chromosomes of a human cell are organized into 23 pairs, and the two members of each pair are said to be **homologues** of one another (with the slight exception of the X and Y chromosomes; see below).

Human sperm and eggs, which have only one homologous chromosome from each pair, are said to be **haploid (1*n*)**. When a sperm and egg fuse, their genetic material combines to form one complete, diploid set of chromosomes. So, for each homologous pair of chromosomes in your genome, one of the homologues comes from your mom and the other from your dad.

The two chromosomes in a homologous pair are generally very similar to one another. They're the same size and shape, and have the same pattern of light and dark bands, as you can see in the human **karyotype** (image of the chromosomes) shown above. Bands appear when the chromosomes are stained with a dye, and the dark bands mark more compacted DNA (usually, with fewer genes), while the light bands mark less compacted DNA (usually, with more genes). Most importantly, the two homologues in a pair carry the same type of genetic information. For instance, there is a gene found near the bottom of chromosome 15 that affects eye color (Note: The Tech Museum of Innovation. (2013). Don't it make your brown eyes blue? In *Stanford at the tech: understanding genetics*. Retrieved from http://genetics.thetech.org/original_news/news39.). A person might have the blue version, or **allele**, of this gene on one homologue, but the brown version on the other. Both homologues have the same type of gene in the same place, but they can (and often do!) have different versions of genes.

In humans, the X and Y chromosomes determine a person's biological sex, with XX for female and XY for male. While the two X chromosomes in a woman's cells are genuinely homologous, the X and Y chromosomes of a man's cells are not. They differ in size and shape, with the X being much larger than the Y, and contain different mostly different genes (although they do have small regions of similarity). The X and Y chromosomes are known as **sex chromosomes**, while the other 44 human chromosomes are called **autosomes**.



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CHROMOSOME STRUCTURE

Learning Outcomes

Understand how DNA is protected and compacted inside cells

The continuity of life from one cell to another has its foundation in the reproduction of cells by way of the cell cycle. The **cell cycle** is an orderly sequence of events that describes the stages of a cell's life from the division of a single parent cell to the production of two new daughter cells. The mechanisms involved in the cell cycle are highly regulated. Part of that regulation involves the physical shape and structure that the DNA has during different phases of the cell cycle.

Eukaryotic Chromosomal Structure and Compaction

If the DNA from all 46 chromosomes in a human cell nucleus was laid out end to end, it would measure approximately two meters; however, its diameter would be only 2 nm. Considering that the size of a typical human cell is about 10 μm (100,000 cells lined up to equal one meter), DNA must be tightly packaged to fit in the cell's nucleus. At the same time, it must also be readily accessible for the genes to be expressed. During some stages of the cell cycle, the long strands of DNA are condensed into compact chromosomes. There are a number of ways that chromosomes are compacted.

In the first level of compaction, short stretches of the DNA double helix wrap around a core of eight **histone** proteins at regular intervals along the entire length of the chromosome (Figure 1). The DNA-histone complex is called **chromatin**. The beadlike, histone DNA complex is called a **nucleosome**, and DNA connecting the nucleosomes is called linker DNA. A DNA molecule in this form is about seven times shorter than the double helix without the histones, and the beads are about 10 nm in diameter, in contrast with the 2-nm diameter of a DNA double helix. The next level of compaction occurs as the nucleosomes and the linker DNA between them are coiled into a 30-nm chromatin fiber. This coiling further shortens the chromosome so that it is now about 50 times shorter than the extended form. In the third level of packing, a variety of fibrous proteins is used to pack the chromatin. These fibrous proteins also ensure that each chromosome in a non-dividing cell occupies a particular area of the nucleus that does not overlap with that of any other chromosome.

DNA replicates in the S phase of interphase. After replication, the chromosomes are composed of two linked **sister chromatids**. The connection between the sister chromatids is closest in a region called the **centromere**. The conjoined sister chromatids, are visible under a light microscope. The centromeric region is highly condensed and thus will appear as a constricted area.

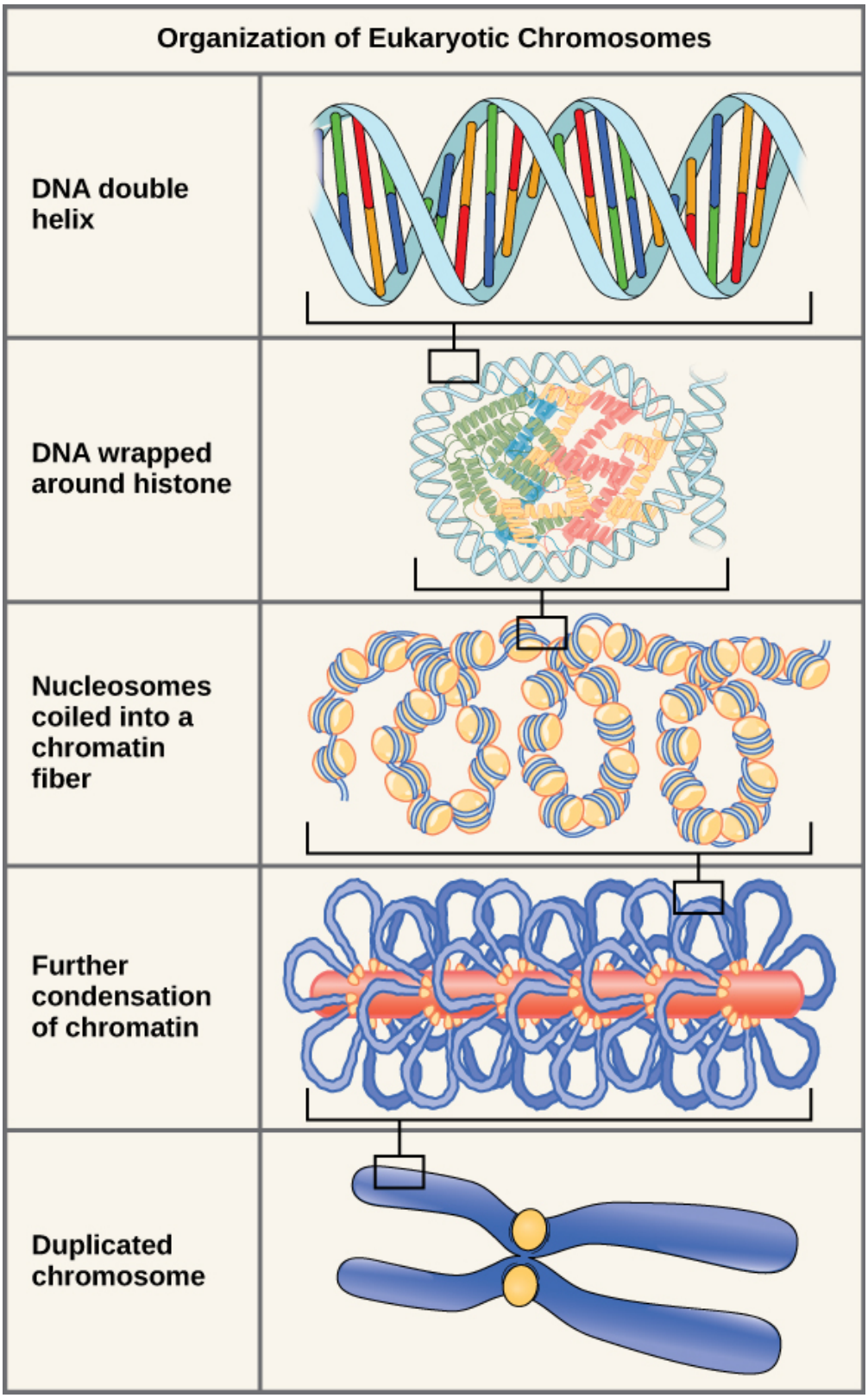

Figure 1. Double-stranded DNA wraps around histone proteins to form nucleosomes that have the appearance of "beads on a string." The nucleosomes are coiled into a 30-nm chromatin fiber. When a cell undergoes mitosis, the chromosomes condense even further.

This animation illustrates the different levels of chromosome packing:
Watch this video online: https://youtu.be/gbSIBhFwQ4s

In Summary: Chromosome Structure

DNA in eukaryotes is highly structured and organized in all stages of an organisms life. Diploid organisms contain a pair of each chromosome; humans have 23 pairs for a total number of 46 chromosomes. Pairs of chromosomes, also known as homologous chromosomes, contain the same genes though there may be differences between the version of gene on each member of the pair. DNA is normally tightly packed into the nucleus of a eukaryotic cell, through protein-DNA complexes that form the characteristic condensed 'chromosome' shape. DNA compacts even further in preparation for cell division.

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INTRODUCTION TO THE CELL CYCLE

What you'll learn to do: Identify the stages of the cell cycle, by picture and by description of major milestones

The **cell cycle** is an ordered series of events involving cell growth and cell division that produces two new daughter cells. Cells on the path to cell division proceed through a series of precisely timed and carefully regulated stages of growth, DNA replication, and division that produces two identical (clone) cells. The cell cycle has two major phases: interphase and the mitotic phase (Figure 1). During **interphase**, the cell grows and DNA is replicated. During the **mitotic phase**, the replicated DNA and cytoplasmic contents are separated, and the cell divides.

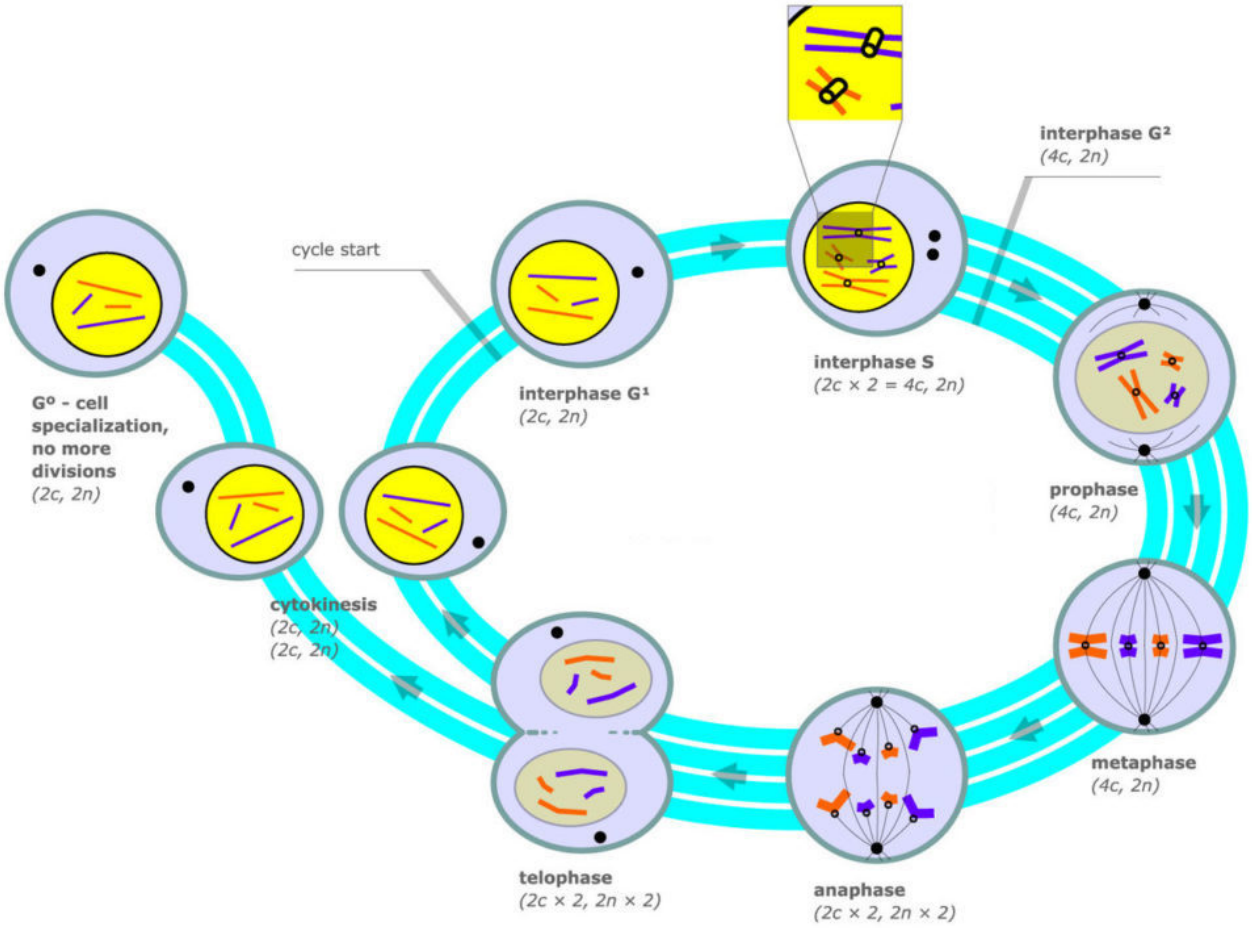

Figure 1. The cell cycle

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INTERPHASE

Learning Outcomes

Identify the characteristics and sub-phases of interphase

Stages of Interphase

During interphase, the cell undergoes normal growth processes while also preparing for cell division. It is the longest phase of the cell cycle, cell spends approximately 90% of its time in this phase. In order for a cell to move from interphase into the mitotic phase, many internal and external conditions must be met.

The three stages of interphase are called G_1, S, and G_2.

G_1 Phase (First Gap)

The first stage of interphase is called the **G_1 phase** (first gap) where the cell is quite active at the biochemical level. The cell is accumulating the building blocks of chromosomal DNA and the associated proteins as well as accumulating sufficient energy reserves to complete the task of replicating each chromosome in the nucleus.

S Phase (Synthesis of DNA)

In the **S phase**, DNA replication results in the formation of identical pairs of DNA molecules—sister chromatids—that are firmly attached to the centromeric region.

G_2 Phase (Second Gap)

In the **G_2 phase**, the cell replenishes its energy stores and synthesizes proteins necessary for chromosome manipulation. Some cell organelles are duplicated, and the cytoskeleton is dismantled to provide resources for the mitotic phase.

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MITOSIS

Learning Outcomes

Identify the characteristics and stages of mitosis

The mitotic phase (also known as M phase) is a multistep process during which the duplicated chromosomes are aligned, separated, and move into two new, identical daughter cells. The first portion of the mitotic phase is called **karyokinesis**, or nuclear division. The second portion of the mitotic phase, called cytokinesis, is the physical separation of the cytoplasmic components into the two daughter cells.

Karyokinesis (Mitosis)

Karyokinesis, also known as mitosis, is divided into a series of phases—prophase, metaphase, anaphase, and telophase—that result in the division of the cell (Figure 1).

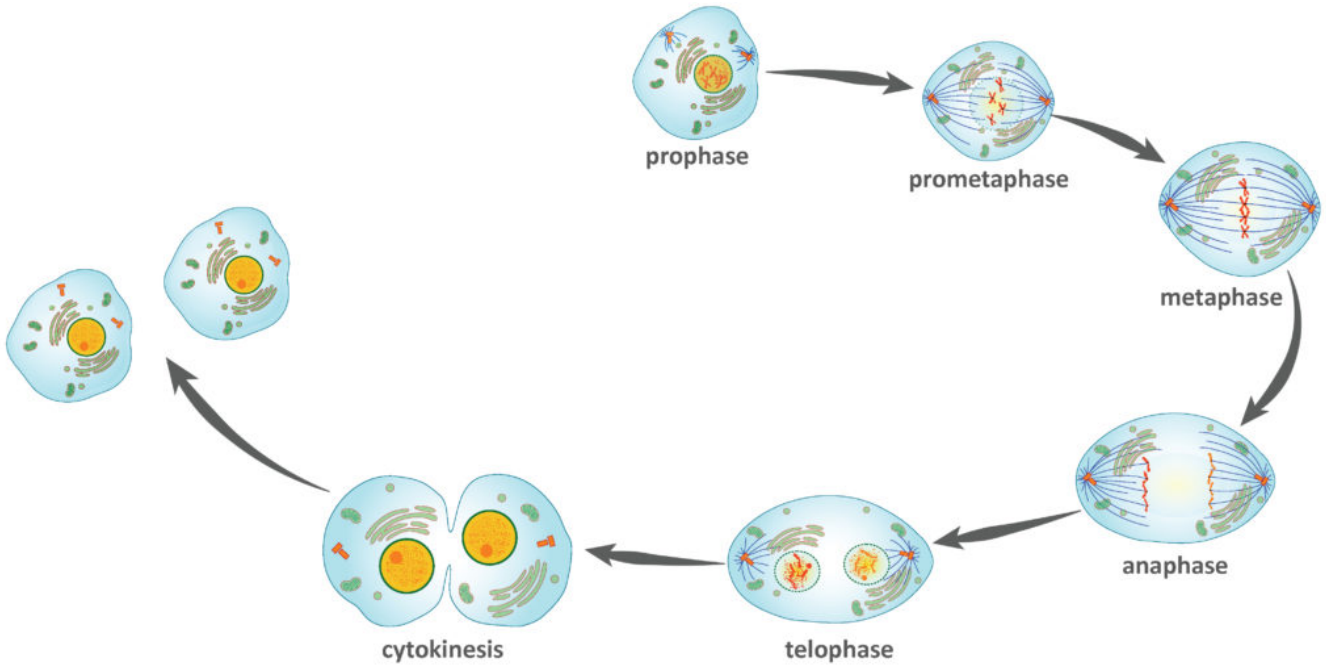

Figure 1. Stages of mitosis

During **prophase**, the "first phase," the nuclear envelope starts to dissociate into small vesicles, and the membranous organelles fragment and disperse toward the periphery of the cell. The nucleolus disappears . The centrosomes begin to move to opposite poles of the cell. Microtubules that will form the mitotic spindle extend between the centrosomes, pushing them farther apart as the microtubule fibers lengthen. The sister chromatids begin to coil more tightly and become visible under a light microscope. Each sister chromatid develops a protein structure called a kinetochore in the centromeric region (Figure 2). The proteins of the kinetochore attract and bind mitotic spindle microtubules.

During **prometaphase**, the nuclear envelope is fully broken down and chromosomes are attached to microtubules from both poles of the mitotic spindle, which begin to move them toward the middle of the cell.

During **metaphase**, all the chromosomes are aligned in a plane called the **metaphase plate**, or the equatorial plane, midway between the two poles of the cell. At this time, the chromosomes are maximally condensed.

During **anaphase**, the sister chromatids separate at the centromere. Each chromatid, now called a chromosome, is pulled rapidly toward the centrosome to which its microtubule is attached. The cell becomes visibly elongated (oval shaped) as the polar microtubules slide against each other at the metaphase plate where they overlap.

During **telophase**, the chromosomes reach the opposite poles and begin to decondense (unravel), relaxing into a chromatin configuration. Nuclear envelopes form around the chromosomes, and nucleosomes appear within the nuclear area.

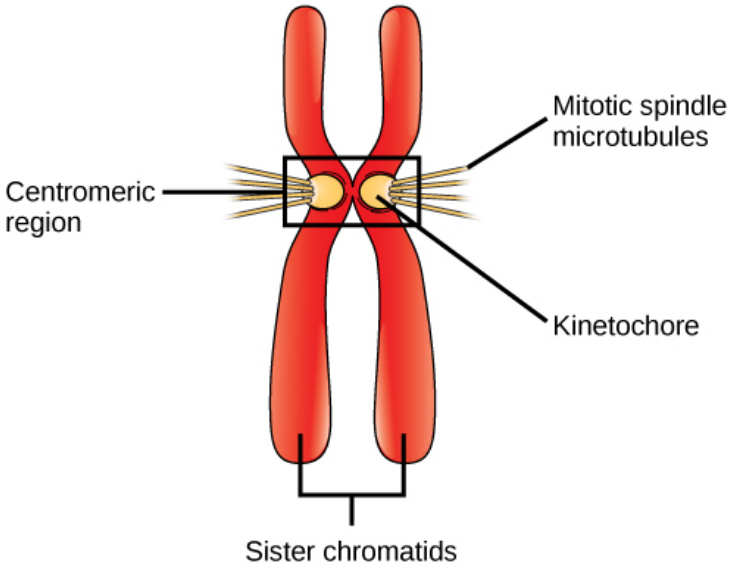

Figure 2. Once a mitotic fiber attaches to a chromosome, the chromosome will be oriented until the kinetochores of sister chromatids face the opposite poles. Eventually, all the sister chromatids will be attached via their kinetochores to microtubules from opposing poles.

The activity below will walk you through mitosis—providing you with the chance to review the different steps of the process and how they work together.

Click here for a text-only version of the activity.

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CYTOKINESIS

Learning Outcomes

Identify the characteristics of cytokinesis

Cytokinesis is the second main stage of the mitotic phase during which cell division is completed via the physical separation of the cytoplasmic components into two daughter cells. Division is not complete until the cell components have been apportioned and completely separated into the two daughter cells. Although the stages of mitosis are similar for most eukaryotes, the process of cytokinesis is quite different for eukaryotes that have cell walls, such as plant cells.

In cells such as animal cells that lack cell walls, cytokinesis follows the onset of anaphase. A contractile ring composed of actin filaments forms just inside the plasma membrane at the former metaphase plate. The actin filaments pull the equator of the cell inward, forming a fissure. This fissure, or "crack," is called the **cleavage furrow**. The furrow deepens as the actin ring contracts, and eventually the membrane is cleaved in two (Figure 1).

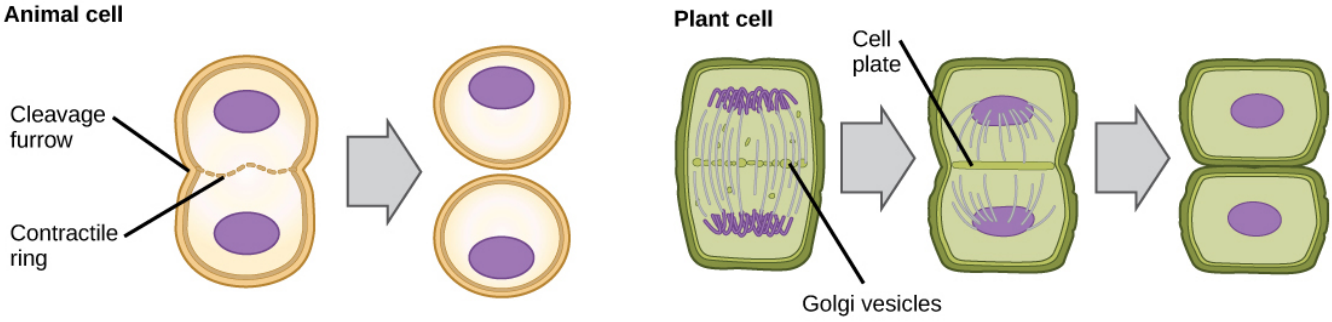

Figure 1. During cytokinesis in animal cells, a ring of actin filaments forms at the metaphase plate. The ring contracts, forming a cleavage furrow, which divides the cell in two. In plant cells, Golgi vesicles coalesce at the former metaphase plate, forming a phragmoplast. A cell plate formed by the fusion of the vesicles of the phragmoplast grows from the center toward the cell walls, and the membranes of the vesicles fuse to form a plasma membrane that divides the cell in two.

In plant cells, a new cell wall must form between the daughter cells. During interphase, the Golgi apparatus accumulates enzymes, structural proteins, and glucose molecules prior to breaking into vesicles and dispersing throughout the dividing cell. During telophase, these Golgi vesicles are transported on microtubules to form a phragmoplast (a vesicular structure) at the metaphase plate. There, the vesicles fuse and coalesce from the center toward the cell walls; this structure is called a **cell plate**. As more vesicles fuse, the cell plate enlarges until it merges with the cell walls at the periphery of the cell. Enzymes use the glucose that has accumulated between the membrane layers to build a new cell wall. The Golgi membranes become parts of the plasma membrane on either side of the new cell wall (Figure 1).

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THE COMPLETE CELL CYCLE

Learning Outcomes

- Identify the characteristics and sub-phases of interphase
- Identify the characteristics and stages of mitosis
- Identify the characteristics of cytokinesis

Remember, mitosis is the process of cell division, but it's just a portion of the full cell cycle. Figure 1 shows approximately how long a cell spends in each stage of the cell cycle:

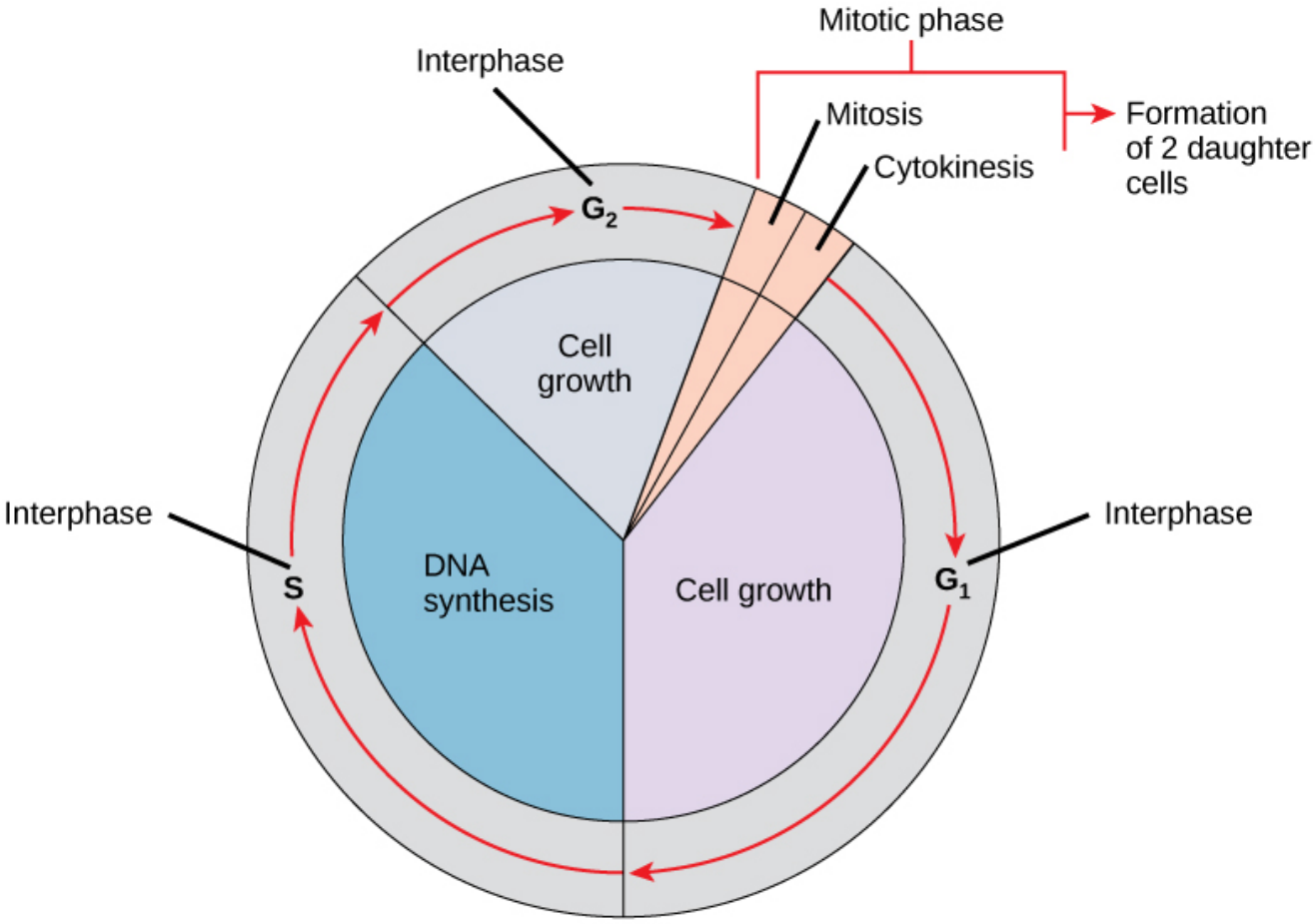

Figure 1. The cell cycle consists of interphase and the mitotic phase. During interphase, the cell grows and the nuclear DNA is duplicated. Interphase is followed by the mitotic phase. During the mitotic phase, the duplicated chromosomes are segregated and distributed into daughter nuclei. The cytoplasm is usually divided as well, resulting in two daughter cells.

As you can see, cells spend most of their time in interphase.

Video Review: The Cell Cycle

This video reviews all the steps of mitosis; seeing it all together is a great review at this stage.
Watch this video online: https://youtu.be/L0k-enzoeOM

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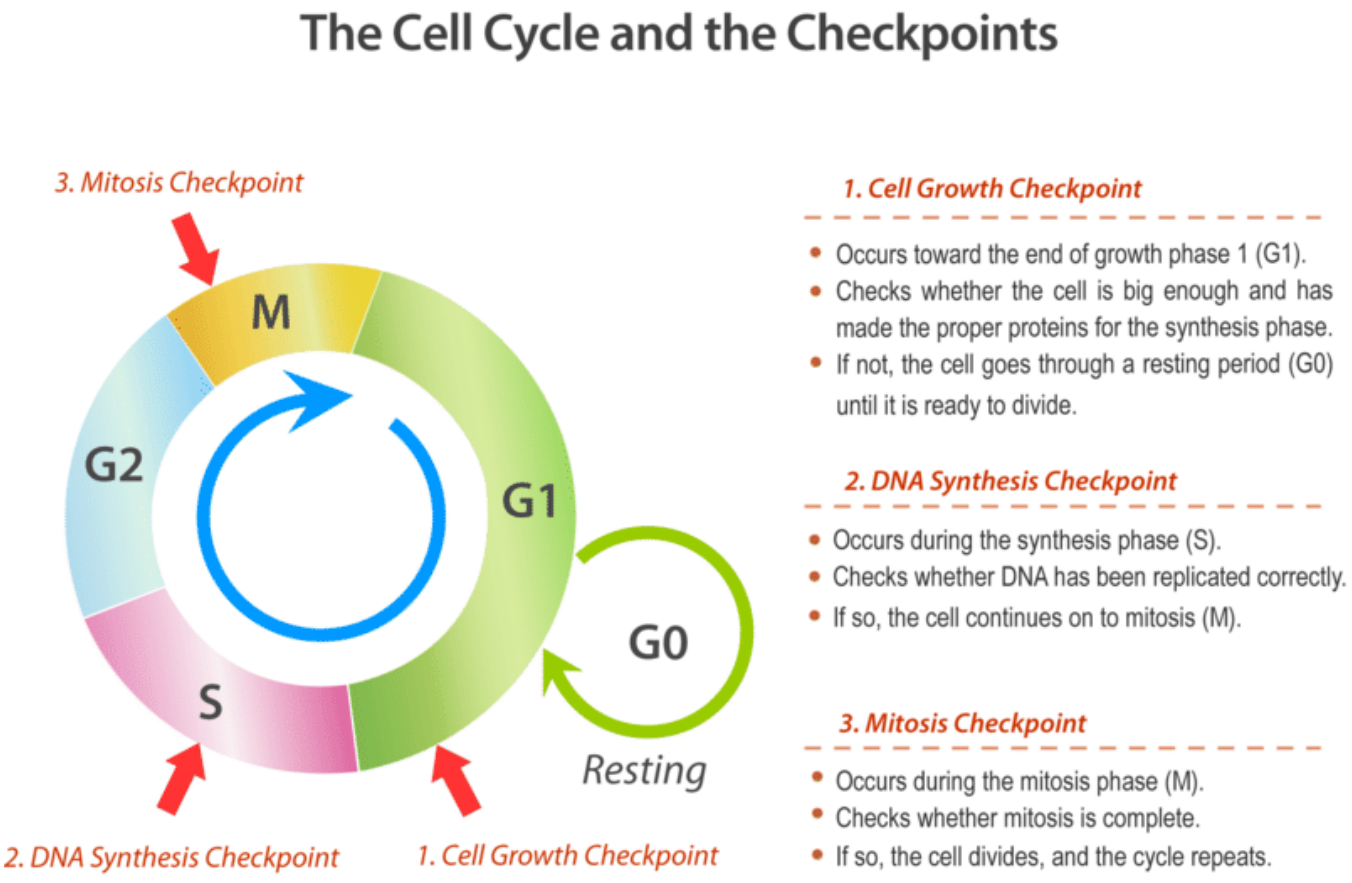
INTRODUCTION TO CELL CYCLE CHECKPOINTS

What you'll learn to do: Identify and explain the important checkpoints that a cell passes through during the cell cycle

As we just learned, the cell cycle is a fairly complicated process. In order to make sure everything goes right, there are checkpoints in the cycle:



Let's learn more about these different checkpoints and how they help control the cell cycle.

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CONTROL OF THE CELL CYCLE

Learning Outcomes

Identify important checkpoints in cell division

The length of the cell cycle is highly variable, even within the cells of a single organism. In humans, the frequency of cell turnover ranges from a few hours in early embryonic development, to an average of two to five days for epithelial cells, and to an entire human lifetime spent in G_0 by specialized cells, such as cortical neurons or cardiac muscle cells. There is also variation in the time that a cell spends in each phase of the cell cycle. When fast-dividing mammalian cells are grown in culture (outside the body under optimal growing conditions), the length of the cycle is about 24 hours. In rapidly dividing human cells with a 24-hour cell cycle, the G_1 phase lasts approximately nine hours, the S phase lasts 10 hours, the G_2 phase lasts about four and one-half hours, and the M phase lasts approximately one-half hour. In early embryos of fruit flies, the cell cycle is completed in about eight minutes. The timing of events in the cell cycle is controlled by mechanisms that are both internal and external to the cell.

Regulation of the Cell Cycle by External Events

Both the initiation and inhibition of cell division are triggered by events external to the cell when it is about to begin the replication process. An event may be as simple as the death of a nearby cell or as sweeping as the release of growth-promoting hormones, such as human growth hormone (HGH). A lack of HGH can inhibit cell division, resulting in dwarfism, whereas too much HGH can result in gigantism. Crowding of cells can also inhibit cell division. Another factor that can initiate cell division is the size of the cell; as a cell grows, it becomes inefficient due to its decreasing surface-to-volume ratio. The solution to this problem is to divide.

Whatever the source of the message, the cell receives the signal, and a series of events within the cell allows it to proceed into interphase. Moving forward from this initiation point, every parameter required during each cell cycle phase must be met or the cycle cannot progress.

Regulation at Internal Checkpoints

It is essential that the daughter cells produced be exact duplicates of the parent cell. Mistakes in the duplication or distribution of the chromosomes lead to mutations that may be passed forward to every new cell produced from an abnormal cell. To prevent a compromised cell from continuing to divide, there are internal control mechanisms that operate at three main cell cycle checkpoints. A checkpoint is one of several points in the eukaryotic cell cycle at which the progression of a cell to the next stage in the cycle can be halted until conditions are favorable. These checkpoints occur near the end of G_1, at the G_2/M transition, and during metaphase (Figure 1).

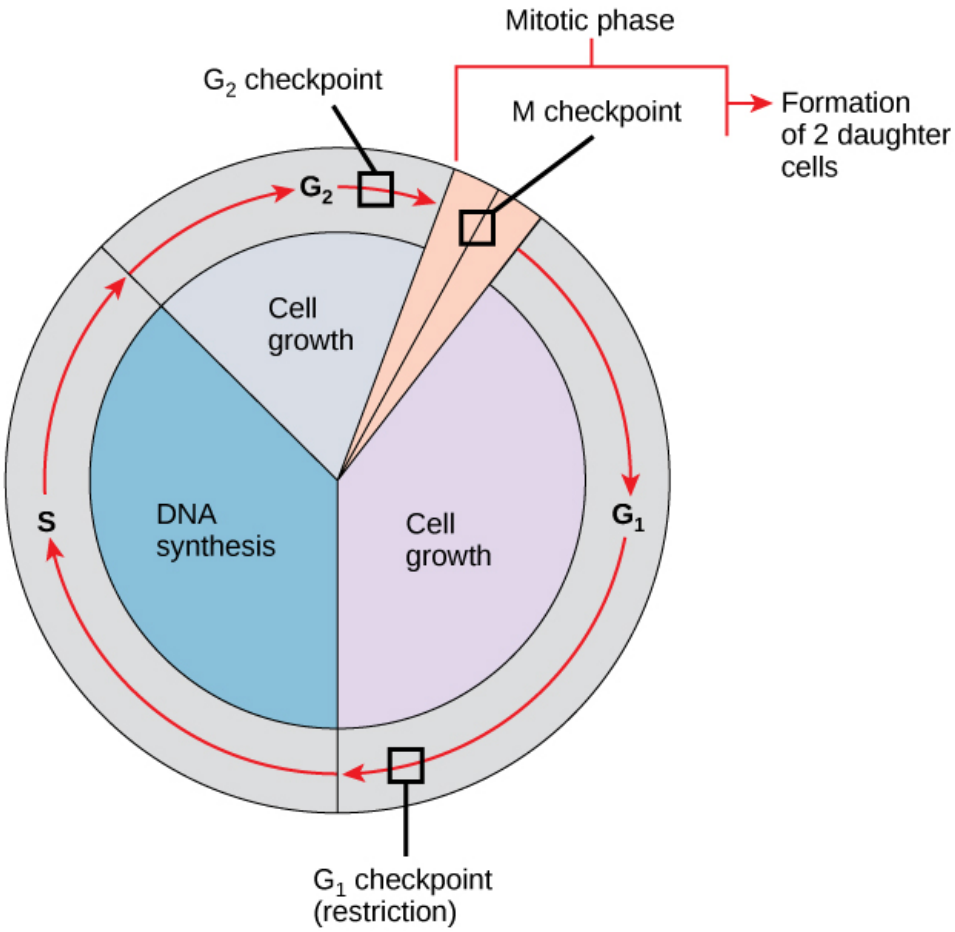

Figure 1. The cell cycle is controlled at three checkpoints. The integrity of the DNA is assessed at the G_1 checkpoint. Proper chromosome duplication is assessed at the G_2 checkpoint. Attachment of each kinetochore to a spindle fiber is assessed at the M checkpoint.

In Summary: Control of the Cell Cycle

Each step of the cell cycle is monitored by internal controls called checkpoints. There are three major checkpoints in the cell cycle: one near the end of G_1, a second at the G_2/M transition, and the third during metaphase. Positive regulator molecules allow the cell cycle to advance to the next stage. Negative regulator molecules monitor cellular conditions and can halt the cycle until specific requirements are met.

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CANCER AND THE CELL CYCLE

Learning Outcomes

Explain how errors in cell division are related to cancer

Cancer comprises many different diseases caused by a common mechanism: uncontrolled cell growth. Despite the redundancy and overlapping levels of cell cycle control, errors do occur. One of the critical processes monitored by the cell cycle checkpoint surveillance mechanism is the proper replication of DNA during the S phase. Even when all of the cell cycle controls are fully functional, a small percentage of replication errors (mutations) will be passed on to the daughter cells. If changes to the DNA nucleotide sequence occur within a coding portion of a gene and are not corrected, a gene mutation results. All cancers start when a gene mutation gives rise to a faulty protein that plays a key role in cell reproduction. Eventually, the pace of the cell cycle speeds up as the effectiveness of the control and repair mechanisms decreases. Uncontrolled growth of the mutated cells outpaces the growth of normal cells in the area, and a tumor (~*oma*) can result.

Proto-oncogenes

The genes that code for the positive cell cycle regulators are called **proto-oncogenes**. Proto-oncogenes are normal genes that, when mutated in certain ways, become **oncogenes**, genes that cause a cell to become cancerous.

Tumor Suppressor Genes

Like proto-oncogenes, many of the negative cell cycle regulatory proteins were discovered in cells that had become cancerous. **Tumor suppressor genes** are segments of DNA that code for negative regulator proteins, the type of regulators that, when activated, can prevent the cell from undergoing uncontrolled division. A cell that carries a mutated form of a negative regulator might not be able to halt the cell cycle if there is a problem. Tumor suppressors are similar to brakes in a vehicle: malfunctioning brakes can contribute to a car crash. Mutated p53 genes have been identified in more than one-half of all human tumor cells.

In Summary: Cancer and the Cell Cycle

This video reviews the ways that cancer is a by-product of broken DNA replication:

Watch this video online: https://youtu.be/RZhL7LDPk8w

Cancer is the result of unchecked cell division caused by a breakdown of the mechanisms that regulate the cell cycle. The loss of control begins with a change in the DNA sequence of a gene that codes for one of the regulatory molecules. Faulty instructions lead to a protein that does not function as it should. Any disruption of the monitoring system can allow other mistakes to be passed on to the daughter cells. Each successive cell division will give rise to daughter cells with even more accumulated damage. Eventually, all checkpoints become nonfunctional, and rapidly reproducing cells crowd out normal cells, resulting in a tumor or leukemia (blood cancer).

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- Cell cycle regulation, cancer, and stem cells. **Authored by**: Sal Khan. **Provided by**: Khan Academy. **Located at**: http://www.khanacademy.org/video?v=RZhL7LDPk8w. **Project**: Biology. **License**: *CC BY-NC-SA: Attribution-NonCommercial-ShareAlike*

INTRODUCTION TO SEXUAL REPRODUCTION

What you'll learn to do: Understand how sexual reproduction leads to different sexual life cycles

Living things can reproduce asexually (offspring have one parent) or sexually (offspring have two parents). In asexual reproduction, offspring are genetically identical to their parents; in sexual reproduction offspring have a mix of traits from their parents.

A vast majority of plants and animals reproduce sexually. Additionally, a portion of organisms that typically reproduce asexually can also reproduce sexually in the right circumstances. There are a variety of methods that living things can use to reproduce. In this outcome we will learn about the primary processes animals and plants use to reproduce, as well as the benefits to sexual reproduction as a whole.

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SEXUAL REPRODUCTION

Learning Outcomes

Understand how sexual reproduction leads to different sexual life cycles

Sexual reproduction was an early evolutionary innovation after the appearance of eukaryotic cells. It appears to have been very successful because most eukaryotes are able to reproduce sexually, and in many animals, it is the only mode of reproduction. And yet, scientists recognize some real disadvantages to sexual reproduction. On the surface, creating offspring that are genetic clones of the parent appears to be a better system. If the parent organism is successfully occupying a habitat, offspring with the same traits would be similarly successful. There is also the obvious benefit to an organism that can produce offspring whenever circumstances are favorable by asexual budding, fragmentation, or asexual eggs. These methods of reproduction do not require another organism of the opposite sex. Indeed, some organisms that lead a solitary lifestyle have retained the ability to reproduce asexually. In addition, in asexual populations, every individual is capable of reproduction. In sexual populations, the males are not producing the offspring themselves, so in theory an asexual population could grow twice as fast.

However, multicellular organisms that exclusively depend on asexual reproduction are exceedingly rare. Why is sexuality (and meiosis) so common? This is one of the important unanswered questions in biology and has been the focus of much research beginning in the latter half of the twentieth century. There are several possible explanations, one of which is that the variation that sexual reproduction creates among offspring is very important to the survival and reproduction of the population. Thus, on average, a sexually reproducing population will leave more descendants than an otherwise similar asexually reproducing population. The only source of variation in asexual organisms is mutation. This is the ultimate source of variation in sexual organisms, but in addition, those different mutations are continually reshuffled from one generation to the next when different parents combine their unique genomes and the genes are mixed into different combinations by crossovers during prophase I and random assortment at metaphase I.

The Red Queen Hypothesis

It is not in dispute that sexual reproduction provides evolutionary advantages to organisms that employ this mechanism to produce offspring. But why, even in the face of fairly stable conditions, does sexual reproduction persist when it is more difficult and costly for individual organisms? Variation is the outcome of sexual reproduction, but why are ongoing variations necessary? Enter the Red Queen hypothesis, first proposed by Leigh Van Valen in 1973. The concept was named in reference to the Red Queen's race in Lewis Carroll's book, *Through the Looking-Glass*.

All species co-evolve with other organisms; for example predators evolve with their prey, and parasites evolve with their hosts. Each tiny advantage gained by favorable variation gives a species an edge over close competitors, predators, parasites, or even prey. The only method that will allow a co-evolving species to maintain its own share of the resources is to also continually improve its fitness. As one species gains an advantage, this increases selection on the other species; they must also develop an advantage or they will be outcompeted. No single species progresses too far ahead because genetic variation among the progeny of sexual reproduction provides all species with a mechanism to improve rapidly. Species that cannot keep up become extinct. The Red Queen's catchphrase was, "It takes all the running you can do to stay in the same place." This is an apt description of co-evolution between competing species.

Life Cycles of Sexually Reproducing Organisms

Fertilization and meiosis alternate in sexual **life cycles**. What happens between these two events depends on the organism. The process of meiosis reduces the chromosome number by half. Fertilization, the joining of two haploid gametes, restores the diploid condition. There are three main categories of life cycles in multicellular organisms: **diploid-dominant**, in which the multicellular diploid stage is the most obvious life stage, such as with most animals including humans; **haploid-dominant**, in which the multicellular haploid stage is the most obvious life stage, such as with all fungi and some algae; and **alternation of generations**, in which the two stages are apparent to different degrees depending on the group, as with plants and some algae.

Diploid-Dominant Life Cycle

Nearly all animals employ a diploid-dominant life-cycle strategy in which the only haploid cells produced by the organism are the gametes. Early in the development of the embryo, specialized diploid cells, called **germ cells**, are produced within the gonads, such as the testes and ovaries. Germ cells are capable of mitosis to perpetuate the cell line and meiosis to produce gametes. Once the haploid gametes are formed, they lose the ability to divide again. There is no multicellular haploid life stage. Fertilization occurs with the fusion of two gametes, usually from different individuals, restoring the diploid state (Figure 1).

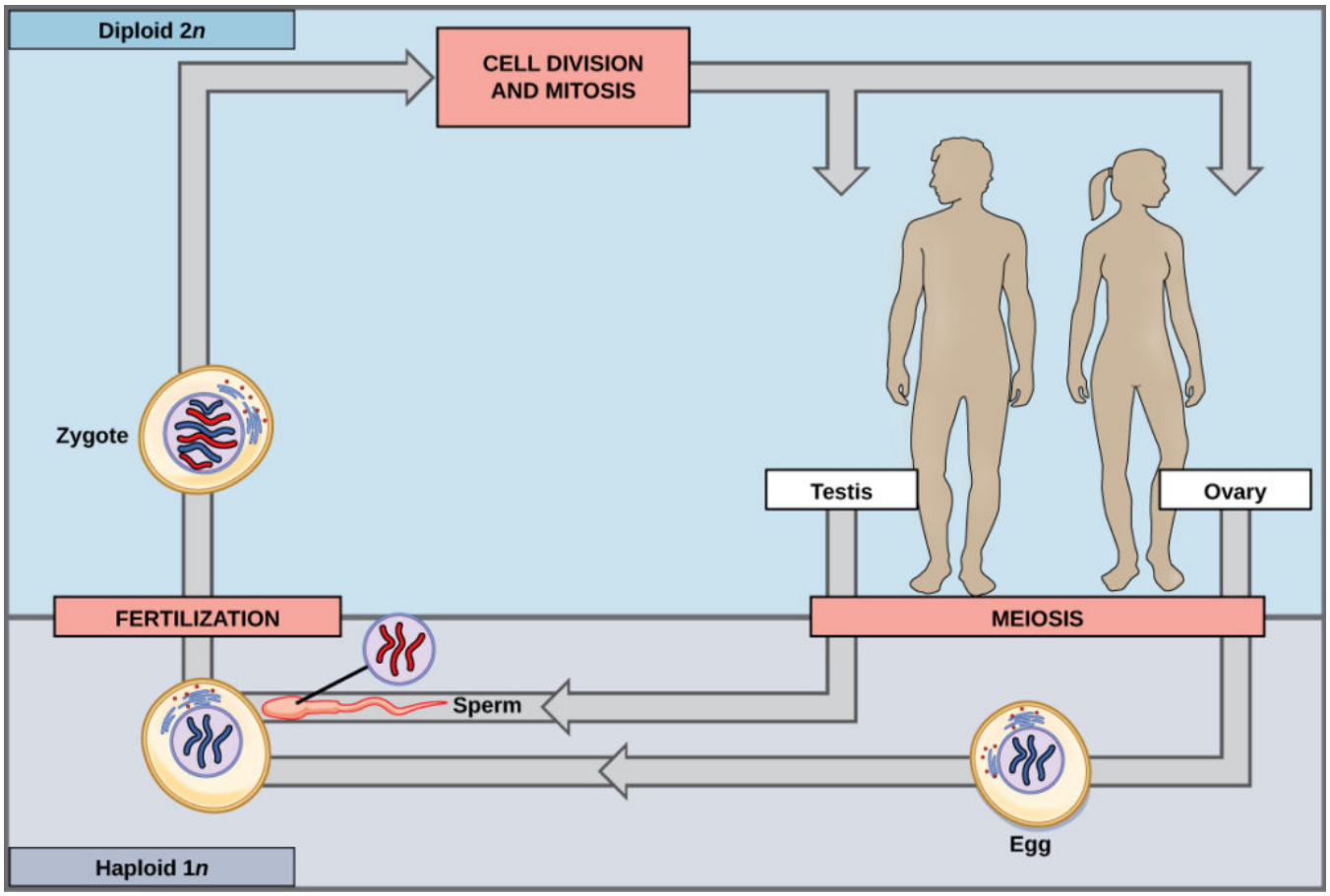

Figure 1. In animals, sexually reproducing adults form haploid gametes from diploid germ cells. Fusion of the gametes gives rise to a fertilized egg cell, or zygote. The zygote will undergo multiple rounds of mitosis to produce a multicellular offspring. The germ cells are generated early in the development of the zygote.

Haploid-Dominant Life Cycle

Most fungi and algae employ a life-cycle type in which the “body” of the organism—the ecologically important part of the life cycle—is haploid. The haploid cells that make up the tissues of the dominant multicellular stage are formed by mitosis. During sexual reproduction, specialized haploid cells from two individuals, designated the (+) and (−) mating types, join to form a diploid zygote. The zygote immediately undergoes meiosis to form four haploid cells called spores. Although haploid like the “parents,” these spores contain a new genetic combination from two parents. The spores can remain dormant for various time periods. Eventually, when conditions are conducive, the spores form multicellular haploid structures by many rounds of mitosis (Figure 2).

Practice Question

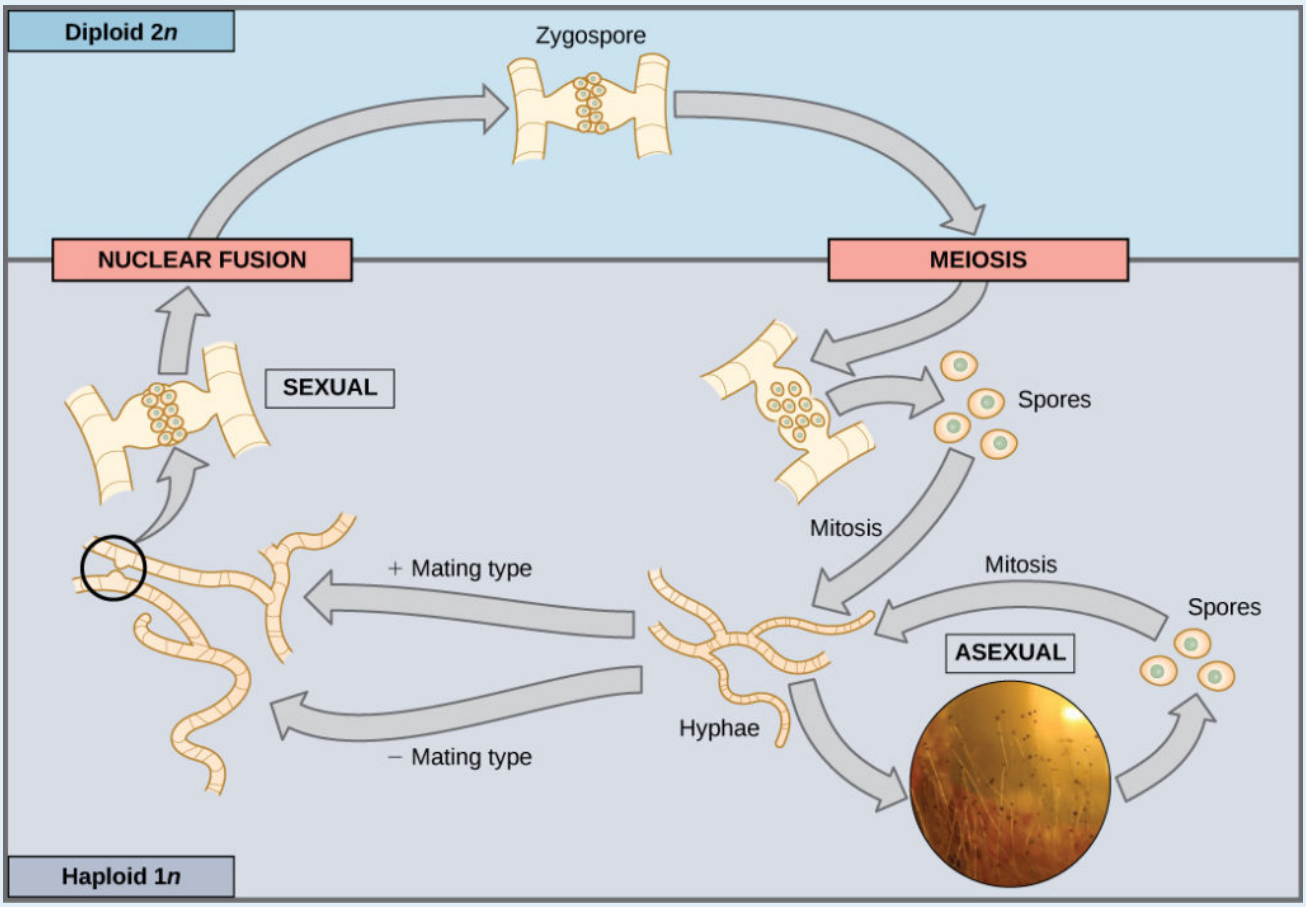

Figure 2. Fungi, such as black bread mold (Rhizopus nigricans), have haploid-dominant life cycles. The haploid multicellular stage produces specialized haploid cells by mitosis that fuse to form a diploid zygote. The zygote undergoes meiosis to produce haploid spores. Each spore gives rise to a multicellular haploid organism by mitosis. (credit "zygomycota" micrograph: modification of work by "Fanaberka"/Wikimedia Commons)

If a mutation occurs so that a fungus is no longer able to produce a minus mating type, will it still be able to reproduce?

Answer

Most likely yes as the fungus can likely reproduce asexually.

Alternation of Generations

The third life-cycle type, employed by some algae and all plants, is a blend of the haploid-dominant and diploid-dominant extremes. Species with alternation of generations have both haploid and diploid multicellular organisms as part of their life cycle. The haploid multicellular plants are called **gametophytes**, because they produce gametes from specialized cells. Meiosis is not directly involved in the production of gametes in this case, because the organism that produces the gametes is already a haploid. Fertilization between the gametes forms a diploid zygote. The zygote will undergo many rounds of mitosis and give rise to a diploid multicellular plant called a **sporophyte**. Specialized cells of the sporophyte will undergo meiosis and produce haploid spores. The spores will subsequently develop into the gametophytes (Figure 3).

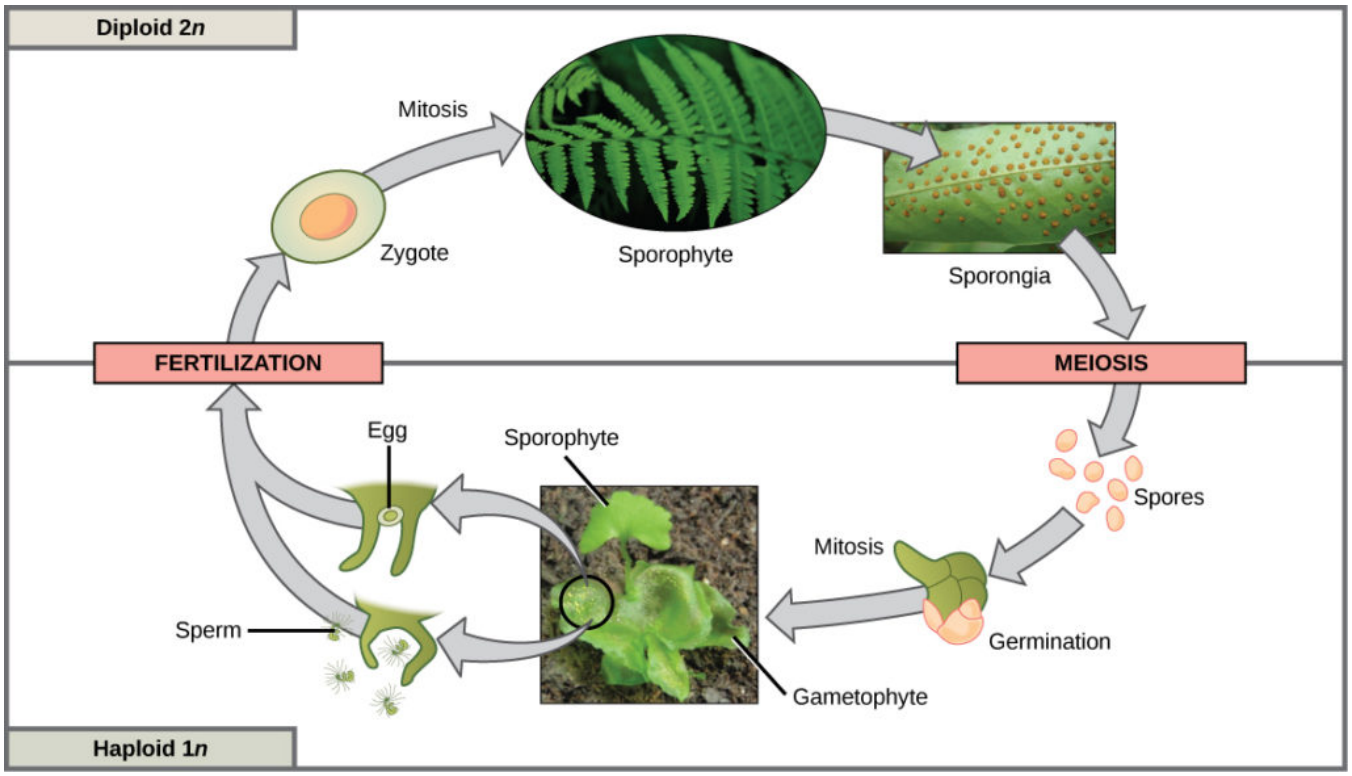

Figure 3. Plants have a life cycle that alternates between a multicellular haploid organism and a multicellular diploid organism. In some plants, such as ferns, both the haploid and diploid plant stages are free-living. The diploid plant is called a sporophyte because it produces haploid spores by meiosis. The spores develop into multicellular, haploid plants called gametophytes because they produce gametes. The gametes of two individuals will fuse to form a diploid zygote that becomes the sporophyte. (credit "fern": modification of work by Cory Zanker; credit "sporangia": modification of work by "Obsidian Soul"/Wikimedia Commons; credit "gametophyte and sporophyte": modification of work by "Vlmastra"/Wikimedia Commons)

Although all plants utilize some version of the alternation of generations, the relative size of the sporophyte and the gametophyte and the relationship between them vary greatly. In plants such as moss, the gametophyte organism is the free-living plant, and the sporophyte is physically dependent on the gametophyte. In other plants, such as ferns, both the gametophyte and sporophyte plants are free-living; however, the sporophyte is much larger. In seed plants, such as magnolia trees and daisies, the gametophyte is composed of only a few cells and, in the case of the female gametophyte, is completely retained within the sporophyte.

Sexual reproduction takes many forms in multicellular organisms. However, at some point in each type of life cycle, meiosis produces haploid cells that will fuse with the haploid cell of another organism. The mechanisms of variation—crossover, random assortment of homologous chromosomes, and random fertilization—are present in all versions of sexual reproduction. The fact that nearly every multicellular organism on Earth employs sexual reproduction is strong evidence for the benefits of producing offspring with unique gene combinations, though there are other possible benefits as well.

In Summary: Sexual Reproduction

Nearly all eukaryotes undergo sexual reproduction. The variation introduced into the reproductive cells by meiosis appears to be one of the advantages of sexual reproduction that has made it so successful. Meiosis and fertilization alternate in sexual life cycles. The process of meiosis produces unique reproductive cells called gametes, which have half the number of chromosomes as the parent cell. Fertilization, the fusion of haploid gametes from two individuals, restores the diploid condition. Thus, sexually reproducing organisms alternate between haploid and diploid stages. However, the ways in which reproductive cells are produced and the timing between meiosis and fertilization vary greatly. There are three main categories of life cycles: diploid-dominant, demonstrated by most animals; haploid-dominant, demonstrated by all fungi and some algae; and the alternation of generations, demonstrated by plants and some algae.

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INTRODUCTION TO MEIOSIS

What you'll learn to do: Identify the stages of meiosis by picture and by description of major milestones; explain why meiosis involves two rounds of nuclear division

Just what is the difference between mitosis and meiosis? We know that mitosis produces autosomes; meiosis produces sex chromosomes. Let's learn just how similar and different these processes are.

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STAGES OF MEIOSIS

Learning Outcomes

Identify the stages of meiosis

The ability to reproduce *in kind* is a basic characteristic of all living things. *In kind* means that the offspring of any organism closely resemble their parent or parents. Hippopotamuses give birth to hippopotamus calves, Joshua trees produce seeds from which Joshua tree seedlings emerge, and adult flamingos lay eggs that hatch into flamingo chicks. *In kind* does not generally mean *exactly the same*.

(a) (b) (c)

Figure 1. Each of us, like these other large multicellular organisms, begins life as a fertilized egg. After trillions of cell divisions, each of us develops into a complex, multicellular organism. (credit a: modification of work by Frank Wouters; credit b: modification of work by Ken Cole, USGS; credit c: modification of work by Martin Pettitt)

As you have learned, mitosis is the part of a cell reproduction cycle that results in identical daughter nuclei that are also genetically identical to the original parent nucleus. In mitosis, both the parent and the daughter nuclei are at the same ploidy level—diploid for most plants and animals. While many unicellular organisms and a few multicellular organisms can produce genetically identical clones of themselves through mitosis, many single-celled organisms and most multicellular organisms reproduce regularly using another method: **meiosis**. Sexual reproduction, specifically meiosis and fertilization, introduces variation into offspring that may account for the evolutionary success of sexual reproduction. The vast majority of eukaryotic organisms, both multicellular and unicellular, can or must employ some form of meiosis and fertilization to reproduce.

Meiosis employs many of the same mechanisms as mitosis. However, the starting nucleus is always diploid and the nuclei that result at the end of a meiotic cell division are haploid. To achieve this reduction in chromosome number, meiosis consists of one round of chromosome duplication and two rounds of nuclear division.

Because the events that occur during each of the division stages are analogous to the events of mitosis, the same stage names are assigned. However, because there are two rounds of division, the major process and the stages are designated with a "I" or a "II." Thus, **meiosis I** is the first round of meiotic division and consists of prophase I, prometaphase I, and so on. **Meiosis II**, in which the second round of meiotic division takes place, includes prophase II, prometaphase II, and so on.

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MEIOSIS I

Learning Outcomes

Describe the steps of meiosis I

Meiosis is preceded by an interphase consisting of the G_1, S, and G_2 phases, which are nearly identical to the phases preceding mitosis. The G_1 phase, which is also called the first gap phase, is the first phase of the interphase and is focused on cell growth. The S phase is the second phase of interphase, during which the DNA of the chromosomes is replicated. Finally, the G_2 phase, also called the second gap phase, is the third and final phase of interphase; in this phase, the cell undergoes the final preparations for meiosis.

During DNA duplication in the S phase, each chromosome is replicated to produce two identical copies, called sister chromatids, that are held together at the centromere. The centrosomes, which are the structures that organize the microtubules of the meiotic spindle, also replicate. This prepares the cell to enter prophase I, the first meiotic phase.

Prophase I

As the nuclear envelope begins to break down, the proteins associated with homologous chromosomes bring the pair close to each other. (Recall that, in mitosis, homologous chromosomes do not pair together. In mitosis, homologous chromosomes line up end-to-end so that when they divide, each daughter cell receives a sister chromatid from both members of the homologous pair.) The tight pairing of the homologous chromosomes is called **synapsis**. In synapsis, the genes on the chromatids of the homologous chromosomes are aligned precisely with each other. The synaptonemal complex supports the exchange of chromosomal segments between non-sister homologous chromatids, a process called **crossing over**. Crossing over can be observed visually after the exchange as **chiasmata** (singular = *chiasma*) (Figure 1).

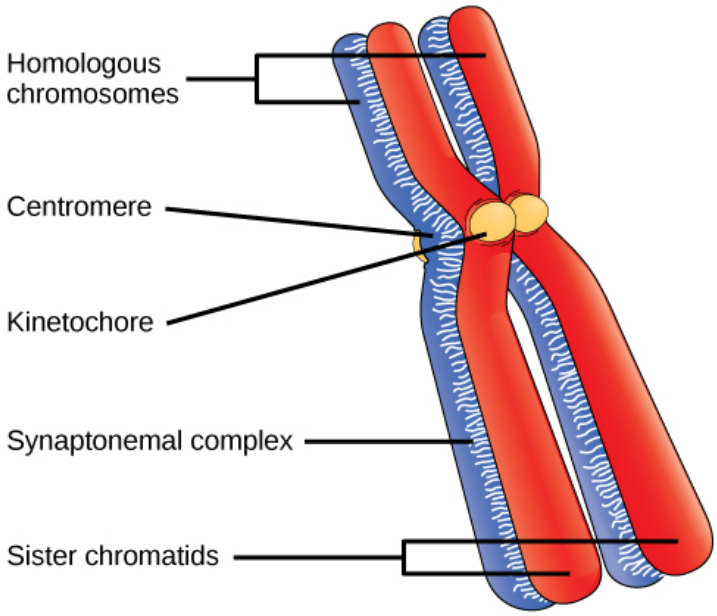

Figure 1. Early in prophase I, homologous chromosomes come together to form a synapse. The chromosomes are bound tightly together and in perfect alignment by a protein lattice called a synaptonemal complex and by cohesin proteins at the centromere.

At the end of prophase I, the pairs are held together only at the chiasmata (Figure 2) and are called **tetrads** because the four sister chromatids of each pair of homologous chromosomes are now visible.

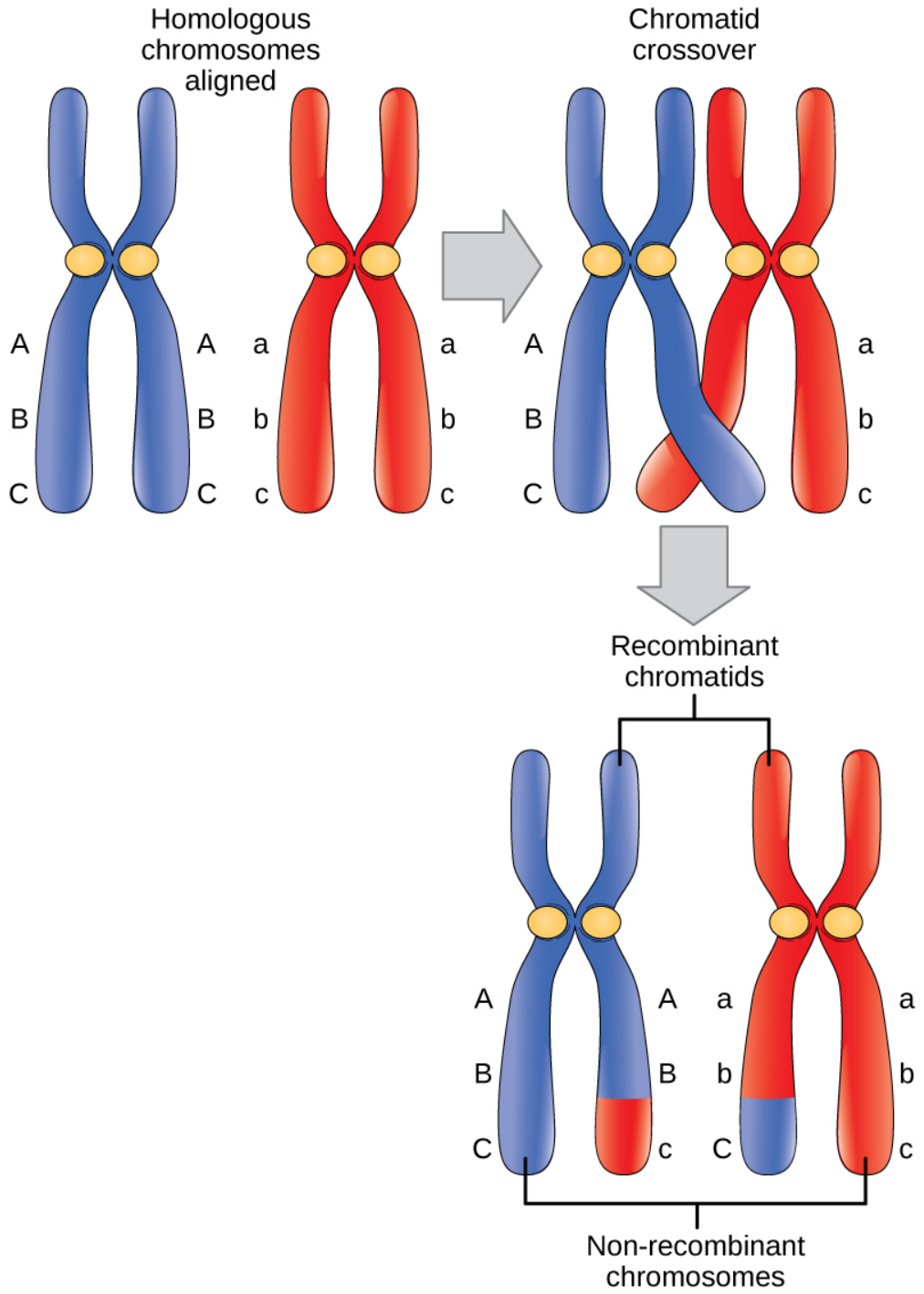

Figure 2. Crossover occurs between non-sister chromatids of homologous chromosomes. The result is an exchange of genetic material between homologous chromosomes.

The crossover events are the first source of genetic variation in the nuclei produced by meiosis. A single crossover event between homologous non-sister chromatids leads to a reciprocal exchange of equivalent DNA between a maternal chromosome and a paternal chromosome. Now, when that sister chromatid is moved into a gamete cell it will carry some DNA from one parent of the individual and some DNA from the other parent. Multiple crossovers in an arm of the chromosome have the same effect, exchanging segments of DNA to create recombinant chromosomes.

A second event in Prophase I is the attachment of the spindle fiber microtubules to the kinetochore proteins at the centromeres. At the end of prometaphase I, each tetrad is attached to microtubules from both poles, with one homologous chromosome facing each pole. The homologous chromosomes are still held together at chiasmata.

In addition, the nuclear membrane has broken down entirely.

Metaphase I

During metaphase I, the homologous chromosomes are arranged in the center of the cell with the kinetochores facing opposite poles. The homologous pairs orient themselves randomly at the equator. Recall that homologous chromosomes are not identical. They contain slight differences in their genetic information, causing each gamete to have a unique genetic makeup. **This randomness is the physical basis for the creation of the second form of genetic variation in offspring**. The number of variations is dependent on the number of chromosomes making up a set. There are two possibilities for orientation at the metaphase plate; the possible number of alignments therefore equals 2^n, where n is the number of chromosomes per set. Humans have 23 chromosome pairs, which results in over eight million (2^{23}) possible genetically-distinct gametes. This number does not include the variability that was previously created in the sister chromatids by crossover. Given these two mechanisms, it is highly unlikely that any two haploid cells resulting from meiosis will have the same genetic composition (Figure 3).

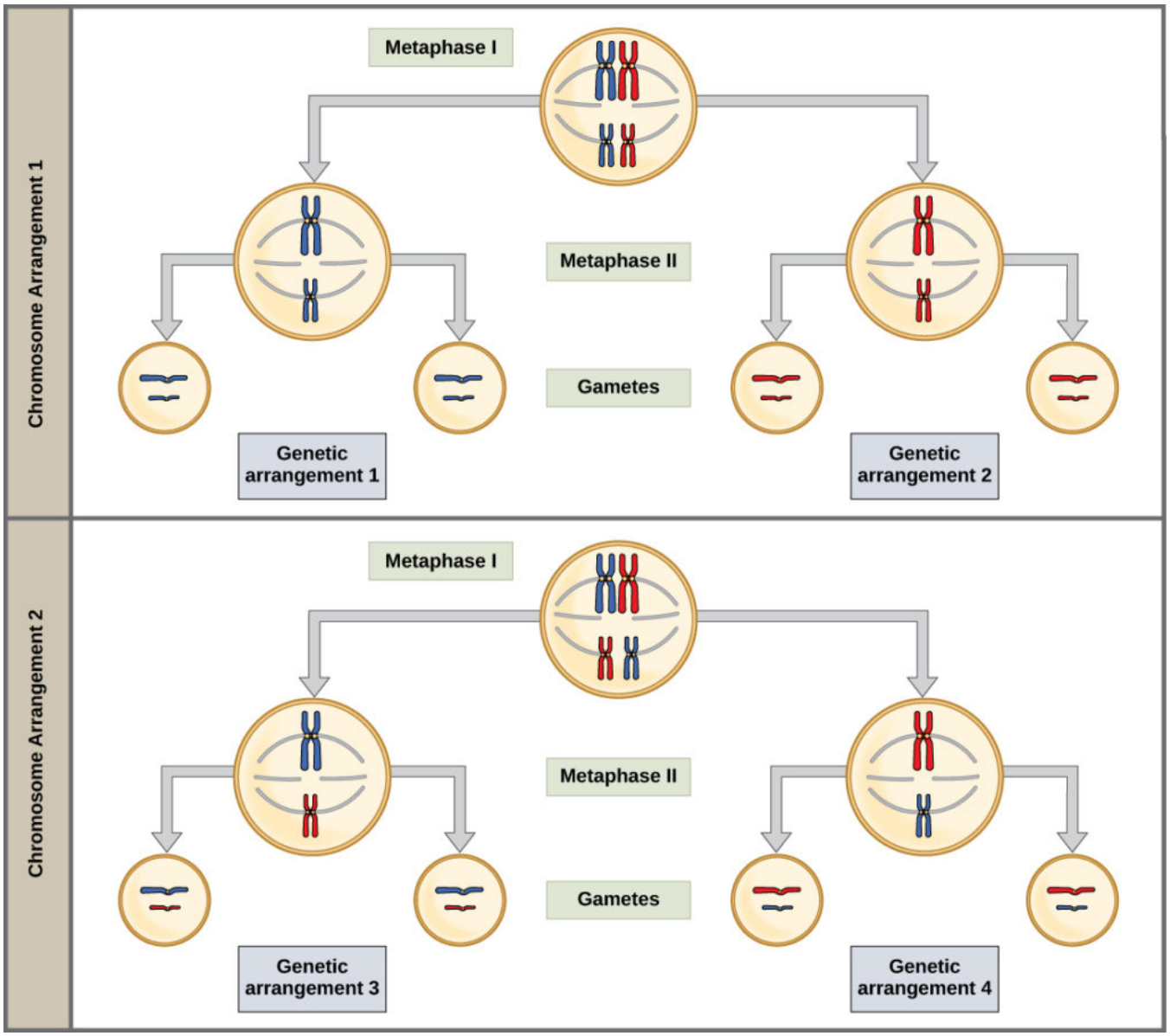

Figure 3. Random, independent assortment during metaphase I can be demonstrated by considering a cell with a set of two chromosomes (n = 2). In this case, there are two possible arrangements at the equatorial plane in metaphase I. The total possible number of different gametes is 2n, where n equals the number of chromosomes in a set. In this example, there are four possible

genetic combinations for the gametes. With n = 23 in human cells, there are over 8 million possible combinations of paternal and maternal chromosomes.

To summarize the genetic consequences of meiosis I, the maternal and paternal genes are recombined by crossover events that occur between each homologous pair during prophase I. In addition, the random assortment of tetrads on the metaphase plate produces a unique combination of maternal and paternal chromosomes that will make their way into the gametes.

Anaphase I

In anaphase I, the microtubules pull the linked chromosomes apart. The sister chromatids remain tightly bound together a the centromere. The chiasmata are broken in anaphase I as the microtubules attached to the fused kinetochores pull the homologous chromosomes apart (Figure 4).

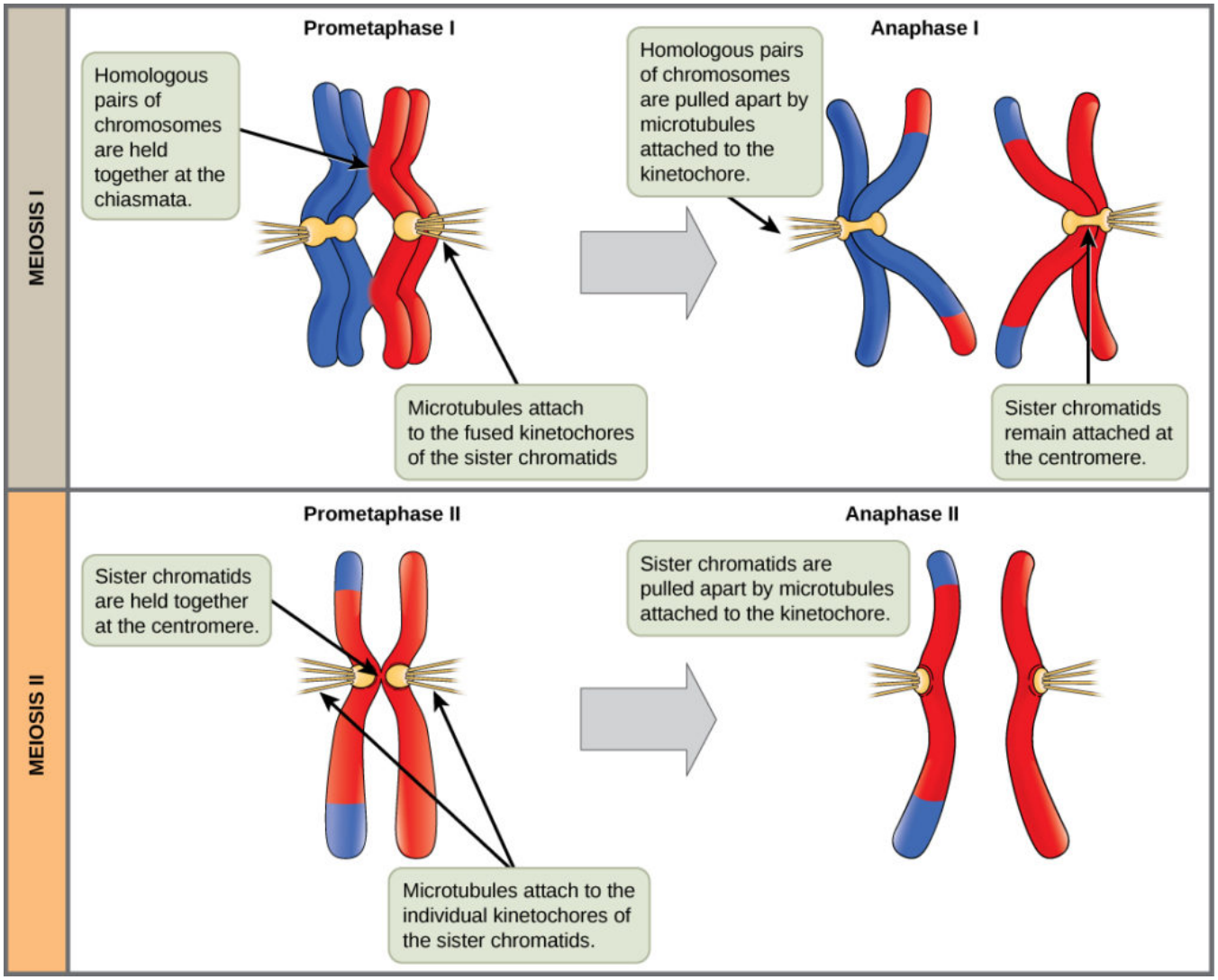

Figure 4. The process of chromosome alignment differs between meiosis I and meiosis II. In prometaphase I, microtubules attach to the fused kinetochores of homologous chromosomes, and the homologous chromosomes are arranged at the midpoint of the cell in metaphase I. In anaphase I, the homologous chromosomes are separated. In prometaphase II, microtubules attach to the kinetochores of sister chromatids, and the sister chromatids are arranged at the midpoint of the cells in metaphase II. In anaphase II, the sister chromatids are separated.

Telophase I and Cytokinesis

In telophase, the separated chromosomes arrive at opposite poles. The remainder of the typical telophase events may or may not occur, depending on the species. In some organisms, the chromosomes decondense and nuclear envelopes form around the chromatids in telophase I. In other organisms, cytokinesis—the physical separation of the cytoplasmic components into two daughter cells—occurs without reformation of the nuclei. In nearly all species of animals and some fungi, cytokinesis separates the cell contents via a cleavage furrow (constriction of the actin ring that leads to cytoplasmic division). In plants, a cell plate is formed during cell cytokinesis by Golgi vesicles fusing at the metaphase plate. This cell plate will ultimately lead to the formation of cell walls that separate the two daughter cells.

Two haploid cells are the end result of the first meiotic division. The cells are haploid because at each pole, there is just one of each pair of the homologous chromosomes. Therefore, only one full set of the chromosomes is present. This is why the cells are considered haploid—there is only one chromosome set, even though each homolog still consists of two sister chromatids. Recall that sister chromatids are merely duplicates of one of the two homologous chromosomes (except for changes that occurred during crossing over). In meiosis II, these two sister chromatids will separate, creating four haploid daughter cells.

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MEIOSIS II

Learning Outcomes

Describe the steps of meiosis II

In some species, cells enter a brief interphase, or **interkinesis**, before entering meiosis II. Interkinesis lacks an S phase, so chromosomes are not duplicated. The two cells produced in meiosis I go through the events of meiosis II in synchrony. During meiosis II, the sister chromatids within the two daughter cells separate, forming four new haploid gametes. The mechanics of meiosis II is similar to mitosis, except that each dividing cell has only one set of homologous chromosomes. Therefore, each cell has half the number of sister chromatids to separate out as a diploid cell undergoing mitosis.

Prophase II

If the chromosomes decondensed in telophase I, they condense again. If nuclear envelopes were formed, they fragment into vesicles. The centrosomes that were duplicated during interkinesis move away from each other toward opposite poles, and new spindles are formed. The nuclear envelopes are completely broken down, and the spindle is fully formed. Each sister chromatid forms an individual kinetochore that attaches to microtubules from opposite poles.

Metaphase II

The sister chromatids are maximally condensed and aligned at the equator of the cell.

Anaphase II

The sister chromatids are pulled apart by the kinetochore microtubules and move toward opposite poles. Non-kinetochore microtubules elongate the cell.

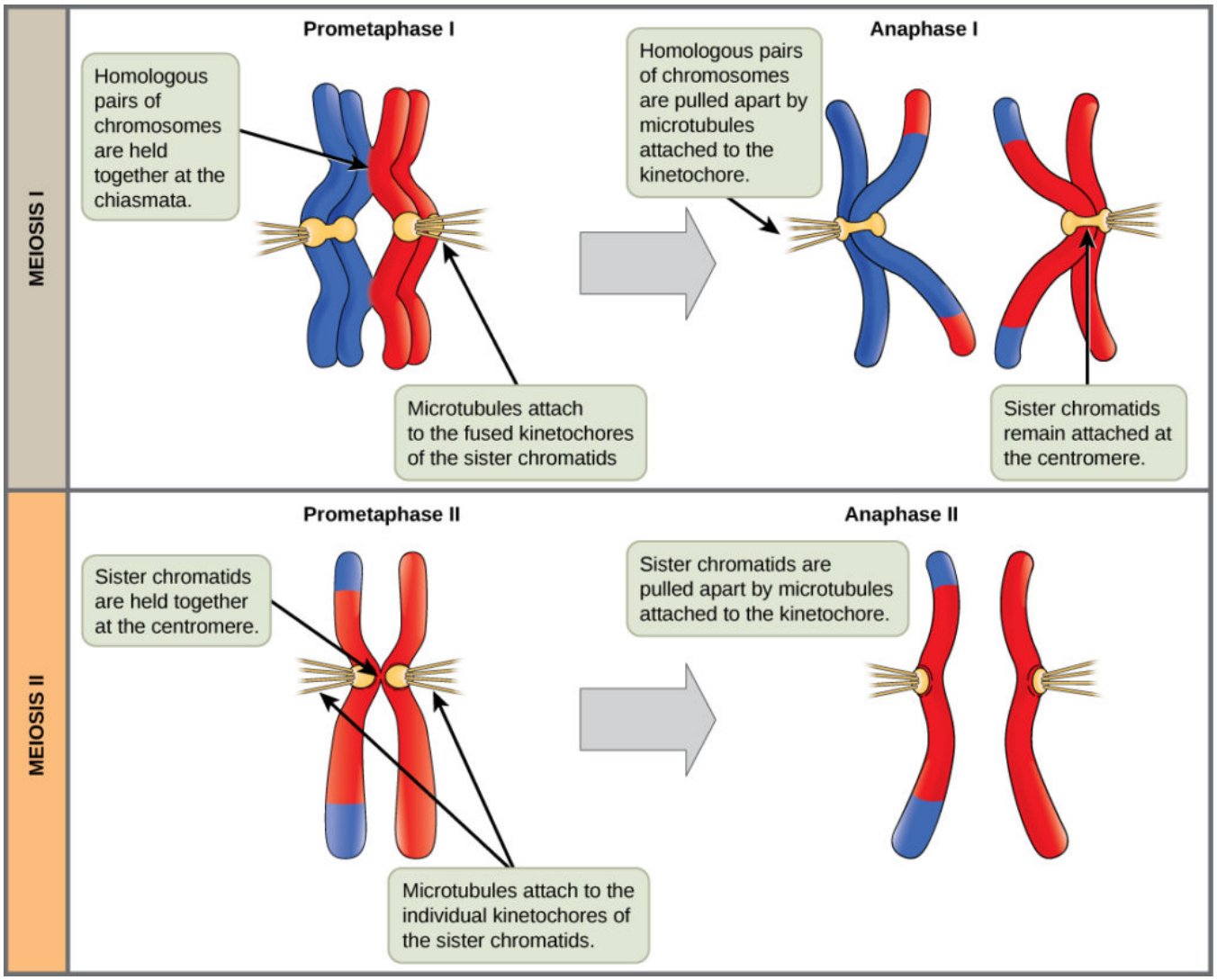


Figure 1. The process of chromosome alignment differs between meiosis I and meiosis II. In prometaphase I, microtubules attach to the fused kinetochores of homologous chromosomes, and the homologous chromosomes are arranged at the midpoint of the cell in metaphase I. In anaphase I, the homologous chromosomes are separated. In prometaphase II, microtubules attach to the kinetochores of sister chromatids, and the sister chromatids are arranged at the midpoint of the cells in metaphase II. In anaphase II, the sister chromatids are separated.

Telophase II and Cytokinesis

The chromosomes arrive at opposite poles and begin to decondense. Nuclear envelopes form around the chromosomes. Cytokinesis separates the two cells into four unique haploid cells. At this point, the newly formed nuclei are both haploid. The cells produced are genetically unique because of the random assortment of paternal and maternal homologs and because of the recombining of maternal and paternal segments of chromosomes (with their sets of genes) that occurs during crossover. The entire process of meiosis is outlined in Figure 2.

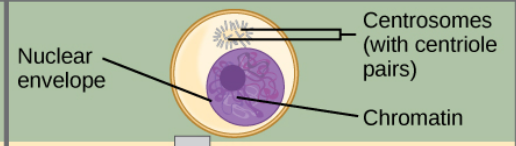
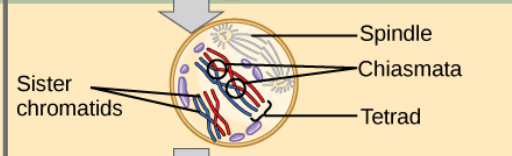
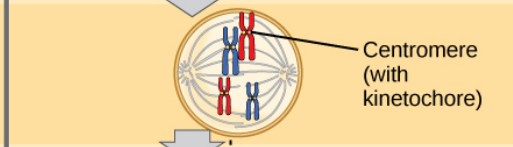
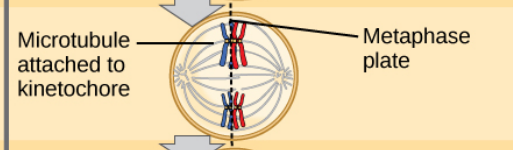
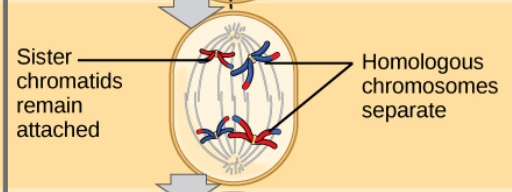
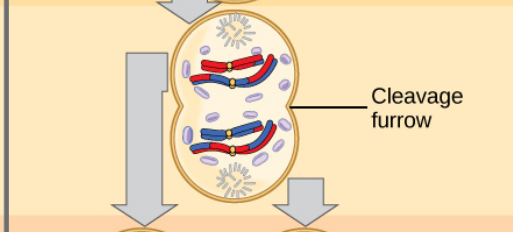
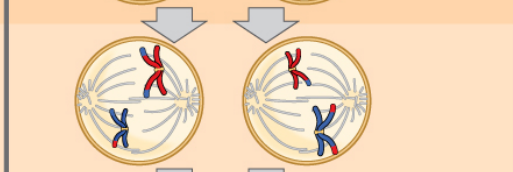
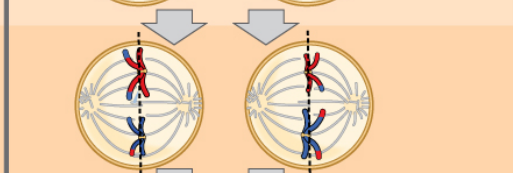
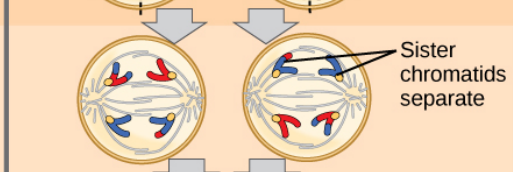
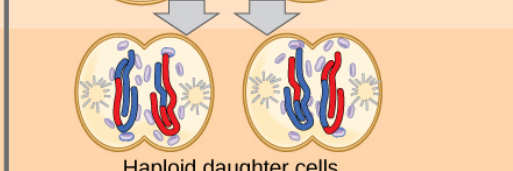
	Stage	Event	Outcome
INTERPHASE	S phase		Chromosomes are duplicated during interphase. The resulting sister chromatids are held together at the centromere. The centrosomes are also duplicated.
MEIOSIS I	Prophase I		Chromosomes condense, and the nuclear envelope fragments. Homologous chromosomes bind firmly together along their length, forming a tetrad. Chiasmata form between non-sister chromatids. Crossing over occurs at the chiasmata. Spindle fibers emerge from the centrosomes.
	Prometaphase I		Homologous chromosomes are attached to spindle microtubules at the fused kinetochore shared by the sister chromatids. Chromosomes continue to condense, and the nuclear envelope completely disappears.
	Metaphase I		Homologous chromosomes randomly assemble at the metaphase plate, where they have been maneuvered into place by the microtubules.
	Anaphase I		Spindle microtubules pull the homologous chromosomes apart. The sister chromatids are still attached at the centromere.
	Telophase I and Cytokinesis		Sister chromatids arrive at the poles of the cell and begin to decondense. A nuclear envelope forms around each nucleus and the cytoplasm is divided by a cleavage furrow. The result is two haploid cells. Each cell contains one duplicated copy of each homologous chromosome pair.
MEIOSIS II	Prophase II		Sister chromatids condense. A new spindle begins to form. The nuclear envelope starts to fragment.
	Prometaphase II		The nuclear envelope disappears, and the spindle fibers engage the individual kinetochores on the sister chromatids.
	Metaphase II		Sister chromatids line up at the metaphase plate.
	Anaphase II		Sister chromatids are pulled apart by the shortening of the kinetochore microtubules. Non-kinetochore microtubules lengthen the cell.
	Telophase II and Cytokinesis		Chromosomes arrive at the poles of the cell and decondense. Nuclear envelopes surround the four nuclei. Cleavage furrows divide the two cells into four haploid cells.

Figure 2. An animal cell with a diploid number of four (2n = 4) proceeds through the stages of meiosis to form four haploid daughter cells.

Review the process of meiosis, observing how chromosomes align and migrate, at Meiosis: An Interactive Animation.

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MEIOSIS: THE COMPLETE CYCLE

Learning Outcomes

- Identify the stages of meiosis
- Describe the steps of meiosis I
- Describe the steps of meiosis II

This video walks you through meiosis I and meiosis II:

Watch this video online: https://youtu.be/qCLmR9-YY7o

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INTRODUCTION TO GENETIC DIVERSITY

What you'll learn to do: Describe and explain a range of mechanisms for generating genetic diversity

Now that we know how meiosis works, let's see how it and its involved processes contribute to genetic diversity.

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GENETIC VARIATION IN MEIOSIS

Learning Outcomes

Understand how meiosis contributes to genetic diversity

The gametes produced in meiosis aren't genetically identical to the starting cell, and they also aren't identical to one another. As an example, consider the meiosis II diagram above, which shows the end products of meiosis for a simple cell with a diploid number of $2n = 4$ chromosomes. The four gametes produced at the end of meiosis II are all slightly different, each with a unique combination of the genetic material present in the starting cell.

As it turns out, there are many more potential gamete types than just the four shown in the diagram, even for a simple cell with with only four chromosomes. This diversity of possible gametes reflects two factors: crossing over and the random orientation of homologue pairs during metaphase of meiosis I.

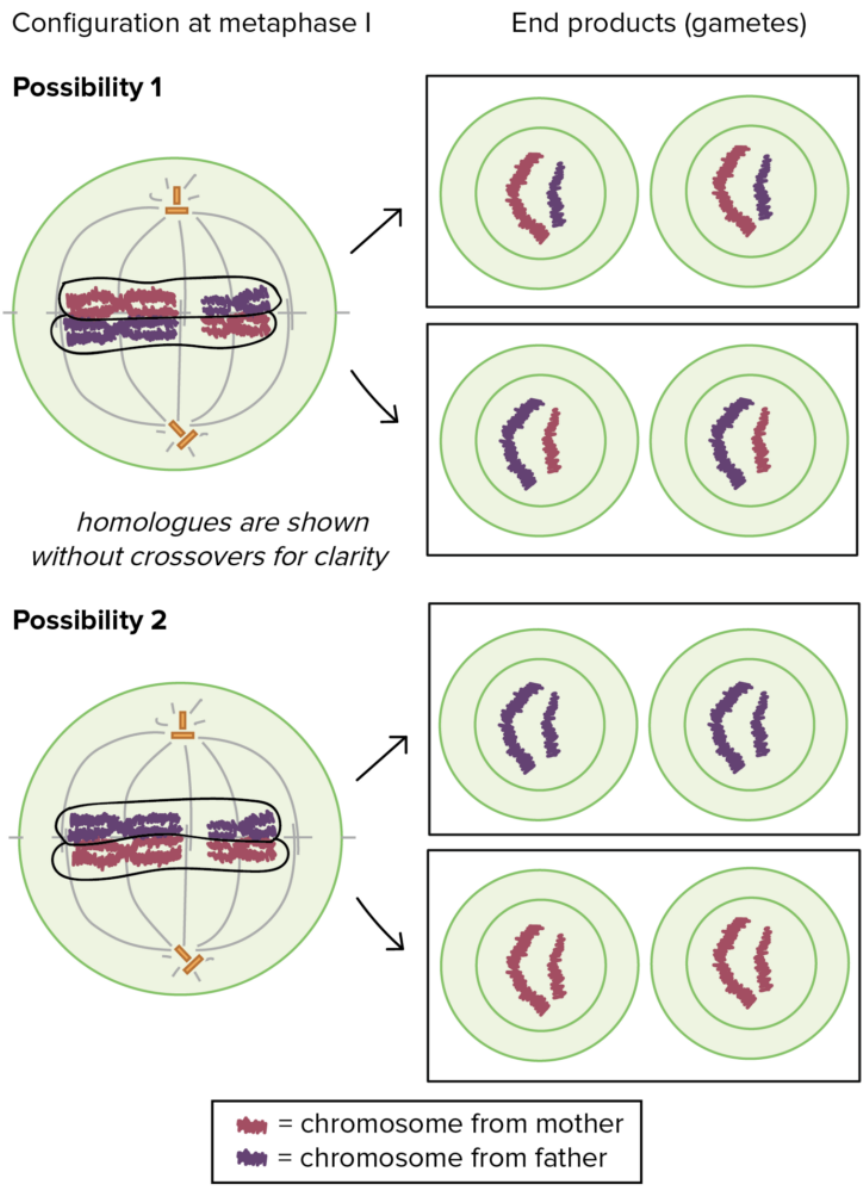
- Crossing over. The points where homologues cross over and exchange genetic material are chosen more or less at random, and they will be different in each cell that goes through meiosis. If meiosis happens many times, as it does in human ovaries and testes, crossovers will happen at many different points. This repetition produces a wide variety of recombinant chromosomes, chromosomes where fragments of DNA have been exchanged between homologues.
- Random orientation of homologue pairs. The random orientation of homologue pairs during metaphase of meiosis I is another important source of gamete diversity.

What exactly does *random orientation* mean here? Well, a homologous pair consists of one homologue from your dad and one from your mom, and you have 23 pairs of homologous chromosomes all together, counting the X and Y as homologous for this purpose. During meiosis I, the homologous pairs will separate to form two equal groups, but it's not usually the case that all the paternal—dad—chromosomes will go into one group and all the maternal—mom—chromosomes into the other.

Instead, each pair of homologues will effectively flip a coin to decide which chromosome goes into which group. In a cell with just two pairs of homologous chromosomes, like the one at right, random metaphase orientation allows for $2^2 = 4$ different types of possible gametes. In a human cell, the same mechanism allows for $2^{23} = 8{,}388{,}608$ different types of possible gametes (Note: Reece, J. B., L. A. Urry, M. L. Cain, S. A. Wasserman, P. V. Minorksy, and R. B. Jackson. "Genetic Variation Produced in Sexual Life Cycles Contributes to Evolution." In *Campbell Biology*, 263–65. 10th ed. San Francisco, CA: Pearson, 2011.). And that's not even considering crossovers!

Given those kinds of numbers, it's very unlikely that any two sperm or egg cells made by a person will be the same. It's even more unlikely that you and your sister or brother will be genetically identical, unless you happen to be identical twins, thanks to the process of fertilization (in which a unique egg from Mom combines with a unique sperm from Dad, making a zygote whose genotype is well beyond one-in-a-trillion!) (Note: Ibid.).

Meiosis and fertilization create genetic variation by making new combinations of gene variants (alleles). In some cases, these new combinations may make an organism more or less fit (able to survive and reproduce), thus providing the raw material for natural selection. Genetic variation is important in allowing a population to adapt via natural selection and thus survive in the long term.



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MITOSIS, MEIOSIS, AND SEXUAL REPRODUCTION

Learning Outcomes

Understand how mitosis, meiosis, and random fertilization all result in genetically unique individuals

As you now know, genetic variation is very important. Genetic variation is introduced in multiple ways, including changes in mitosis, crossing over and random orientation in meiosis, and random fertilization. The video below offers you a nice overview of how each contributes to genetic diversity.

Watch this video online: https://youtu.be/kaSIjIzAtYA

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INTRODUCTION TO ERRORS IN CHROMOSOME NUMBER

What you'll learn to do: Examine karyotypes and identify the effects of significant changes in chromosome number

We previously learned how errors in mitosis can potentially lead to cancer. What could errors in meiosis result in? In this outcome, we'll learn what happens when errors occur in chromosome number.

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KARYOTYPES

Learning Outcomes

Identify a karyotype and describe its uses in biology

The isolation and microscopic observation of chromosomes forms the basis of cytogenetics and is the primary method by which clinicians detect chromosomal abnormalities in humans. A **karyotype** is the number and appearance of chromosomes, and includes their length, banding pattern, and centromere position. To obtain a view of an individual's karyotype, cytologists photograph the chromosomes and then cut and paste each chromosome into a chart, or **karyogram**, also known as an ideogram (Figure 1). The simplest use of a karyotype (or its karyogram image) is to identify abnormal chromosomal numbers.

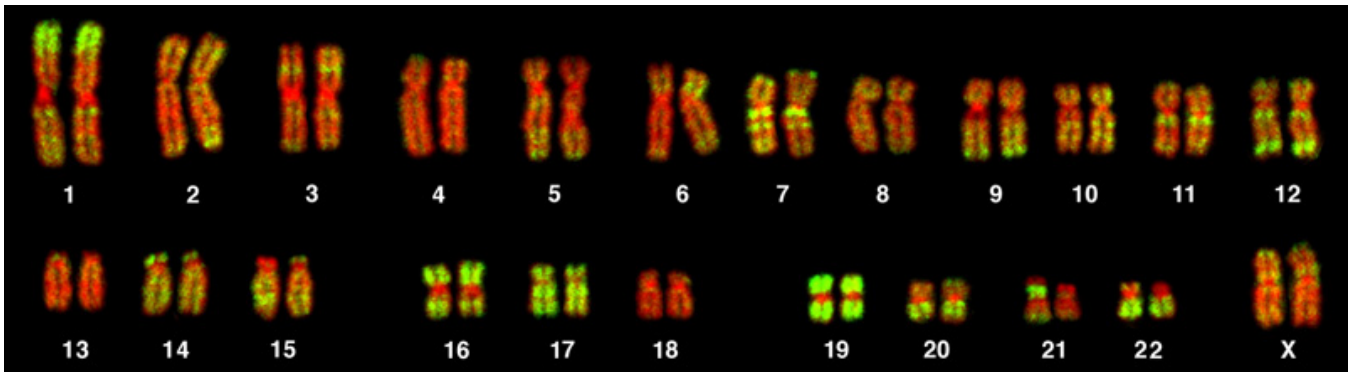

Figure 1. This karyotype is of a female human. Notice that homologous chromosomes are the same size, and have the same centromere positions and banding patterns. A human male would have an XY chromosome pair instead of the XX pair shown. (credit: Andreas Blozer et al)

In a given species, chromosomes can be identified by their number, size, centromere position, and banding pattern. In a human karyotype, **autosomes** or "body chromosomes" (all of the non–sex chromosomes) are generally organized in approximate order of size from largest (chromosome 1) to smallest (chromosome 22). The X and Y chromosomes are not autosomes. However, chromosome 21 is actually shorter than chromosome 22. This was discovered after the naming of Down syndrome as trisomy 21, reflecting how this disease results from possessing one extra chromosome 21 (three total). Not wanting to change the name of this important disease, chromosome 21 retained its numbering, despite describing the shortest set of chromosomes. The chromosome "arms" projecting from either end of the centromere may be designated as short or long, depending on their relative lengths. The short arm is abbreviated *p* (for "petite"), whereas the long arm is abbreviated *q* (because it follows "p" alphabetically). Each arm is further subdivided and denoted by a number. Using this naming system, locations on chromosomes can be described consistently in the scientific literature.

Geneticists Use Karyograms to Identify Chromosomal Aberrations

Although Mendel is referred to as the "father of modern genetics," he performed his experiments with none of the tools that the geneticists of today routinely employ. One such powerful cytological technique is karyotyping, a method in which traits characterized by chromosomal abnormalities can be identified from a single cell. To observe an individual's karyotype, a person's cells (like white blood cells) are first collected from a blood sample or other tissue. In the laboratory, the isolated cells are stimulated to begin actively dividing. A chemical called colchicine is then applied to cells to arrest condensed chromosomes in metaphase. Cells are then made to swell using a hypotonic solution so the chromosomes spread apart. Finally, the sample is preserved in a fixative and applied to a slide.

The geneticist then stains chromosomes with one of several dyes to better visualize the distinct and reproducible banding patterns of each chromosome pair. Following staining, the chromosomes are viewed using bright-field microscopy. A common stain choice is the Giemsa stain. Giemsa staining results in approximately 400–800 bands (of tightly coiled DNA and condensed proteins) arranged along all of the 23 chromosome pairs; an experienced geneticist can identify each band. In addition to the banding patterns, chromosomes are further identified on the basis of size and centromere location. To obtain the classic depiction of the karyotype in which homologous pairs of chromosomes are aligned in numerical order from longest to shortest, the geneticist obtains a digital image, identifies each chromosome, and manually arranges the chromosomes into this pattern (Figure 1).

At its most basic, the karyogram may reveal genetic abnormalities in which an individual has too many or too few chromosomes per cell. Examples of this are Down Syndrome, which is identified by a third copy of chromosome 21, and Turner Syndrome, which is characterized by the presence of only one X chromosome in women instead of the normal two. Geneticists can also identify large deletions or insertions of DNA. For instance, Jacobsen Syndrome—which involves distinctive facial features as well as heart and bleeding defects—is identified by a deletion on chromosome 11. Finally, the karyotype can pinpoint translocations, which occur when a segment of genetic material breaks from one chromosome and reattaches to another chromosome. Translocations are implicated in certain cancers, including chronic myelogenous leukemia.

During Mendel's lifetime, inheritance was an abstract concept that could only be inferred by performing crosses and observing the traits expressed by offspring. By observing a karyogram, today's geneticists can actually visualize the chromosomal composition of an individual to confirm or predict genetic abnormalities in offspring, even before birth.

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COMMON DISORDERS

Learning Outcomes

- Identify common errors that can create an abnormal karyotype
- Identify syndromes that result from a significant change in chromosome number

Of all of the chromosomal disorders, abnormalities in chromosome number are the most obviously identifiable from a karyogram. Disorders of chromosome number include the duplication or loss of entire chromosomes, as well as changes in the number of complete sets of chromosomes. They are caused by **nondisjunction**, which occurs when pairs of homologous chromosomes or sister chromatids fail to separate during meiosis. Misaligned or incomplete synapsis, or a dysfunction of the spindle apparatus that facilitates chromosome migration, can cause nondisjunction. The risk of nondisjunction occurring increases with the age of the parents.

Nondisjunction can occur during either meiosis I or II, with differing results (Figure 1). If homologous chromosomes fail to separate during meiosis I, the result is two gametes that lack that particular chromosome and two gametes with two copies of the chromosome. If sister chromatids fail to separate during meiosis II, the result is one gamete that lacks that chromosome, two normal gametes with one copy of the chromosome, and one gamete with two copies of the chromosome.

Practice Question

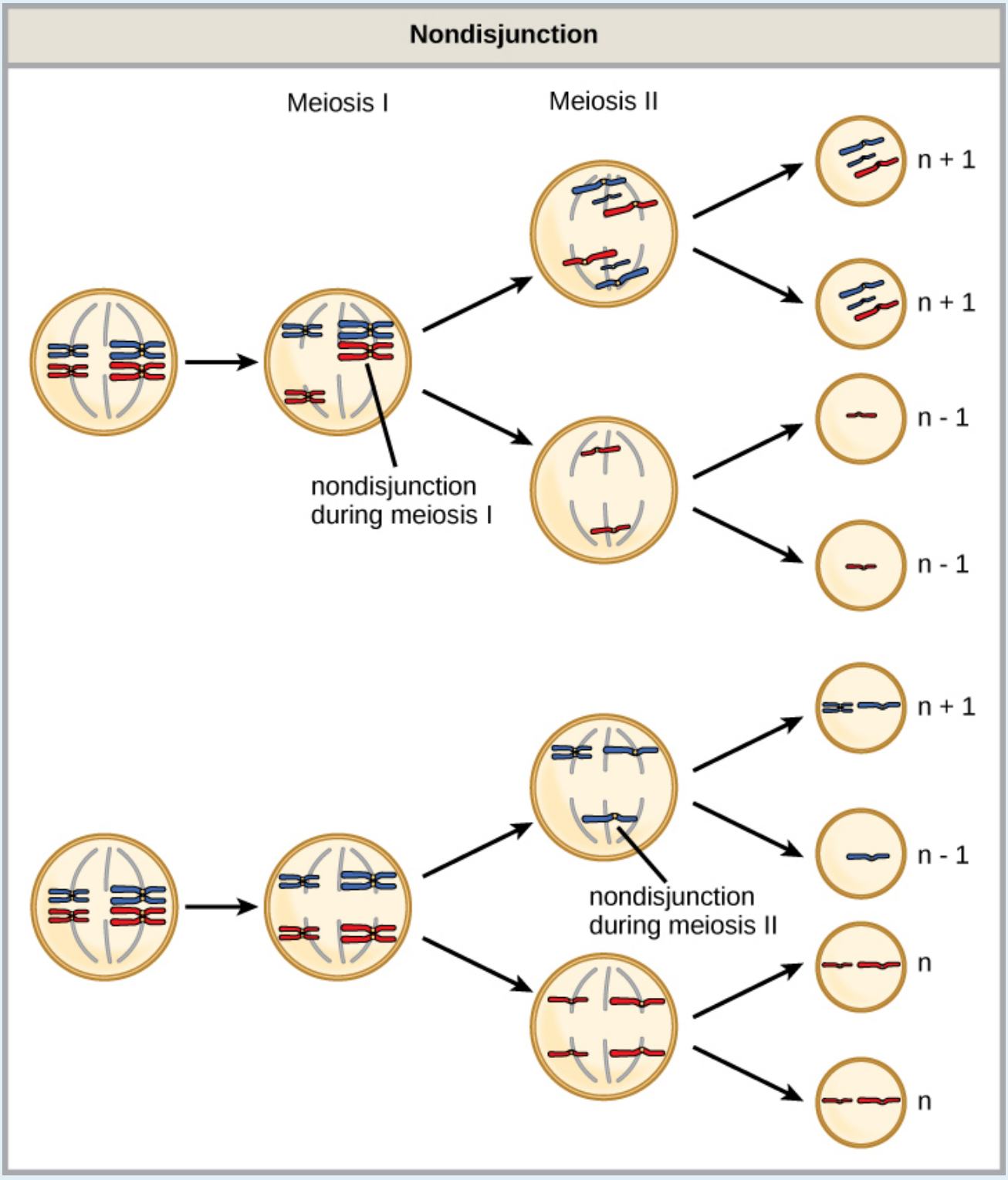

Figure 1. Nondisjunction occurs when homologous chromosomes or sister chromatids fail to separate during meiosis, resulting in an abnormal chromosome number. Nondisjunction may occur during meiosis I or meiosis II.

Which of the following statements about nondisjunction is true?

a. Nondisjunction only results in gametes with n+1 or n–1 chromosomes.
b. Nondisjunction occurring during meiosis II results in 50 percent normal gametes.
c. Nondisjunction during meiosis I results in 50 percent normal gametes.
d. Nondisjunction always results in four different kinds of gametes.

Answer

Answer b is true.

Aneuploidy

An individual with the appropriate number of chromosomes for their species is called **euploid**; in humans, euploidy corresponds to 22 pairs of autosomes and one pair of sex chromosomes. An individual with an error in chromosome number is described as **aneuploid**, a term that includes **monosomy** (loss of one chromosome) or **trisomy** (gain of an extraneous chromosome). Monosomic human zygotes missing any one copy of an autosome invariably fail to develop to birth because they lack essential genes. This underscores the importance of "gene dosage" in humans. Most autosomal trisomies also fail to develop to birth; however, duplications of some of the smaller chromosomes (13, 15, 18, 21, or 22) can result in offspring that survive for several weeks to many years. Trisomic individuals suffer from a different type of genetic imbalance: an excess in gene dose. Individuals with an extra chromosome may synthesize an abundance of the gene products encoded by that chromosome. This extra dose (150 percent) of specific genes can lead to a number of functional challenges and often precludes development. The most common trisomy among viable births is that of chromosome 21, which corresponds to Down Syndrome. Individuals with this inherited disorder are characterized by short stature and stunted digits, facial distinctions that include a broad skull and large tongue, and significant developmental delays. The incidence of Down syndrome is correlated with maternal age; older women are more likely to become pregnant with fetuses carrying the trisomy 21 genotype (Figure 2).

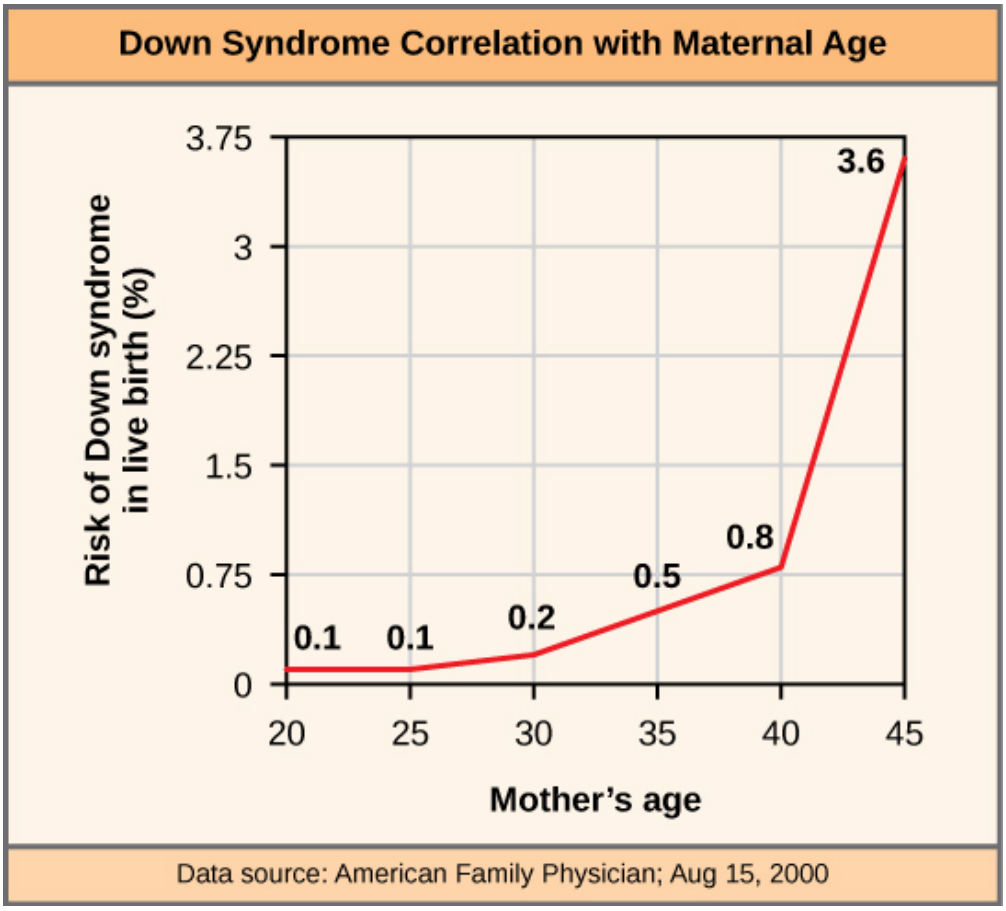

Figure 2. The incidence of having a fetus with trisomy 21 increases dramatically with maternal age.

Polyploidy

An individual with more than the correct number of chromosome sets (two for diploid species) is called **polyploid**. For instance, fertilization of an abnormal diploid egg with a normal haploid sperm would yield a triploid zygote. Polyploid animals are extremely rare, with only a few examples among the flatworms, crustaceans, amphibians, fish, and lizards. Polyploid animals are sterile because meiosis cannot proceed normally and instead produces mostly aneuploid daughter cells that cannot yield viable zygotes. Rarely, polyploid animals can reproduce asexually by haplodiploidy, in which an unfertilized egg divides mitotically to produce offspring. In contrast, polyploidy is very common in the plant kingdom, and polyploid plants tend to be larger and more robust than euploids of their species (Figure 3).

Figure 3. As with many polyploid plants, this triploid orange daylily (Hemerocallis fulva) is particularly large and robust, and grows flowers with triple the number of petals of its diploid counterparts. (credit: Steve Karg)

Sex Chromosome Nondisjunction in Humans

Humans display dramatic deleterious effects with autosomal trisomies and monosomies. Therefore, it may seem counterintuitive that human females and males can function normally, despite carrying different numbers of the X chromosome. Rather than a gain or loss of autosomes, variations in the number of sex chromosomes are associated with relatively mild effects. In part, this occurs because of a molecular process called X inactivation. Early in development, when female mammalian embryos consist of just a few thousand cells (relative to trillions in the newborn), one X chromosome in each cell inactivates by tightly condensing into a quiescent (dormant) structure called a Barr body. The chance that an X chromosome (maternally or paternally derived) is inactivated in each cell is random, but once the inactivation occurs, all cells derived from that one will have the same inactive X chromosome or Barr body. By this process, females compensate for their double genetic dose of X chromosome.

In so-called “tortoiseshell” cats, embryonic X inactivation is observed as color variegation (Figure 4). Females that are heterozygous for an X-linked coat color gene will express one of two different coat colors over different regions of their body, corresponding to whichever X chromosome is inactivated in the embryonic cell progenitor of that region.

Figure 4. In cats, the gene for coat color is located on the X chromosome. In the embryonic development of female cats, one of the two X chromosomes is randomly inactivated in each cell, resulting in a tortoiseshell pattern if the cat has two different alleles for coat color. Male cats, having only one X chromosome, never exhibit a tortoiseshell coat color. (credit: Michael Bodega)

An individual carrying an abnormal number of X chromosomes will inactivate all but one X chromosome in each of her cells. However, even inactivated X chromosomes continue to express a few genes, and X chromosomes must reactivate for the proper maturation of female ovaries. As a result, X-chromosomal abnormalities are typically associated with mild mental and physical defects, as well as sterility. If the X chromosome is absent altogether, the individual will not develop in utero.

Several errors in sex chromosome number have been characterized. Individuals with three X chromosomes, called triplo-X, are phenotypically female but express developmental delays and reduced fertility. The XXY genotype, corresponding to one type of Klinefelter syndrome, corresponds to phenotypically male individuals with small testes, enlarged breasts, and reduced body hair. More complex types of Klinefelter syndrome exist in which the individual has as many as five X chromosomes. In all types, every X chromosome except one undergoes inactivation to compensate for the excess genetic dosage. This can be seen as several Barr bodies in each cell nucleus. Turner syndrome, characterized as an X0 genotype (i.e., only a single sex chromosome), corresponds to a phenotypically female individual with short stature, webbed skin in the neck region, hearing and cardiac impairments, and sterility.

Duplications and Deletions

In addition to the loss or gain of an entire chromosome, a chromosomal segment may be duplicated or lost. Duplications and deletions often produce offspring that survive but exhibit physical and mental abnormalities. Duplicated chromosomal segments may fuse to existing chromosomes or may be free in the nucleus. Cri-du-chat (from the French for “cry of the cat”) is a syndrome associated with nervous system abnormalities and identifiable physical features that result from a deletion of most of 5p (the small arm of chromosome 5) (Figure 5). Infants with this genotype emit a characteristic high-pitched cry on which the disorder’s name is based.

Figure 5. This individual with cri-du-chat syndrome is shown at two, four, nine, and 12 years of age. (credit: Paola Cerruti Mainardi)

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PUTTING IT TOGETHER: CELL DIVISION

Mitosis and meiosis are both forms of division of the nucleus in eukaryotic cells. They share some similarities, but also exhibit distinct differences that lead to very different outcomes (Figure 1 and Table 1). Mitosis is a single nuclear division that results in two nuclei that are usually partitioned into two new cells. The nuclei resulting from a mitotic division are genetically identical to the original nucleus. They have the same number of sets of chromosomes, one set in the case of haploid cells and two sets in the case of diploid cells. In most plants and all animal species, it is typically diploid cells that undergo mitosis to form new diploid cells. In contrast, meiosis consists of two nuclear divisions resulting in four nuclei that are usually partitioned into four new cells. The nuclei resulting from meiosis are not genetically identical and they contain one chromosome set only. This is half the number of chromosome sets in the original cell, which is diploid.

The main differences between mitosis and meiosis occur in meiosis I, which is a very different nuclear division than mitosis. In meiosis I, the homologous chromosome pairs become associated with each other, are bound together with the synaptonemal complex, develop chiasmata and undergo crossover between sister chromatids, and line up along the metaphase plate in tetrads with kinetochore fibers from opposite spindle poles attached to each kinetochore of a homolog in a tetrad. All of these events occur only in meiosis I.

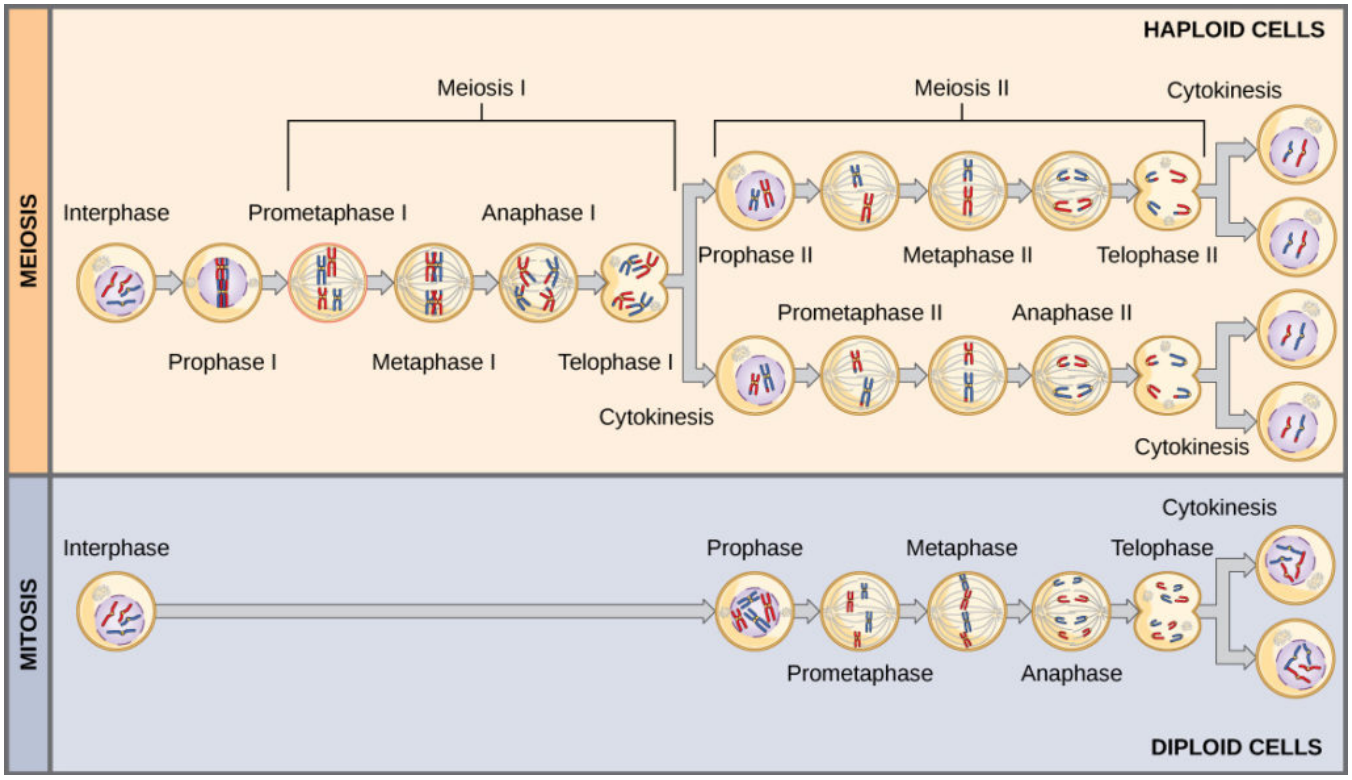

Figure 1. Meiosis and mitosis are both preceded by one round of DNA replication; however, meiosis includes two nuclear divisions. The four daughter cells resulting from meiosis are haploid and genetically distinct. The daughter cells resulting from mitosis are diploid and identical to the parent cell.

Table 1. Meiosis v. Mitosis

	Meiosis	Mitosis
DNA Synthesis	Occurs in S phase of Interphase	Occurs in S phase of Interphase
Synapsis of homolgous chromosomes	During prophase I	Does not occur in mitosis
Crossover	During prophase I	Does not occur in mitosis
Homologous chromosomes line up at metaphase plate	During metaphase I	Does not occur in mitosis
Sister chromatids line up at metaphase plate	During metaphase II	During metaphase
Outcome: Number and genetic composition of daughter cells	Four haploid cells at the end of meiosis II	Two diploid cells at the end of mitosis

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MODULE 8: DNA STRUCTURE AND REPLICATION

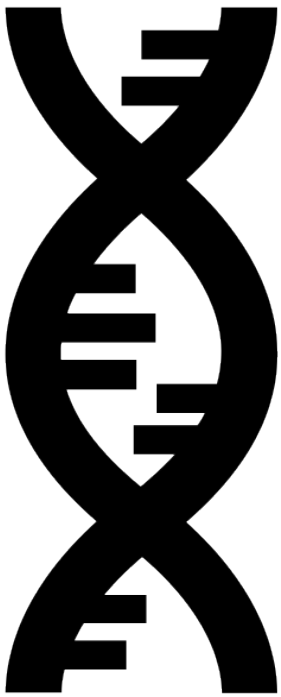
WHY IT MATTERS: DNA STRUCTURE AND REPLICATION

Why learn about DNA structure and the process of DNA replication?

You're one of a kind. It's not just your eyes, smile, and personality. Your health, risk for disease, and the ways you respond to medicines are also unique. Medicines that work well for some people may not help you at all. In fact, they might even cause problems. Wouldn't it be nice if treatments and preventive care could be designed just for you?

The careful matching of your biology to your medical care is known as personalized medicine. It's already being used by health care providers nationwide.

The story of personalized medicine begins with the unique DNA you inherited from your parents. DNA is responsible for all the physical traits that make you function as a human organism. It plays a vital role in life on this planet. DNA stores genetic information.



Genes are stretches of DNA that serve as a sort of instruction manual telling your body how to make the proteins and perform the other tasks that your body needs. The same genes often differ slightly between people. Bases may be switched, missing, or added here and there. Most of these variations have no effect on your health. But some can create unusual proteins that might boost your risk for certain diseases. Some variants can affect how well a medicine works in your body. Or they might cause a medicine to have different side effects in you than in someone else.

It's becoming more common for doctors to test for gene variants before prescribing certain drugs. "If doctors know your genes, they can predict drug response and incorporate this information into the medical decisions they make," says Dr. Rochelle Long, a pharmacogenomics expert at NIH. "By screening to know who shouldn't get certain drugs, we can prevent life-threatening side effects," Long says.

Even one of the oldest and most common drugs, aspirin, can have varying effects based on your genes. Millions of people take a daily aspirin to lower their risk for heart attack and stroke. Aspirin helps by preventing blood clots that could clog arteries. But aspirin doesn't reduce heart disease risk in everyone.

Personalized Medicine in Practice

Here are just a few treatments that benefit from personalization:

- Blood clot treatments
- Colorectal cancer treatments
- Breast cancer treatments

So what does this mean for you? Is personalized medicine something you should look into? Let's learn more about DNA and genetics to see if we can answer this question.

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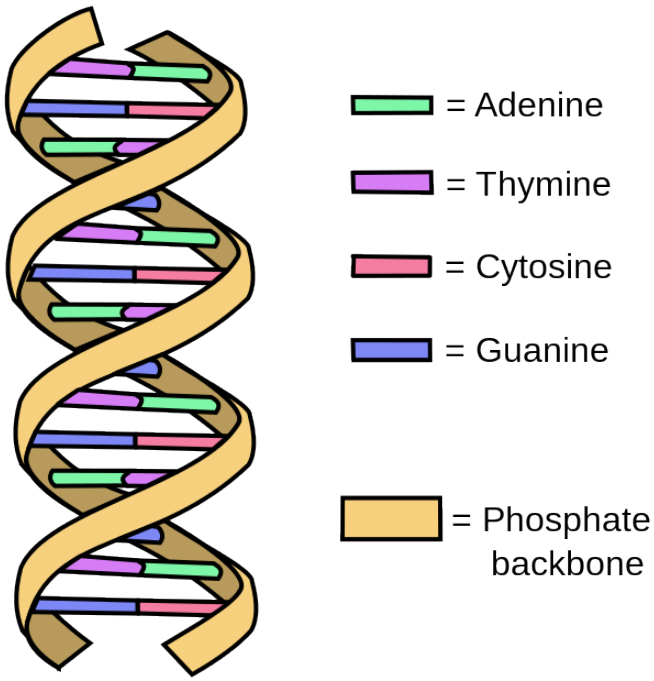
INTRODUCTION TO STORING GENETIC INFORMATION

What you'll learn to do: Explain how DNA stores genetic information

The unique structure of DNA is key to its ability to store and replicated genetic information:



DNA

In this outcome, you will learn to describe the double helix structure of DNA: its sugar-phosphate backbone ladder with nitrogenous base “rungs” of ladder.

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STRUCTURE OF DNA

Learning Outcomes

Diagram the structure of DNA

The building blocks of DNA are **nucleotides**. The important components of each nucleotide are a nitrogenous base, deoxyribose (5-carbon sugar), and a phosphate group (see Figure 1). Each nucleotide is named depending on its nitrogenous base. The nitrogenous base can be a **purine**, such as adenine (A) and guanine (G), or a **pyrimidine**, such as cytosine (C) and thymine (T). Uracil (U) is also a pyrimidine (as seen in Figure 1), but it only occurs in RNA, which we will talk more about later.

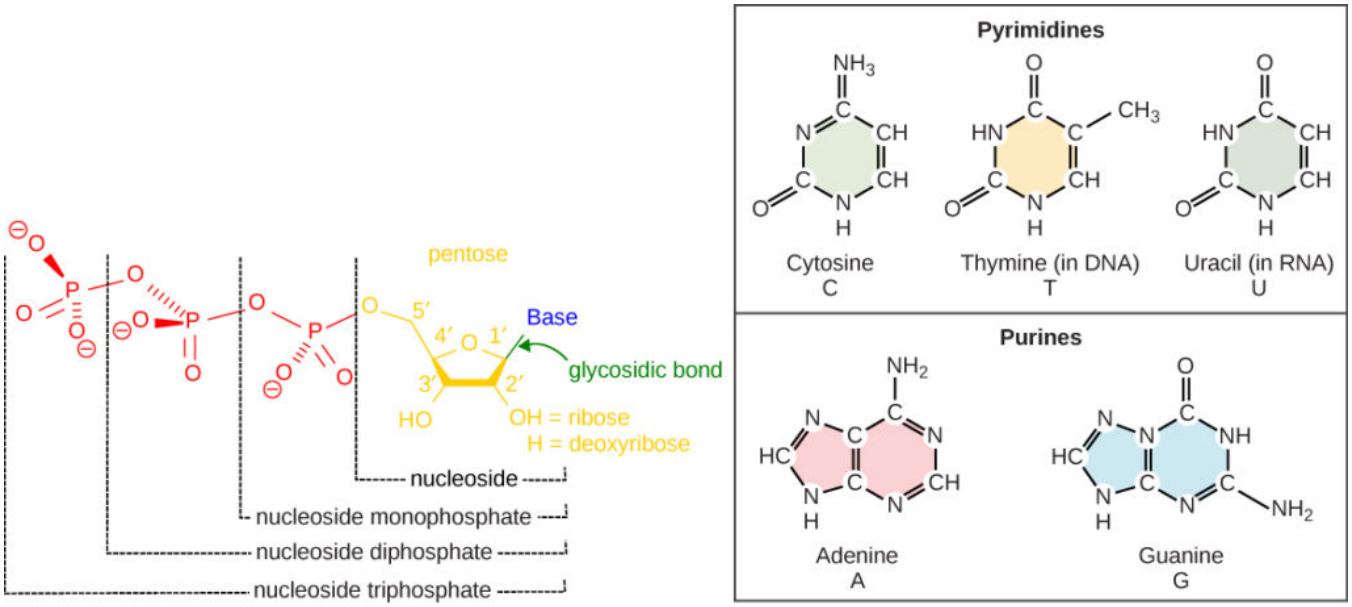

Figure 1. Each nucleotide is made up of a sugar, a phosphate group, and a nitrogenous base. The sugar is deoxyribose in DNA and ribose in RNA.

The nucleotides combine with each other by covalent bonds known as **phosphodiester bonds** or linkages. The phosphate residue is attached to the hydroxyl group of the 5′ carbon of one sugar of one nucleotide and the hydroxyl group of the 3′ carbon of the sugar of the next nucleotide, thereby forming a 5′-3′ phosphodiester bond.

In the 1950s, Francis **Crick** and James **Watson** worked together to determine the structure of DNA at the University of Cambridge, England. Other scientists like Linus Pauling and Maurice Wilkins were also actively exploring this field. Pauling had discovered the secondary structure of proteins using X-ray crystallography. In Wilkins' lab, researcher Rosalind Franklin was using X-ray diffraction methods to understand the structure of DNA. Watson and Crick were able to piece together the puzzle of the DNA molecule on the basis of Franklin's data because Crick had also studied X-ray diffraction (Figure 2). In 1962, James Watson, Francis Crick, and Maurice Wilkins were awarded the Nobel Prize in Medicine. Unfortunately, by then Franklin had died, and Nobel prizes are not awarded posthumously.

(a) (b)

Figure 2. The work of pioneering scientists (a) James Watson, Francis Crick, and Maclyn McCarty led to our present day understanding of DNA. Scientist Rosalind Franklin discovered (b) the X-ray diffraction pattern of DNA, which helped to elucidate its double helix structure. (credit a: modification of work by Marjorie McCarty, Public Library of Science)

Watson and Crick proposed that DNA is made up of *two strands* that are twisted around each other to form a right-handed *helix*. *Base pairing* takes place between a purine and pyrimidine; namely, *A pairs with T and G pairs with C*. Adenine and thymine are complementary base pairs, and cytosine and guanine are also complementary base pairs. The base pairs are stabilized by *hydrogen bonds*; adenine and thymine form two hydrogen bonds and cytosine and guanine form three hydrogen bonds. The two strands are *anti-parallel* in nature; that is, the 3′ end of one strand faces the 5′ end of the other strand. The sugar and phosphate of the nucleotides form the backbone of the structure, whereas the nitrogenous bases are stacked inside. Each base pair is separated from the other base pair by a distance of 0.34 nm, and each turn of the helix measures 3.4 nm. Therefore, ten base pairs are present per turn of the helix. The diameter of the DNA double helix is 2 nm, and it is uniform throughout. Only the pairing between a purine and pyrimidine can explain the uniform diameter. The twisting of the two strands around each other results in the formation of uniformly spaced major and minor grooves (Figure 3).

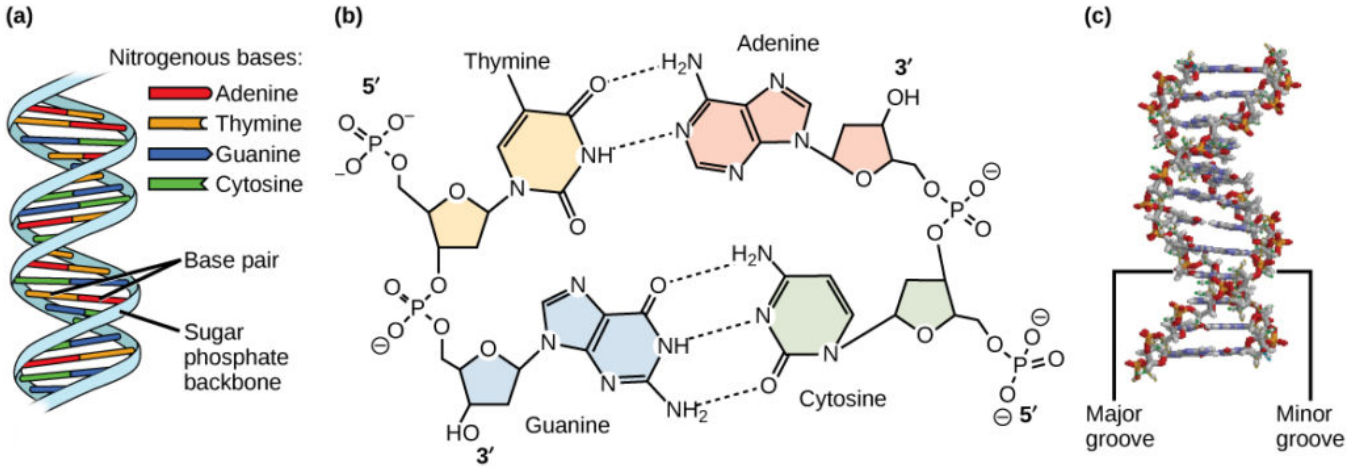

Figure 3. DNA has (a) a double helix structure and (b) phosphodiester bonds. The (c) major and minor grooves are binding sites for DNA binding proteins during processes such as transcription (the copying of RNA from DNA) and replication.

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GENETIC INFORMATION

Learning Outcomes

Relate the structure of DNA to the storage of genetic information

The genetic information of an organism is stored in DNA molecules. How can one kind of molecule contain all the instructions for making complicated living beings like ourselves? What component or feature of DNA can contain this information? It has to come from the nitrogen bases, because, as you already know, the backbone of all DNA molecules is the same. But there are only four bases found in DNA: G, A, C, and T. The sequence of these four bases can provide all the instructions needed to build any living organism. It might be hard to imagine that 4 different “letters” can communicate so much information. But think about the English language, which can represent a huge amount of information using just 26 letters. Even more profound is the binary code used to write

computer programs. This code contains only ones and zeros, and think of all the things your computer can do. The DNA alphabet can encode very complex instructions using just four letters, though the messages end up being really long. For example, the *E. coli* bacterium carries its genetic instructions in a DNA molecule that contains more than five million nucleotides. The human genome (all the DNA of an organism) consists of around three billion **nucleotides** divided up between 23 paired DNA molecules, or **chromosomes**.

The information stored in the order of bases is organized into **genes**: each gene contains information for making a functional product. The genetic information is first copied to another **nucleic acid polymer**, **RNA** (ribonucleic acid), preserving the order of the nucleotide bases. Genes that contain instructions for making proteins are converted to messenger RNA (mRNA). Some specialized genes contain instructions for making functional RNA molecules that don't make **proteins**. These RNA molecules function by affecting cellular processes directly; for example some of these RNA molecules regulate the expression of mRNA. Other genes produce RNA molecules that are required for **protein synthesis**, **transfer RNA** (**tRNA**), and **ribosomal RNA** (**rRNA**).

In order for DNA to function effectively at storing information, two key processes are required. First, information stored in the DNA molecule must be copied, with minimal errors, every time a cell divides. This ensures that both daughter cells inherit the complete set of genetic information from the parent cell. Second, the information stored in the DNA molecule must be **translated**, or expressed. In order for the stored information to be useful, cells must be able to access the instructions for making specific proteins, so the correct proteins are made in the right place at the right time.

Both copying and reading the information stored in DNA relies on base pairing between two **nucleic acid** polymer strands. Recall that DNA structure is a double helix (see Figure 1).

The sugar deoxyribose with the **phosphate group** forms the scaffold or backbone of the molecule (highlighted in yellow in Figure 1). Bases point inward. Complementary bases form hydrogen bonds with each other within the double helix. See how the bigger bases (**purines**) pair with the smaller ones (**pyrimidines**). This keeps the width of the double helix constant. More specifically, A pairs with T and C pairs with G. As we discuss the function of DNA in subsequent sections, keep in mind that there is a chemical reason for specific pairing of bases.

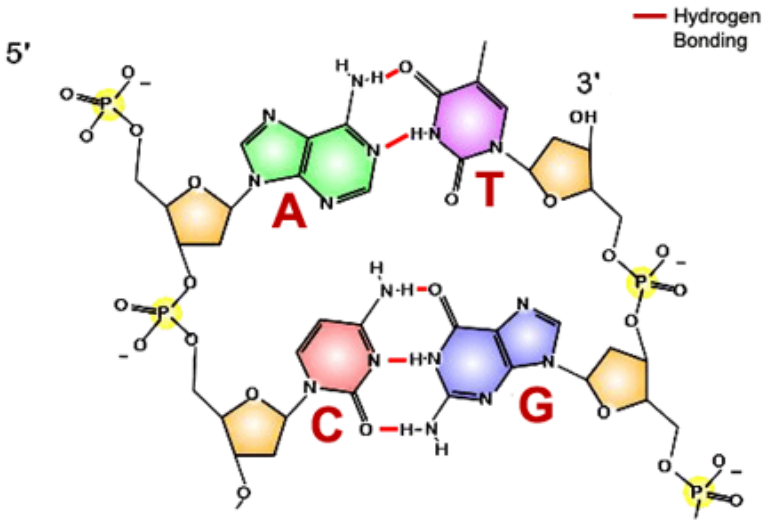

Figure 1. DNA's double helix. Graphic modified from "DNA chemical structure," by Madeleine Price Ball, CC-BY-SA-2.0

To illustrate the connection between information in DNA and an observable characteristic of an organism, let's consider a gene that provides the instructions for building the hormone insulin. Insulin is responsible for regulating blood sugar levels. The insulin gene contains instructions for assembling the protein insulin from individual amino acids. Changing the sequence of nucleotides in the DNA molecule can change the amino acids in the final protein, leading to protein malfunction. If insulin does not function correctly, it might be unable to bind to another protein (insulin receptor). On the organismal level of organization, this molecular event (change of DNA sequence) can lead to a disease state—in this case, diabetes.

Practice Question

The order of nucleotides in a gene (in DNA) is the key to how information is stored. For example, consider these two words: stable and tables. Both words are built from the same letters (subunits), but the different order of these subunits results in very different meanings. In DNA, the information is stored in units of 3 letters. Use the following key to decode the encrypted message. This should help you to see how information can be stored in the linear order of nucleotides in DNA.

ABC = a	DEF = d	GHI = e	JKL = f
MNO = h	PQR = i	STU = m	VWX = n
YZA = o	BCD = r	EFG = s	HIJ = t
KLM = w	NOP = j	QRS = p	TUV = y

Encrypted Message: HIJMNOPQREFG – PQREFG – MNOYZAKLM – DEFVWXABC – EFGHIJYZABCDGHIEFG – PQRVWXJKLYZABCDSTUABCHIJPQRYZAVWX

Answer

This is how DNA stores information.

Practice Question

Where in the DNA is information stored?

a. The shape of the DNA
b. The sugar-phosphate backbone
c. The sequence of bases
d. The presence of two strands.

Answer

Answer c. The sequence of the bases codes for the instructions for protein synthesis. The shape is DNA is not related to information storage. The sugar-phosphate backbone only acts as a scaffold. The presence of two strands is important for replication, but their information content is equivalent, as they are complementary to each other.

Practice Question

Which statement is correct?

a. The sequence of DNA bases is arranged into chromosomes, most of which contain the instructions to build an amino acid.
b. The sequence of DNA strands is arranged into chromosomes, most of which contain the instructions to build a protein.
c. The sequence of DNA bases is arranged into genes, most of which contain the instructions to build a protein.
d. The sequence of DNA phosphates is arranged into genes, most of which contain the instructions to build a cell.

Answer

Answer c. **The sequence of DNA bases is arranged into genes, most of which contain the instructions to build a protein.** DNA stores information in the sequence of its bases. The information is grouped into genes. Protein is what is mainly coded.

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INTRODUCTION TO DNA REPLICATION

What you'll learn to do: Explain the role of complementary base pairing in the precise replication process of DNA

In this outcome, we'll learn more about the precise structure of DNA and how it replicates. Watch this video for a quick introduction to this topic:

Watch this video online: https://youtu.be/TNKWgcFPHqw

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BASICS OF DNA REPLICATION

Learning Outcomes

Outline the basic steps in DNA replication

The elucidation of the structure of the double helix provided a hint as to how DNA divides and makes copies of itself. This model suggests that the two strands of the double helix separate during replication, and each strand serves as a template from which the new complementary strand is copied. What was not clear was how the replication took place. There were three models suggested: conservative, semi-conservative, and dispersive (see Figure 1).

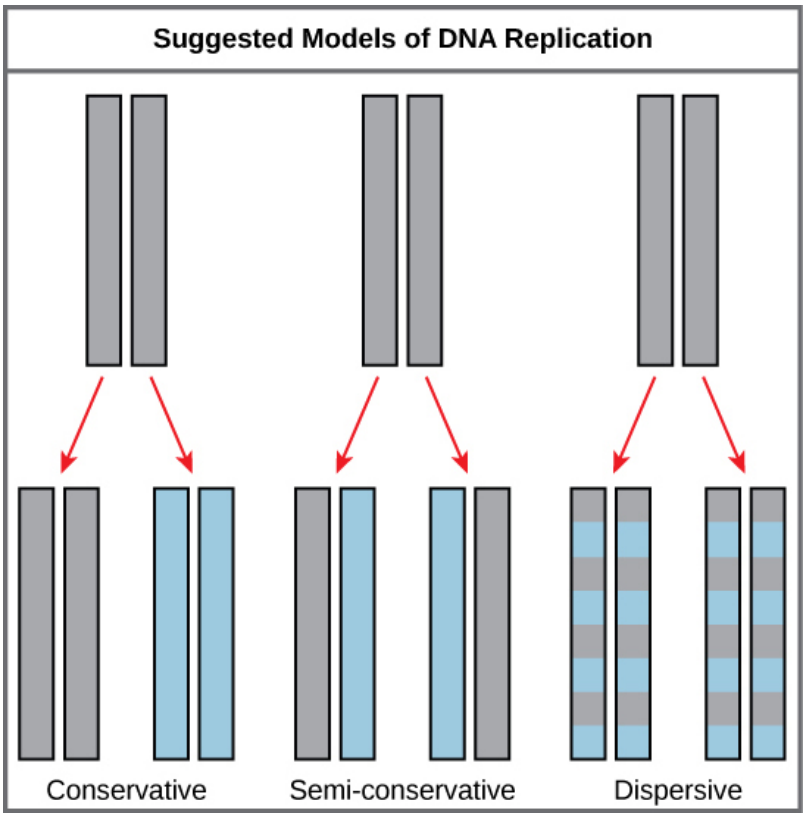

Figure 1. The three suggested models of DNA replication. Grey indicates the original DNA strands, and blue indicates newly synthesized DNA.

In **conservative replication**, the parental DNA remains together, and the newly formed daughter strands are together. The **semi-conservative** method suggests that each of the two parental DNA strands act as a template for new DNA to be synthesized; after replication, each double-stranded DNA includes one parental or "old" strand and one "new" strand. In the **dispersive model**, both copies of DNA have double-stranded segments of parental DNA and newly synthesized DNA interspersed.

Meselson and Stahl were interested in understanding how DNA replicates. They grew *E. coli* for several generations in a medium containing a "heavy" isotope of nitrogen (^{15}N) that gets incorporated into nitrogenous bases, and eventually into the DNA (Figure 2).

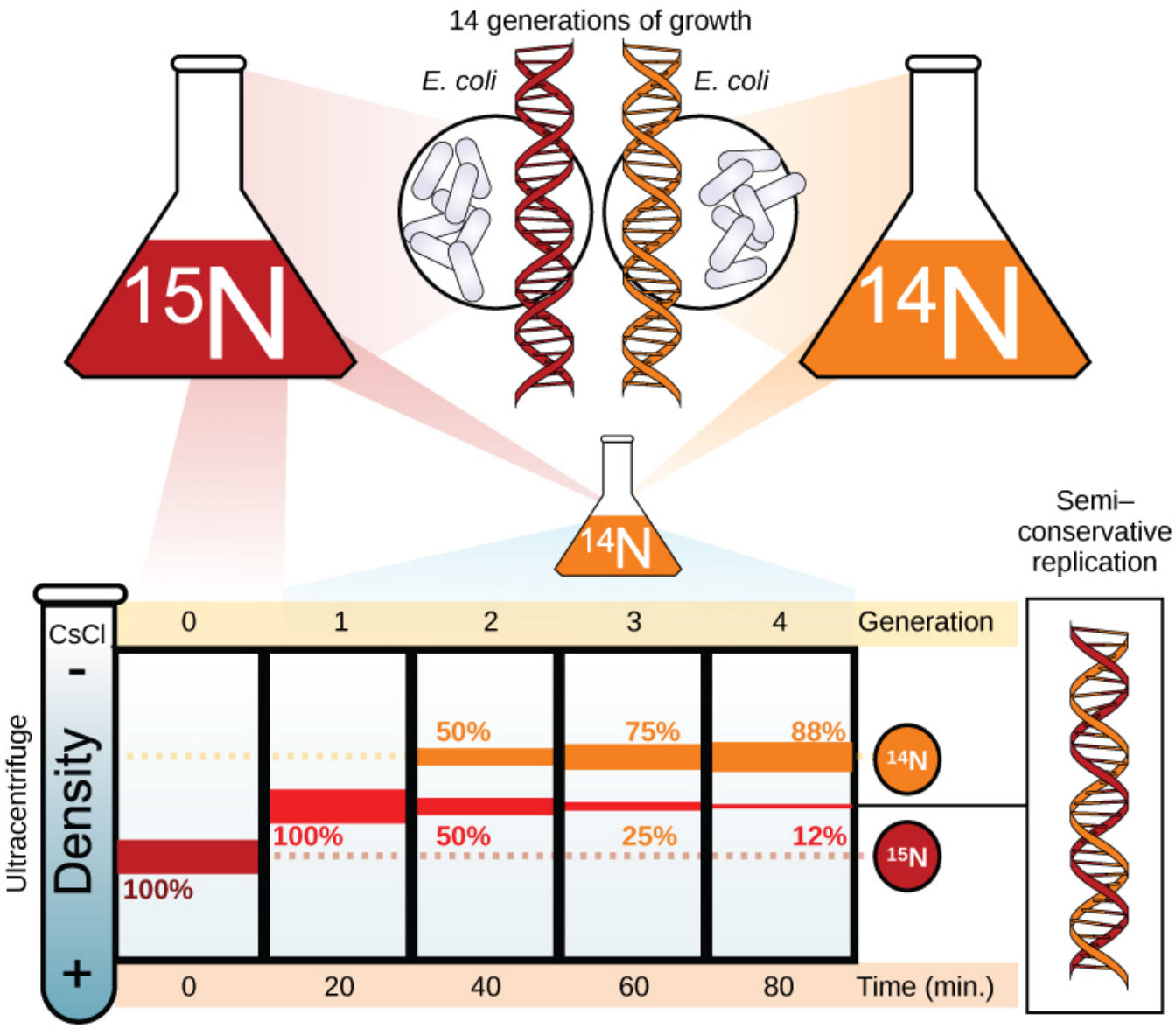

Figure 2. Meselson and Stahl experimented with E. coli grown first in heavy nitrogen (^{15}N) then in ^{14}N. DNA grown in ^{15}N (red band) is heavier than DNA grown in ^{14}N (orange band), and sediments to a lower level in cesium chloride solution in an ultracentrifuge. When DNA grown in ^{15}N is switched to media containing ^{14}N, after one round of cell division the DNA sediments halfway between the ^{15}N and ^{14}N levels, indicating that it now contains fifty percent ^{14}N. In subsequent cell divisions, an increasing amount of DNA contains ^{14}N only. This data supports the semi-conservative replication model. (credit: modification of work by Mariana Ruiz Villareal)

The *E. coli* culture was then shifted into medium containing ^{14}N and allowed to grow for one generation. The cells were harvested and the DNA was isolated. The DNA was centrifuged at high speeds in an ultracentrifuge. Some cells were allowed to grow for one more life cycle in ^{14}N and spun again. During the density gradient centrifugation, the DNA is loaded into a gradient (typically a salt such as cesium chloride or sucrose) and spun at high speeds of 50,000 to 60,000 rpm. Under these circumstances, the DNA will form a band according to its density in the gradient. DNA grown in ^{15}N will band at a higher density position than that grown in ^{14}N. Meselson and Stahl noted that after one generation of growth in ^{14}N after they had been shifted from ^{15}N, the single band observed was intermediate in position in between DNA of cells grown exclusively in ^{15}N and ^{14}N. This suggested either a semi-conservative or dispersive mode of replication. The DNA harvested from cells grown for two generations in ^{14}N formed two bands: one DNA band was at the intermediate position between ^{15}N and ^{14}N, and

the other corresponded to the band of ^{14}N DNA. These results could only be explained if DNA replicates in a semi-conservative manner. Therefore, the other two modes were ruled out.

During DNA replication, each of the two strands that make up the double helix serves as a template from which new strands are copied. The new strand will be complementary to the parental or “old” strand. When two daughter DNA copies are formed, they have the same sequence and are divided equally into the two daughter cells.

Click through this tutorial on DNA replication.

In Summary: Basics of DNA Replication

The model for DNA replication suggests that the two strands of the double helix separate during replication, and each strand serves as a template from which the new complementary strand is copied. In conservative replication, the parental DNA is conserved, and the daughter DNA is newly synthesized. The semi-conservative method suggests that each of the two parental DNA strands acts as template for new DNA to be synthesized; after replication, each double-stranded DNA includes one parental or “old” strand and one “new” strand. The dispersive mode suggested that the two copies of the DNA would have segments of parental DNA and newly synthesized DNA. Experimental evidence showed DNA replication is semi-conservative.

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MAJOR ENZYMES

Learning Outcomes

Identify the major enzymes that play a role in DNA replication

The process of **DNA replication** is catalyzed by a type of enzyme called **DNA polymerase** (*poly* meaning many, *mer* meaning pieces, and *–ase* meaning enzyme; so an enzyme that attaches many pieces of DNA). Observe Figure 1: the double helix of the original DNA molecule separates (blue) and new strands are made to match the separated strands. The result will be two DNA molecules, each containing an old and a new strand. Therefore, DNA replication is called semiconservative. The term *semiconservative* refers to the fact that half of the original molecule (one of the two strands in the double helix) is “conserved” in the new molecule. The original strand is referred to as the *template strand* because it provides the information, or template, for the newly synthesized strand.

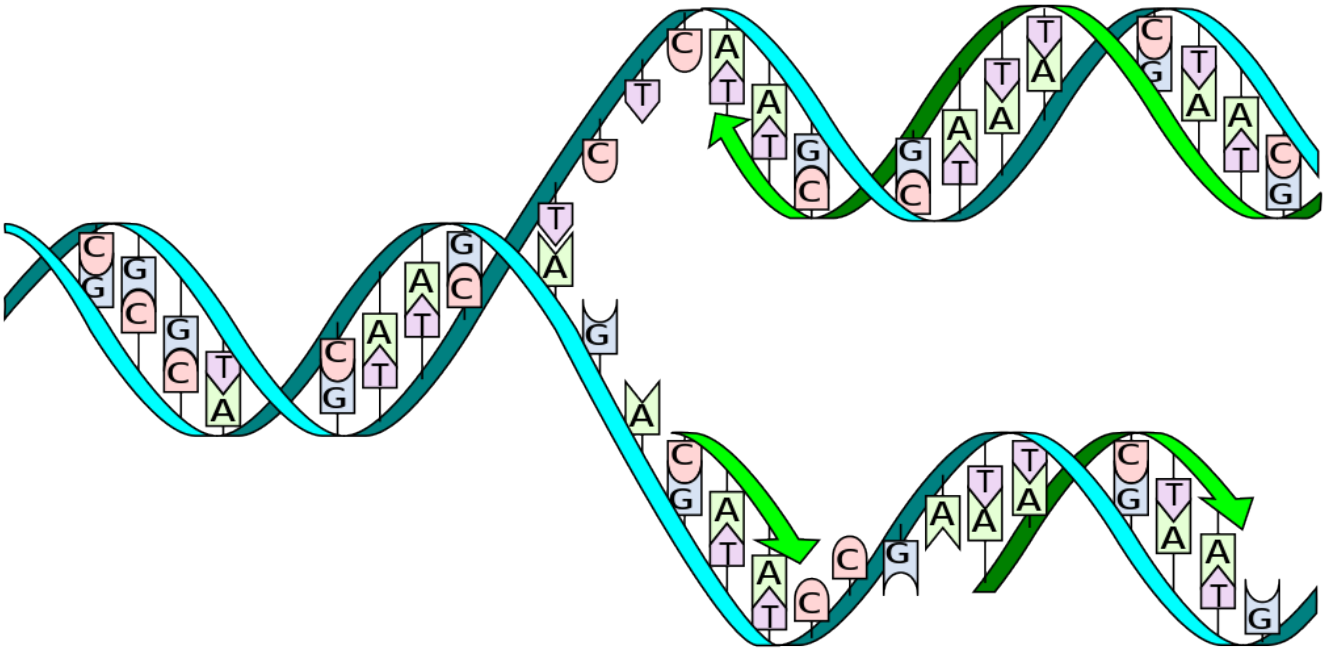

Figure 1. By Madprime(wikipedia) (DNA replication split horizontal) CC BY-SA 2.0

DNA polymerase needs an "anchor" to start adding nucleotides: a short sequence of DNA or RNA that is complementary to the template strand will work to provide a free 3′ end. This sequence is called a *primer* (Figure 2).

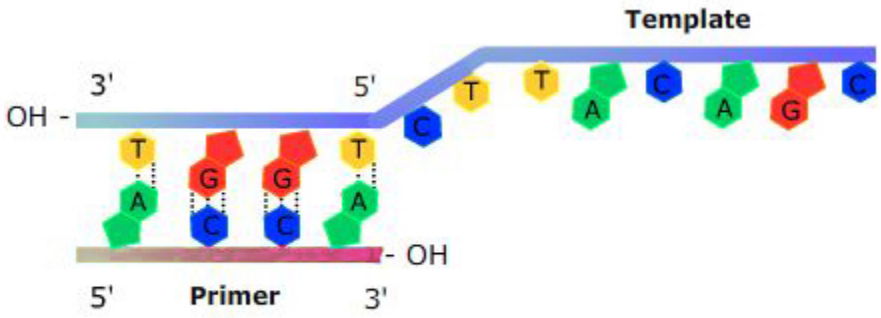

Figure 2. Primer and Template

How does **DNA polymerase** know in what order to add nucleotides? Specific base pairing in DNA is the key to copying the DNA: if you know the sequence of one strand, you can use base pairing rules to build the other strand. Bases form pairs (base pairs) in a very specific way.

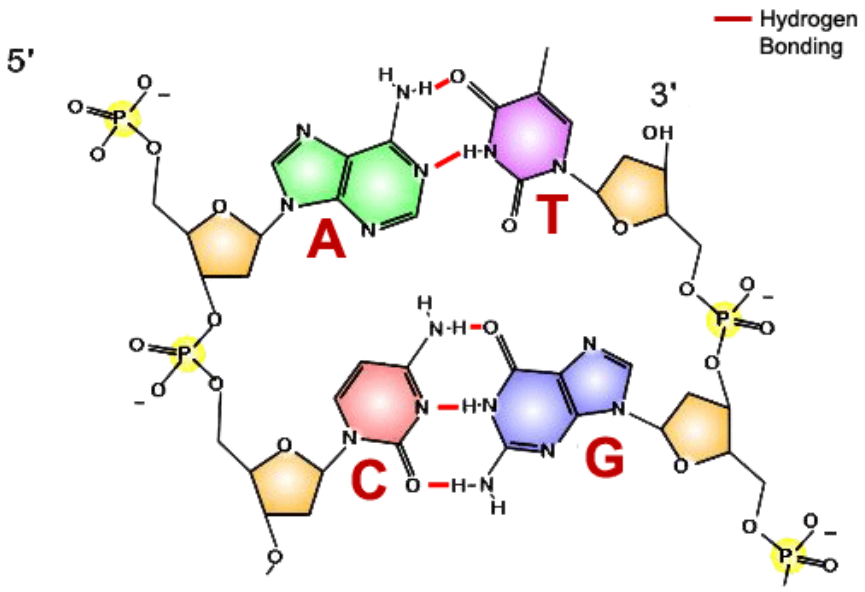

Figure 3. DNA chemical structure. Modification of DNA chemical structure by Madeleine Price Ball; CC-BY-SA-2.0

Practice Question

True/False: DNA replication requires an enzyme.

Answer

True. Most biological reactions rely on the enzyme to speed up the reaction. In the case of DNA replication, this enzyme is DNA polymerase.

Practice Question

What are the building blocks on DNA?

a. Deoxyribonucleotides
b. Fatty acids
c. Ribonucleotides
d. Amino acids

Answer

Answer a. DNA is a double helix made up of two long chains of deoxyribonucleotides.

Practice Question

True/False: DNA replication requires energy.

Answer

True. Making large molecules from small subunits (anabolism) requires energy. What supplies the energy? The building blocks themselves serve as a source of energy. As they get incorporated into the DNA polymer, two phosphate groups are broken off to release energy, some of which is used for making the polymer. Deoxyribonucleotides differ from nucleotides like ATP only by one missing oxygen atom.

Practice Question

We have the building blocks, a source of energy, and a catalyst. What's missing? We need instruction about the order of nucleotides in the new polymer. Which molecule provides these instructions?

a. Protein
b. DNA
c. Carbohydrate
d. Lipid

Answer

Answer b. We refer to this DNA as a template. The original information stored in the order of bases will direct the synthesis of the new DNA via base pairing.

Practice Question

There is one more thing required by the DNA polymerase. It cannot just start making a DNA copy of the template strand; it needs a short piece of DNA or RNA with a free hydroxyl group in the right place to attach the nucleotides to. (Remember that synthesis always occurs in one direction—new building blocks are added to the 3′ end.) This component starts the process by giving DNA polymerase something to bind to. What might you call this short piece of nucleic acid?

a. A solvent
b. A primer
c. A converter
d. A sealant

Answer

Answer b. A primer is used to start this process by giving DNA polymerase something to bind the new nucleotide to.

Now that you understand the basics of **DNA replication**, we can add a bit of complexity. The two strands of DNA have to be temporarily separated from each other; this job is done by a special enzyme, ***helicase***, that helps unwind and separate the DNA helices (Figure 4). Another issue is that the DNA polymerase only works in one direction along the strand (5′ to 3′), but the double-stranded DNA has two strands oriented in opposite directions. This problem is solved by synthesizing the two strands slightly differently: one new strand grows continuously, the other in bits and pieces. Short fragments of RNA are used as primers for the DNA polymerase.

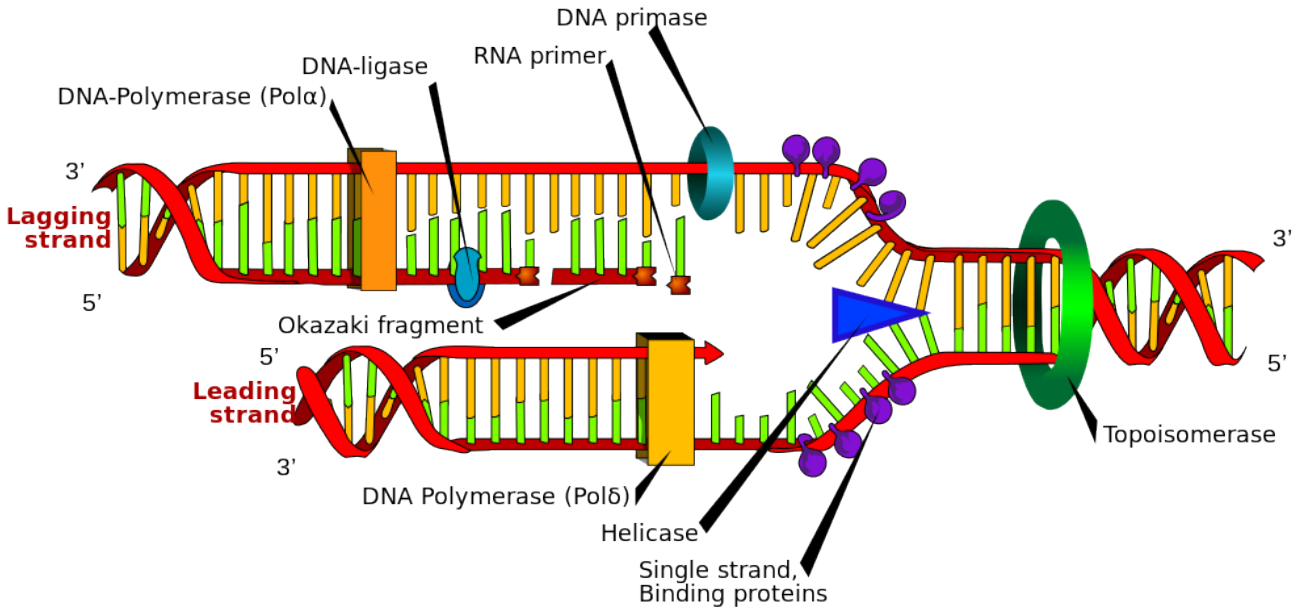

Figure 4. By Mariana Ruiz (DNA replication) Public Domain

Practice Question

Which of these separates the two complementary strands of DNA?

a. DNA polymerase
b. helicase
c. RNA primer
d. single-strand binding protein

Answer

Answer b. Helicase breaks the hydrogen bonds holding together the two strands of DNA.

Practice Question

Which of these attaches complementary bases to the template strand?

a. DNA polymerase
b. helicase
c. RNA primer
d. single-strand binding protein

Answer

Answer a. DNA polymerase builds the new strand of DNA.

Practice Question

Which of these is later replaced with DNA bases?

a. DNA polymerase

b. helicase
c. RNA primer
d. single-strand binding protein

Answers

Answer c. the RNA primer is replaced with DNA nucleotides.

In Summary: Major Enzymes

Replication in eukaryotes starts at multiple origins of replication. A primer is required to initiate synthesis, which is then extended by DNA polymerase as it adds nucleotides one by one to the growing chain. The leading strand is synthesized continuously, whereas the lagging strand is synthesized in short stretches called Okazaki fragments. The RNA primers are replaced with DNA nucleotides; the DNA remains one continuous strand by linking the DNA fragments with DNA ligase. Below is a summary table of the major enzymes addressed in this reading, listed in rough order of activity during replication.

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PROOFREADING DNA

Learning Outcomes

Identify the key proofreading processes in DNA replication

DNA replication is a highly accurate process, but mistakes can occasionally occur, such as a DNA polymerase inserting a wrong base. Uncorrected mistakes may sometimes lead to serious consequences, such as cancer. Repair mechanisms correct the mistakes. In rare cases, mistakes are not corrected, leading to mutations; in other cases, repair enzymes are themselves mutated or defective.

Most of the mistakes during DNA replication are promptly corrected by DNA polymerase by proofreading the base that has been just added (Figure 1). In proofreading, the DNA pol reads the newly added base before adding the next one, so a correction can be made. The polymerase checks whether the newly added base has paired correctly with the base in the template strand. If it is the right base, the next nucleotide is added. If an incorrect base has been added, the enzyme makes a cut at the phosphodiester bond and releases the wrong nucleotide. This is performed by the exonuclease action of DNA pol III. Once the incorrect nucleotide has been removed, a new one will be added again.

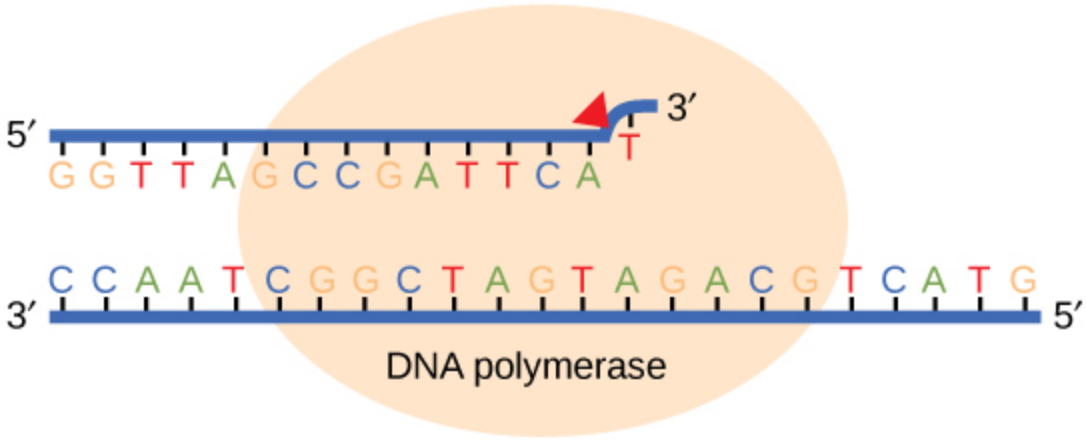

Figure 1. Proofreading by DNA polymerase corrects errors during replication.

Some errors are not corrected during replication, but are instead corrected after replication is completed; this type of repair is known as **mismatch repair** (Figure 2). The enzymes recognize the incorrectly added nucleotide and excise it; this is then replaced by the correct base. If this remains uncorrected, it may lead to more permanent damage. How do mismatch repair enzymes recognize which of the two bases is the incorrect one? In *E. coli*, after replication, the nitrogenous base adenine acquires a methyl group; the parental DNA strand will have methyl groups, whereas the newly synthesized strand lacks them. Thus, DNA polymerase is able to remove the wrongly incorporated bases from the newly synthesized, non-methylated strand. In eukaryotes, the mechanism is not very well understood, but it is believed to involve recognition of unsealed nicks in the new strand, as well as a short-term continuing association of some of the replication proteins with the new daughter strand after replication has completed.

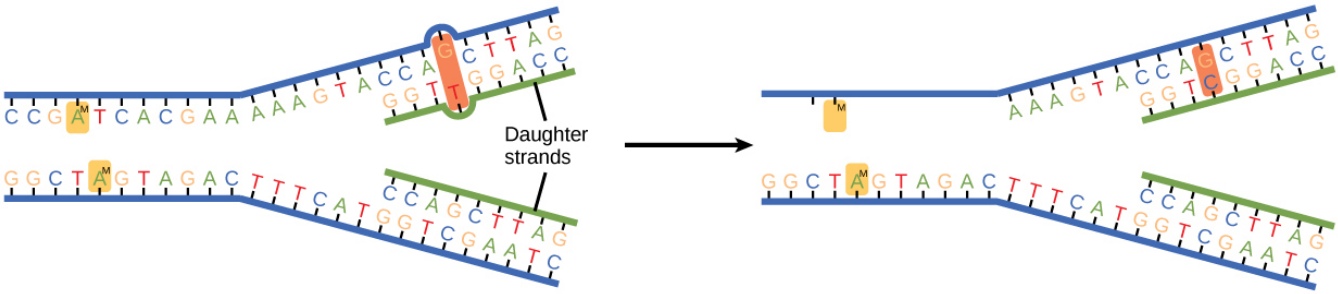

Figure 2. In mismatch repair, the incorrectly added base is detected after replication. The mismatch repair proteins detect this base and remove it from the newly synthesized strand by nuclease action. The gap is now filled with the correctly paired base.

In another type of repair mechanism, **nucleotide excision repair**, enzymes replace incorrect bases by making a cut on both the 3′ and 5′ ends of the incorrect base (Figure 3).

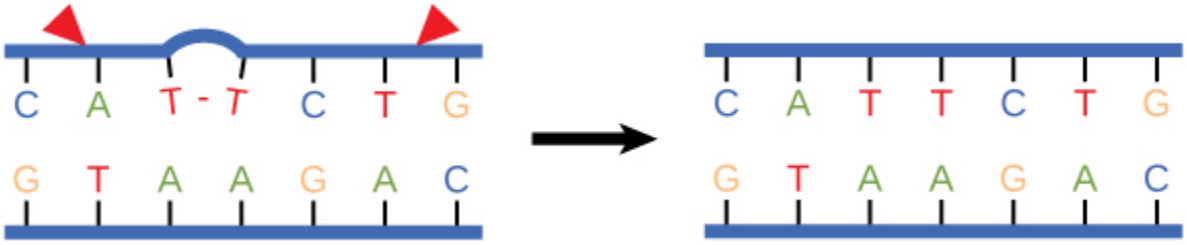

Figure 3. Nucleotide excision repairs thymine dimers. When exposed to UV, thymines lying adjacent to each other can form thymine dimers. In normal cells, they are excised and replaced.

The segment of DNA is removed and replaced with the correctly paired nucleotides by the action of DNA pol. Once the bases are filled in, the remaining gap is sealed with a phosphodiester linkage catalyzed by DNA ligase. This repair mechanism is often employed when UV exposure causes the formation of pyrimidine dimers.

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INTRODUCTION TO VIRUS REPLICATION

What you'll learn to do: Identify different viruses and how they replicate

While viruses technically aren't living things (they don't have cells), they still have DNA or RNA. Despite being "nonliving," viruses play an important role in evolutionary pressures on all living things, so it is important to study them.

Viruses are diverse entities. They vary in their structure, their replication methods, and in their target hosts. Nearly all forms of life—from bacteria and archaea to eukaryotes such as plants, animals, and fungi—have viruses that infect them. While most biological diversity can be understood through evolutionary history, such as how species have adapted to conditions and environments, much about virus origins and evolution remains unknown.

In this section, we'll learn how viruses reproduce. As we do, you can compare viral replication to DNA replication in living things. We will finish by looking at other nonliving infectious agents.

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VIRAL MORPHOLOGY

Learning Outcomes

Discuss the basics of virus structure

Viruses are **acellular**, meaning they are biological entities that do not have a cellular structure. They therefore lack most of the components of cells, such as organelles, ribosomes, and the plasma membrane. Viruses are sometimes called *virions*: a virion is a 'complete' virus free in the environment (not in a host). A virion consists of at least a **nucleic acid core** and an **outer protein coating or capsid**; sometimes a virus will have an outer **envelope** made of protein and phospholipid membranes derived from the host cell. Viruses may also contain additional proteins, such as enzymes. The most obvious difference between members of viral families is their morphology, which is quite diverse. An interesting feature of viral complexity is that the complexity of the host does not correlate with the complexity of the virion. Some of the most complex virion structures are observed in **bacteriophages**, viruses that infect the simplest living organisms, bacteria.

Types of Nucleic Acid

Unlike nearly all living organisms that use DNA as their genetic material, viruses may use either DNA or RNA as theirs. The **virus core** contains the genome or total genetic content of the virus. Viral genomes tend to be small, containing only those genes that encode proteins that the virus cannot get from the host cell. This genetic material may be single- or double-stranded. It may also be *linear or circula*r.

DNA viruses cause human diseases, such as chickenpox, hepatitis B, and some venereal diseases, like herpes and genital warts. Human diseases caused by RNA viruses include hepatitis C, measles, and rabies.

Morphology

Viruses come in many shapes and sizes, but these are consistent and distinct for each viral family. All virions have a nucleic acid genome covered by a protective layer of proteins, called a **capsid**. The capsid is made up of protein subunits called **capsomeres**. Some viral capsids are simple polyhedral “spheres,” whereas others are quite complex in structure.

Many viruses use some sort of glycoprotein to attach to their host cells via molecules on the cell called **viral receptors** (Figure 1).

Among the most complex virions known, the T4 bacteriophage, which infects the *Escherichia coli* bacterium, has a tail structure that the virus uses to attach to host cells and a head structure that houses its DNA.

Overall, the shape of the virion and the presence or absence of an envelope tell us little about what disease the virus may cause or what species it might infect, but they are still useful means to begin viral classification (Figure 2).

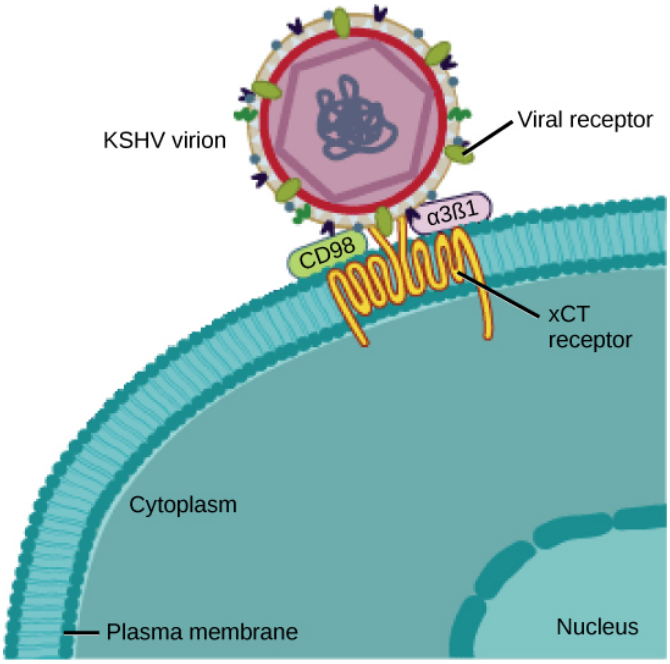

Figure 1. The KSHV virus binds the xCT receptor on the surface of human cells. (credit: modification of work by NIAID, NIH)

Practice Question

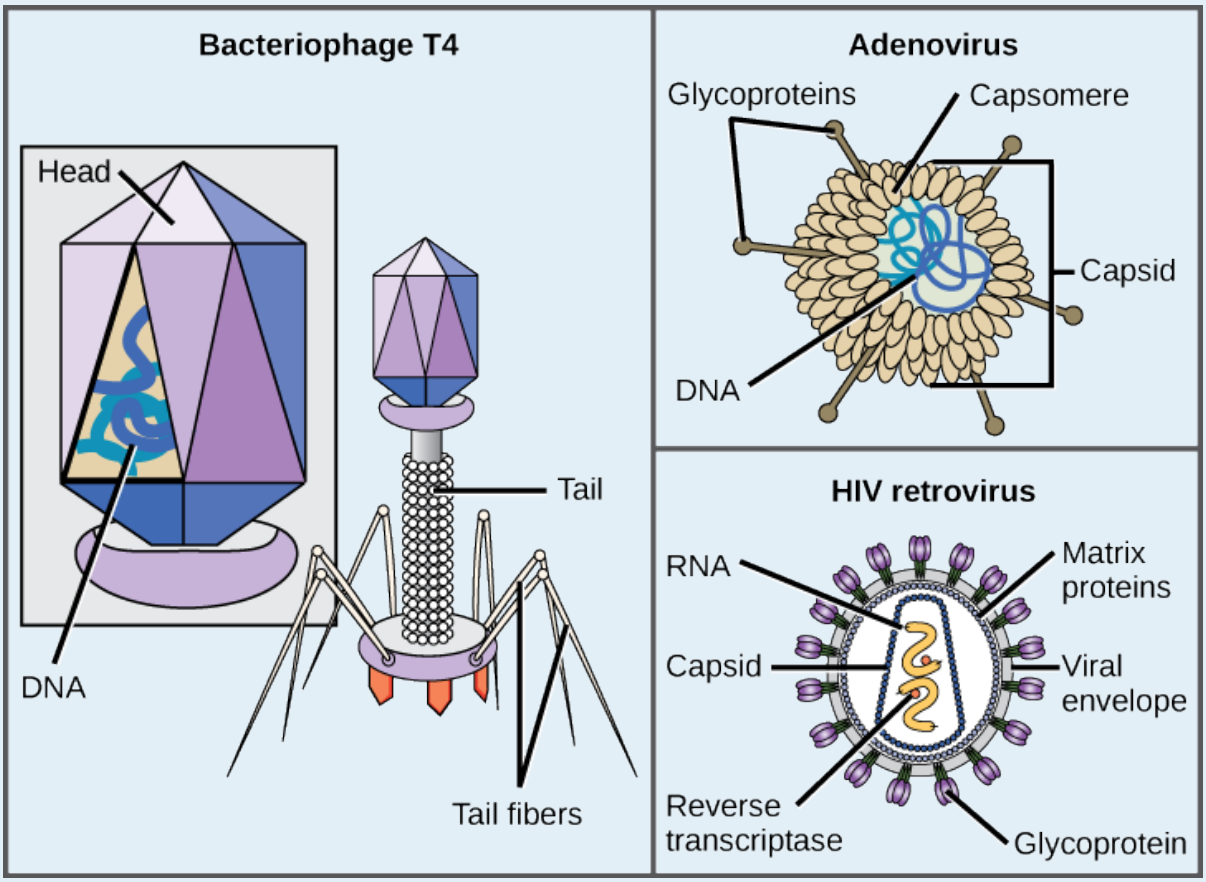

Figure 2. Viruses can be either complex in shape or relatively simple. This figure shows three relatively complex virions: the bacteriophage T4, with its DNA-containing head group and tail fibers that attach to host cells; adenovirus, which uses spikes from its capsid to bind to host cells; and HIV, which uses glycoproteins embedded in its envelope to bind to host cells. Notice that HIV has proteins called matrix proteins, internal to the envelope, which help stabilize virion shape. (credit "bacteriophage, adenovirus": modification of work by NCBI, NIH; credit "HIV retrovirus": modification of work by NIAID, NIH)

Which of the following statements about virus structure is true?

a. All viruses are encased in a viral membrane.
b. The capsomere is made up of small protein subunits called capsids.
c. DNA is the genetic material in all viruses.
d. Glycoproteins help the virus attach to the host cell.

Answer

Statement d is true.

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VIRAL INFECTIOUS CYCLES

Learning Outcomes

Understand the different types of viral infections, based on the host cell

As you've learned, viruses are often very specific as to which hosts and which cells within the host they will infect. This feature of a virus makes it specific to one or a few species of life on Earth. On the other hand, so many different types of viruses exist on Earth that nearly every living organism has its own set of viruses that tries to infect its cells. Even the smallest and simplest of cells, prokaryotic bacteria, may be attacked by specific types of viruses. Viruses that target bacteria are known as **bacteriophages**.

A bacteriophage has both **lytic** and **lysogenic** cycles. In the **lytic** cycle, the phage replicates and lyses the host cell. In the **lysogenic** cycle, phage DNA is incorporated into the host genome, where it is passed on to subsequent generations. When the phage DNA is incorporated into the host cell genome, it is called a **prophage**. Environmental stressors such as starvation or exposure to toxic chemicals may cause the prophage to excise and enter the lytic cycle.

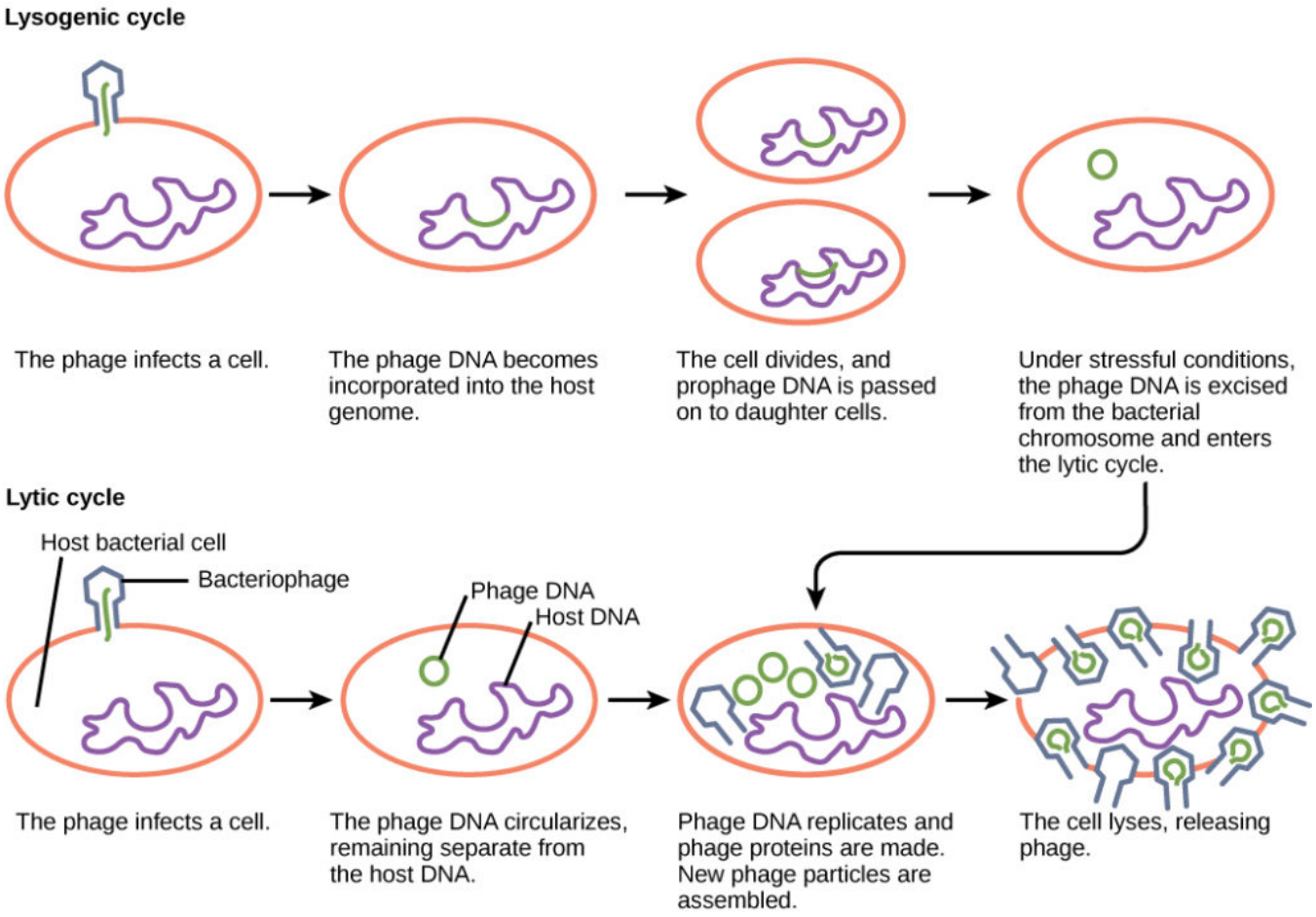

Figure 1. Click for a larger image

Practice Question

Which of the following statements is false?

a. In the lytic cycle, new phage are produced and released into the environment.
b. In the lysogenic cycle, phage DNA is incorporated into the host genome.
c. An environmental stressor can cause the phage to initiate the lysogenic cycle.
d. Cell lysis only occurs in the lytic cycle.

Answer

Statement c is false.

Animal Viruses

Animal viruses, unlike the viruses of plants and bacteria, do not have to penetrate a cell wall to gain access to the host cell. Non-enveloped or “naked” animal viruses may enter cells in two different ways. As a protein in the viral capsid binds to its receptor on the host cell, the virus may be taken inside the cell via a vesicle during the normal cell process of receptor-mediated endocytosis. An alternative method of cell penetration used by non-enveloped viruses is for capsid proteins to undergo shape changes after binding to the receptor, creating channels in the host cell membrane. The viral genome is then “injected” into the host cell through these channels in a manner analogous to that used by many bacteriophages. Enveloped viruses also have two ways of entering cells after binding to their receptors: receptor-mediated endocytosis, or **fusion**. Many enveloped viruses enter the cell by receptor-mediated endocytosis in a fashion similar to some non-enveloped viruses. On the other hand, fusion only occurs with enveloped virions. These viruses, which include HIV among others, use special fusion proteins in their envelopes to cause the envelope to fuse with the plasma membrane of the cell, thus releasing the genome and capsid of the virus into the cell cytoplasm.

After making their proteins and copying their genomes, animal viruses complete the assembly of new virions and exit the cell. As we have already discussed using the example of HIV, enveloped animal viruses may bud from the cell membrane as they assemble themselves, taking a piece of the cell’s plasma membrane in the process. On the other hand, non-enveloped viral progeny, such as rhinoviruses, accumulate in infected cells until there is a signal for lysis or apoptosis, and all virions are released together.

Animal viruses are associated with a variety of human diseases. Some of them follow the classic pattern of **acute disease**, where symptoms get increasingly worse for a short period followed by the elimination of the virus from the body by the immune system and eventual recovery from the infection. Examples of acute viral diseases are the common cold and influenza. Other viruses cause long-term **chronic infections**, such as the virus causing hepatitis C, whereas others, like herpes simplex virus, only cause **intermittent** symptoms. Still other viruses, such as human herpesviruses 6 and 7, which in some cases can cause the minor childhood disease roseola, often successfully cause productive infections without causing any symptoms at all in the host, and thus we say these patients have an **asymptomatic infection**.

In hepatitis C infections, the virus grows and reproduces in liver cells, causing low levels of liver damage. The damage is so low that infected individuals are often unaware that they are infected, and many infections are detected only by routine blood work on patients with risk factors such as intravenous drug use. On the other hand, since many of the symptoms of viral diseases are caused by immune responses, a lack of symptoms is an indication of a weak immune response to the virus. This allows for the virus to escape elimination by the immune system and persist in individuals for years, all the while producing low levels of progeny virions in what is known as a chronic viral disease. Chronic infection of the liver by this virus leads to a much greater chance of developing liver cancer, sometimes as much as 30 years after the initial infection.

As already discussed, herpes simplex virus can remain in a state of latency in nervous tissue for months, even years. As the virus “hides” in the tissue and makes few if any viral proteins, there is nothing for the immune response to act against, and immunity to the virus slowly declines. Under certain conditions, including various types of physical and psychological stress, the latent herpes simplex virus may be reactivated and undergo a lytic

replication cycle in the skin, causing the lesions associated with the disease. Once virions are produced in the skin and viral proteins are synthesized, the immune response is again stimulated and resolves the skin lesions in a few days by destroying viruses in the skin. As a result of this type of replicative cycle, appearances of cold sores and genital herpes outbreaks only occur intermittently, even though the viruses remain in the nervous tissue for life. Latent infections are common with other herpesviruses as well, including the varicella-zoster virus that causes chickenpox. After having a chickenpox infection in childhood, the varicella-zoster virus can remain latent for many years and reactivate in adults to cause the painful condition known as “shingles” (Figure 2).

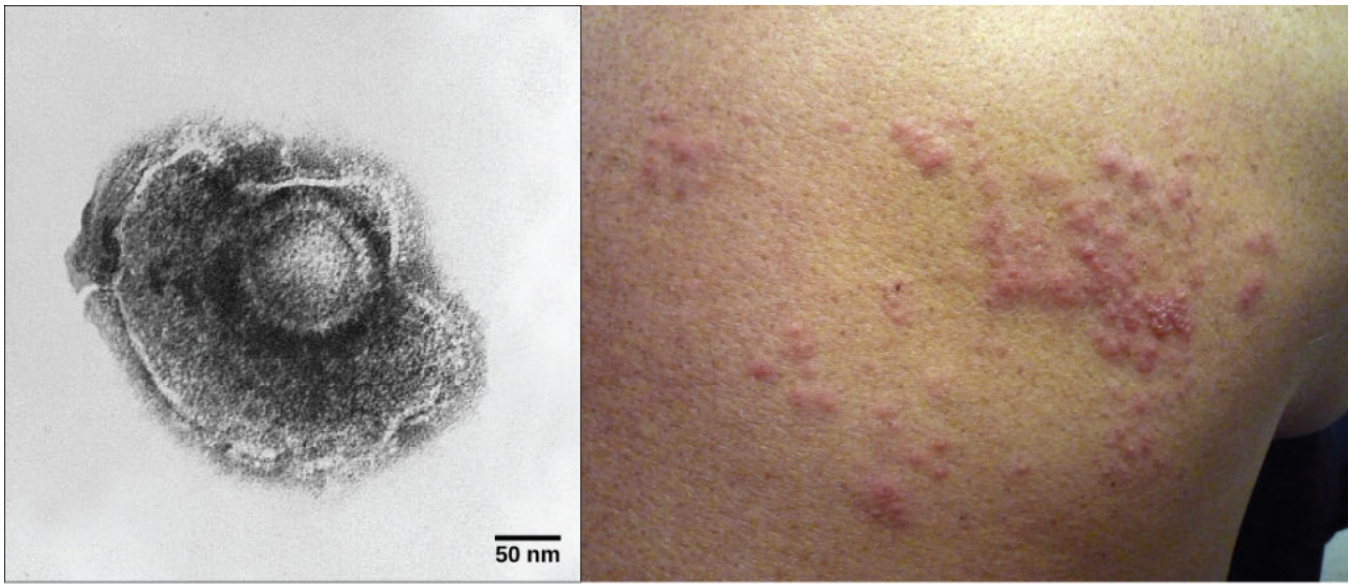

Figure 2. (a) Varicella-zoster, the virus that causes chickenpox, has an enveloped icosahedral capsid visible in this transmission electron micrograph. Its double-stranded DNA genome becomes incorporated in the host DNA and can reactivate after latency in the form of (b) shingles, often exhibiting a rash. (credit a: modification of work by Dr. Erskine Palmer, B. G. Martin, CDC; credit b: modification of work by “rosmary”/Flickr; scale-bar data from Matt Russell)

Some animal-infecting viruses, including the hepatitis C virus discussed above, are known as oncogenic viruses: They have the ability to cause cancer. These viruses interfere with the normal regulation of the host cell cycle either by either introducing genes that stimulate unregulated cell growth (oncogenes) or by interfering with the expression of genes that inhibit cell growth. Oncogenic viruses can be either DNA or RNA viruses.

Cancers known to be associated with viral infections include cervical cancer caused by human papillomavirus (HPV) , liver cancer caused by hepatitis B virus, T-cell leukemia, and several types of lymphoma.

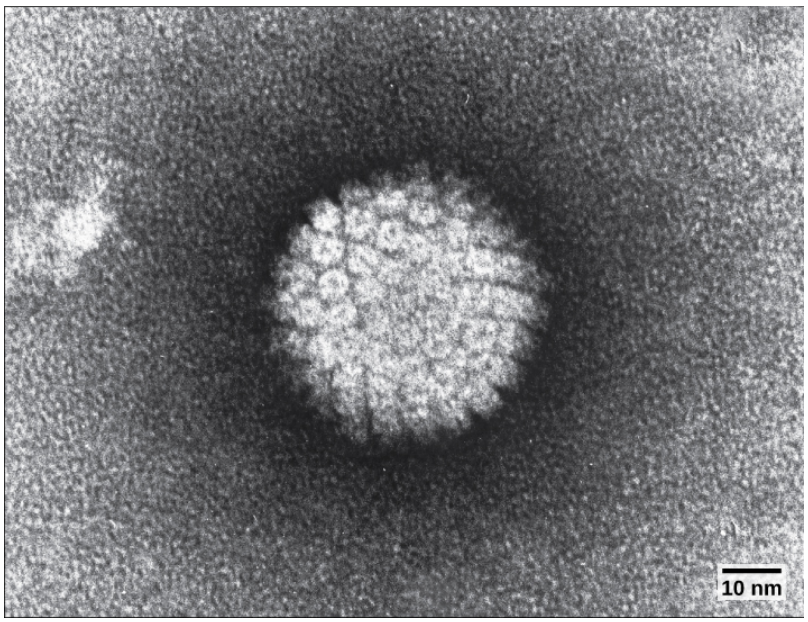

Figure 3. HPV, or human papillomavirus (credit: modification of work by NCI, NIH; scale-bar data from Matt Russell)

HPV, or human papillomavirus (as seen in Figure 3), has a naked icosahedral capsid visible in this transmission electron micrograph and a double-stranded DNA genome that is incorporated into the host DNA. The virus, which is sexually transmitted, is oncogenic and can lead to cervical cancer.

Visit the interactive animations showing the various stages of the replicative cycles of animal viruses and click on the flash animation links.

Plant Viruses

Plant viruses, like other viruses, contain a core of either DNA or RNA. You have already learned about one of these, the tobacco mosaic virus. As plants have a cell wall to protect their cells, these viruses do not use receptor-mediated endocytosis to enter host cells as is seen with animal viruses. For many plant viruses to be transferred from plant to plant, damage to some of the plants' cells must occur to allow the virus to enter a new host. This damage is often caused by weather, insects, animals, fire, or human activities like farming or landscaping. Additionally, plant offspring may inherit viral diseases from parent plants. Plant viruses can be transmitted by a variety of vectors, through contact with an infected plant's sap, by living organisms such as insects and nematodes, and through pollen. When plants viruses are transferred between different plants, this is known as **horizontal transmission**, and when they are inherited from a parent, this is called **vertical transmission**.

Symptoms of viral diseases vary according to the virus and its host (see the table below). One common symptom is **hyperplasia**, the abnormal proliferation of cells that causes the appearance of plant tumors known as **galls**. Other viruses induce **hypoplasia**, or decreased cell growth, in the leaves of plants, causing thin, yellow areas to appear. Still other viruses affect the plant by directly killing plant cells, a process known as **cell necrosis**. Other symptoms of plant viruses include malformed leaves, black streaks on the stems of the plants, altered growth of stems, leaves, or fruits, and ring spots, which are circular or linear areas of discoloration found in a leaf.

Table 1. Some Common Symptoms of Plant Viral Diseases

Symptom	Appears as
Hyperplasia	Galls (tumors)
Hypoplasia	Thinned, yellow splotches on leaves
Cell necrosis	Dead, blackened stems, leaves, or fruit
Abnormal growth patterns	Malformed stems, leaves, or fruit
Discoloration	Yellow, red, or black lines, or rings in stems, leaves, or fruit

Plant viruses can seriously disrupt crop growth and development, significantly affecting our food supply. They are responsible for poor crop quality and quantity globally, and can bring about huge economic losses annually. Others viruses may damage plants used in landscaping. Some viruses that infect agricultural food plants include the name of the plant they infect, such as tomato spotted wilt virus, bean common mosaic virus, and cucumber mosaic virus. In plants used for landscaping, two of the most common viruses are peony ring spot and rose mosaic virus. There are far too many plant viruses to discuss each in detail, but symptoms of bean common mosaic virus result in lowered bean production and stunted, unproductive plants. In the ornamental rose, the rose mosaic disease causes wavy yellow lines and colored splotches on the leaves of the plant.

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PRIONS AND VIROIDS

Learning Outcomes

- Describe prions and their basic properties
- Define viroids and their targets of infection

Prions and viroids are pathogens (agents with the ability to cause disease) that have simpler structures than viruses but, in the case of prions, still can produce deadly diseases.

Prions

Prions, so-called because they are proteinaceous, are infectious particles—smaller than viruses—that contain no nucleic acids (neither DNA nor RNA).

Fatal neurodegenerative diseases, such as **kuru** in humans and bovine spongiform encephalopathy (BSE) in cattle (commonly known as "**mad cow disease**"), were shown to be transmitted by prions. The disease was spread by the consumption of meat, nervous tissue, or internal organs from infected individuals, usually by members of the same species. Individuals with kuru and BSE show symptoms of loss of motor control and unusual behaviors, such as uncontrolled bursts of laughter with kuru, followed by death. These symptoms are due to lesions in the brain tissue.

The cause of spongiform encephalopathies, such as kuru and BSE, is an infectious structural variant of a normal cellular protein called PrP (prion protein). It is this variant that constitutes the prion particle. PrP exists in two forms, **PrP^{C}**, the normal form of the protein, and **PrP^{SC}**, the infectious form. Once introduced into the body, the PrP^{SC} contained within the prion binds to PrP^{C} and converts it to PrP^{SC}. This leads to an exponential increase of the PrP^{SC} protein, which aggregates. PrP^{SC} is folded abnormally, and the resulting conformation (shape) is directly responsible for the lesions seen in the brains of infected cattle. Thus, although not fully accepted among scientists, the prion seems likely to be an entirely new form of infectious agent, the first one found whose transmission is not reliant upon genes made of DNA or RNA.

Viroids

Viroids are plant pathogens: small, single-stranded, circular RNA particles that are much simpler than a virus. They do not have a capsid or outer envelope, but like viruses can reproduce only within a host cell. Viroids do not, however, manufacture any proteins, and they only produce a single, specific RNA molecule. Human diseases caused by viroids have yet to be identified.

Viroids are known to infect plants and are responsible for crop failures and the loss of millions of dollars in agricultural revenue each year. Some of the plants they infect include potatoes, cucumbers, tomatoes, chrysanthemums, avocados, and coconut palms. For example, the potato spindle tuber viroid (PSTVd), which typically spreads when

infected knives cut healthy potatoes in preparation for planting, can affect potatoes and tomatoes. The symptoms of PSTVd can be seen in Figure 1.

Figure 1. These potatoes have been infected by the potato spindle tuber viroid. (credit: Pamela Roberts, University of Florida Institute of Food and Agricultural Sciences, USDA ARS)

In Summary: Prions and Viroids

Prions are infectious agents that consist of protein, but no DNA or RNA, and seem to produce their deadly effects by duplicating their shapes and accumulating in tissues. They are thought to contribute to several progressive brain disorders, including mad cow disease and Creutzfeldt-Jakob disease. Viroids are single-stranded RNA pathogens that infect plants. Their presence can have a severe impact on the agriculture industry.

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PUTTING IT TOGETHER: DNA STRUCTURE AND REPLICATION

Let's expand just a bit on how the process of personalized medicine works. Within any given genome there will be small differences in the DNA sequence. All humans, for example, are 99.99% identical and only differ by 0.01%. These small differences account for many features in an individual, including how he or she responds to disease treatments.

Personalized Medicine in Practice

Let's return to our examples from the beginning of the chapter:

- **Blood clot treatments**: Screening the genome of a patient prior to prescribing Warafin for blood clots allows clinicians to determine if the drug will even work and if so, to pinpoint a specific dosage.
- **Colorectal cancer treatments**: In the case of colorectal cancer, the KRAS protein can be examined to determine if cetuximab will be effective. This is an important step as this drug is ineffective in about 40% of patients.
- **Breast cancer treatments**: Anti-breast cancer treatments can be assessed prior to trial-and-error in a patient based on the DNA sequence of key receptors in the body.

So, should you have your genes tested? Unfortunately, the decision about whether to get a particular genetic test can be complicated.

Getting a genetic test usually isn't difficult. Doctors generally take a sample of body fluid or tissue, such as blood, saliva or skin, and send it to a lab. Most genetic tests used today analyze just one or a few genes, often to help diagnose disease. Newborns, for example, are routinely screened for several genetic disorders by taking a few drops of blood from their heels. When life-threatening conditions are caught early, infants can be treated right away to prevent problems.

Genetic tests are now available for about 2,500 diseases, and that number keeps growing. Your doctor might advise you to get tested for specific genetic diseases if they tend to run in your family or if you have certain symptoms.

"While there are many genetic tests, they vary as to how well they predict risk," says Dr. Lawrence Brody, a genetic testing expert at NIH.

For some diseases, such as sickle cell anemia or cystic fibrosis, inheriting 2 copies of abnormal genes means a person will get that disease. But for other diseases and conditions, the picture is more complex. For type 2 diabetes, testing positive for some specific gene variants may help predict risk, but no better than other factors—such as obesity, high blood pressure and having a close relative with the disease.

The latest approach to personalized medicine is to get your whole genome sequenced. That's still expensive, but the cost has dropped dramatically over the past decade and will likely continue to fall. Since your genome essentially stays the same over time, this information might one day become part of your medical record, so doctors could consult it as needed.

You can start to get a sense of your genetic risks by putting together your family's health history. A free online tool called My Family Health Portrait from the U.S. Surgeon General can help you and your doctor spot early warning signs of conditions that run in your family.

But personalized medicine isn't just about genes. You can learn a lot about your health risks by taking a close look at your current health and habits. Smoking, a poor diet, and lack of exercise can raise your risks for life-threatening health problems, such as heart disease and cancer. Talk to your health care provider about the steps you can take to understand and reduce your unique health risks.

Learn More

Visit this site to learn more about how FDA regulations come into play.

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MODULE 9: DNA TRANSCRIPTION AND TRANSLATION

WHY IT MATTERS: DNA TRANSCRIPTION AND TRANSLATION

Why learn about DNA transcription and translation?

By now you're familiar with genes and DNA sequences and how changes in the DNA can have a big impact. But the question still remains . . . how? How does DNA cause anything to happen?

Our bodies contain trillions of cells that need to be constructed in very precise ways. Much like when a construction company creates blueprints before they actually begin a project, your body needs some kind of plan to accomplish this.

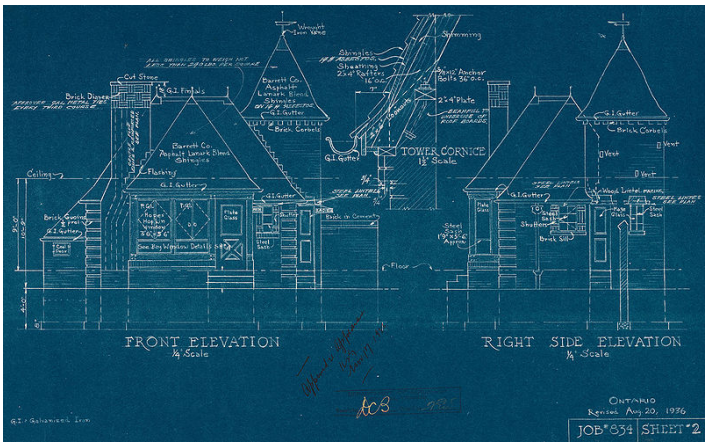

Figure 1. Before buildings are created, plans are drawn up. The plans direct the construction process.

DNA acts as a blueprint. With this plan in every cell, your body is able to convert DNA into action molecules, which are proteins, by way of an intermediary, RNA. This idea is so central to biology that it is often called the **central dogma of biology**: **DNA** is transcribed to **RNA** which is translated to **protein**. This multi-step process is one of the reasons for the diversity we see in the world around us.

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INTRODUCTION TO TRANSCRIPTION

What you'll learn to do: Outline the process of transcription

Have you ever had to transcribe something? Maybe someone left a message on your voicemail, and you had to write it down on paper. Or maybe you took notes in class, then rewrote them neatly to help you review.

As these examples show, *transcription* is a process in which information is rewritten. Transcription is something we do in our everyday lives, and it's also something our cells must do, in a more specialized and narrowly defined way. In biology, **transcription** is the process of copying out the DNA sequence of a gene in the similar alphabet of RNA.

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STEPS OF TRANSCRIPTION

Learning Outcomes

Understand the basic steps in the transcription of DNA into RNA

The process of **Transcription** takes place in the cytoplasm in prokaryotes and in nucleus in eukaryotes. It uses DNA as a template to make an RNA (mRNA) molecule. During transcription, a strand of mRNA is made that is complementary to a strand of DNA. Figure 1 shows how this occurs. Eventually portions of the transcribed mRNA will be made into functional proteins.

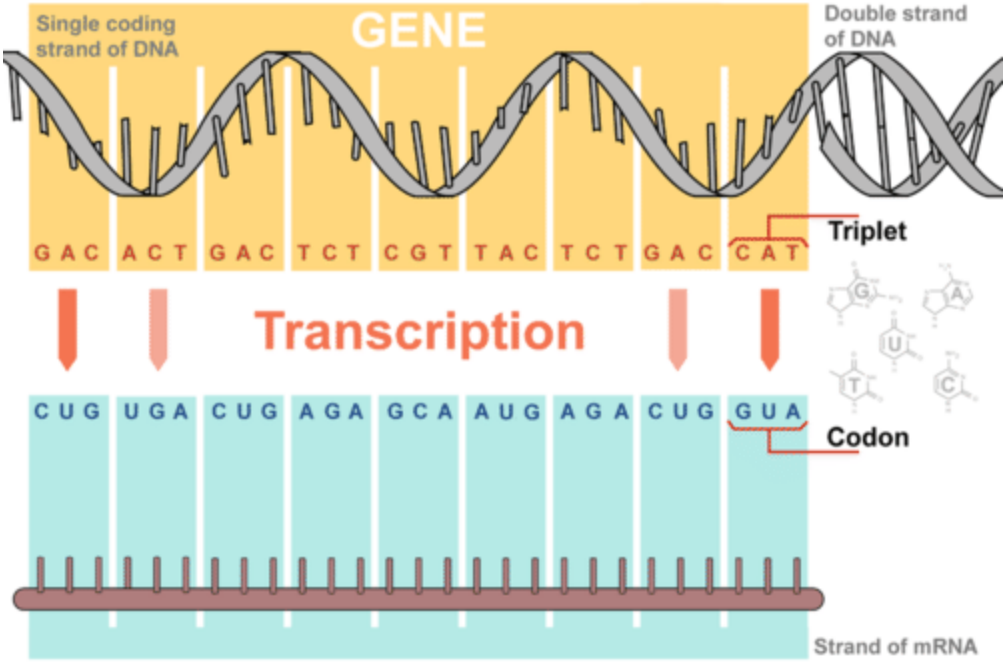

Figure 1. Overview of Transcription. Transcription uses the sequence of bases in a strand of DNA to make a complementary strand of mRNA. Triplets are groups of three successive nucleotide bases in DNA. Codons are complementary groups of bases in mRNA.

You can also watch this more detailed video about transcription.

Steps of Transcription

Transcription takes place in three steps: initiation, elongation, and termination. The steps are illustrated in Figure 2.

Step 1: Initiation

Initiation is the beginning of transcription. It occurs when the enzyme **RNA polymerase** binds to a region of a gene called the **promoter**. This signals the DNA to unwind so the enzyme can "read" the bases in one of the DNA strands. The enzyme is now ready to make a strand of mRNA with a complementary sequence of bases.

Step 2: Elongation

Elongation is the addition of nucleotides to the mRNA strand. RNA polymerase reads the unwound DNA strand and builds the mRNA molecule, using complementary base pairs. During this process, an adenine (A) in the DNA binds to an uracil (U) in the RNA.

Step 3: Termination

Termination is the ending of transcription, and occurs when RNA polymerase crosses a **stop (termination) sequence** in the gene. The mRNA strand is complete, and it detaches from DNA.

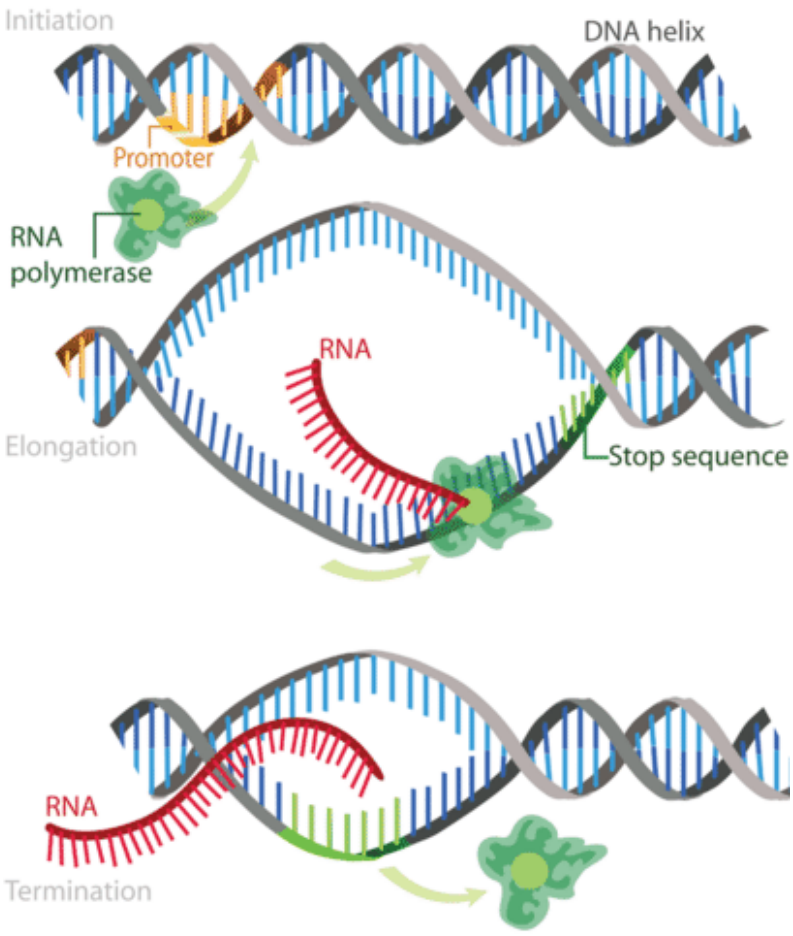

Figure 2. Transcription occurs in the three steps—initiation, elongation, and termination—all shown here.

This video provides a review of these steps. You can stop watching the video at 5:35. (After this point, it discusses translation, which we'll discuss in the next outcome.)

Watch this video online: https://youtu.be/h3b9ArupXZg

Visit this BioStudio animation to see the process of prokaryotic transcription.

Watch this video online: https://youtu.be/WsofH466lqk

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PRE-RNA AND MRNA

Learning Outcomes

Understand the difference between pre-RNA and mRNA

After transcription, eukaryotic **pre-mRNA**s must undergo several processing steps before they can be translated. Eukaryotic (and prokaryotic) tRNAs and rRNAs also undergo processing before they can function as components in the protein synthesis machinery.

mRNA Processing

The eukaryotic pre-mRNA undergoes extensive processing before it is ready to be translated. The additional steps involved in eukaryotic mRNA maturation create a molecule with a much longer half-life than a prokaryotic mRNA. Eukaryotic mRNAs last for several hours, whereas the typical *E. coli* mRNA lasts no more than five seconds.

The three most important steps of pre-mRNA processing are the addition of stabilizing and signaling factors at the 5′ and 3′ ends of the molecule, and the removal of intervening sequences that do not specify the appropriate amino acids.

5′ Capping

A cap is added to the 5′ end of the growing transcript by a phosphate linkage. This addition protects the mRNA from degradation. In addition, factors involved in protein synthesis recognize the cap to help initiate translation by ribosomes.

3′ Poly-A Tail

Once elongation is complete, an enzyme called poly-A polymerase adds a string of approximately 200 A residues, called the **poly-A tail** to the pre-mRNA. This modification further protects the pre-mRNA from degradation and signals the export of the cellular factors that the transcript needs to the cytoplasm.

Pre-mRNA Splicing

Eukaryotic genes are composed of **exons**, which correspond to protein-coding sequences (*ex*-on signifies that they are *ex*pressed), and *int*ervening sequences called **introns** (*int*ron denotes their *int*ervening role), which are removed from the pre-mRNA during processing. Intron sequences in mRNA do not encode functional proteins.

All of a pre-mRNA's introns must be completely and precisely removed before protein synthesis. If the process errs by even a single nucleotide, the reading frame of the rejoined exons would shift, and the resulting protein would be dysfunctional. The process of removing introns and reconnecting exons is called **splicing** (Figure 1).

Practice Question

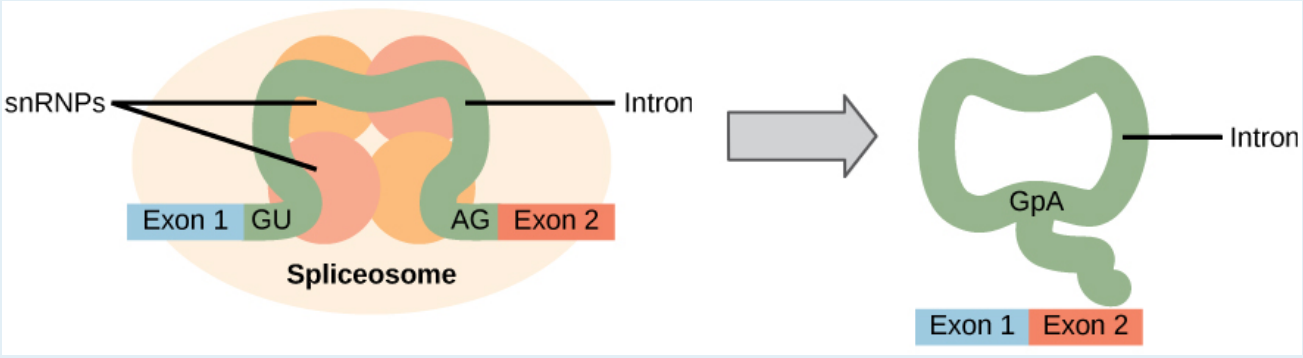

Figure 1. Pre-mRNA splicing involves the precise removal of introns from the primary RNA transcript. The splicing process is catalyzed by protein complexes called spliceosomes that are composed of proteins and RNA molecules called snRNAs. Spliceosomes recognize sequences at the 5′ and 3′ end of the intron.

Errors in splicing are implicated in cancers and other human diseases. What kinds of mutations might lead to splicing errors?

Answer

Think of different possible outcomes if splicing errors occur. Mutations in the spliceosome recognition sequence at each end of the intron, or in the proteins and RNAs that make up the spliceosome, may impair splicing. Mutations may also add new spliceosome recognition sites. Splicing errors could lead to introns being retained in spliced RNA, exons being excised, or changes in the location of the splice site.

See how introns are removed during RNA splicing at this website.

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INTRODUCTION TO TRANSLATION

What you'll learn to do: Summarize the process of translation

Take a moment to look at your hands. The bone, skin, and muscle you see are made up of cells. And each of those cells contains many millions of proteins As a matter of fact, proteins are key molecular "building blocks" for every organism on Earth!

How are these proteins made in a cell? For starters, the instructions for making proteins are "written" in a cell's DNA in the form of genes. Basically, a gene is used to build a protein in a two-step process:

- Step 1: transcription (which we just learned about)! Here, the DNA sequence of a gene is "rewritten" in the form of RNA. In eukaryotes like you and me, the RNA is processed (and often has a few bits snipped out of it) to make the final product, called a messenger RNA or mRNA.

- Step 2: translation! In this stage, the mRNA is “decoded” to build a protein (or a chunk/subunit of a protein) that contains a specific series of amino acids.

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REQUIREMENTS FOR TRANSLATION

Learning Outcomes

Describe the components needed for translation

The process of translation, or protein synthesis, involves the decoding of an mRNA message into a polypeptide product. Amino acids are covalently strung together by interlinking peptide bonds. Each individual amino acid has an amino group (NH_2) and a carboxyl (COOH) group. Polypeptides are formed when the amino group of one amino acid forms an amide (i.e., peptide) bond with the carboxyl group of another amino acid (Figure 1).

This reaction is catalyzed by ribosomes and generates one water molecule.

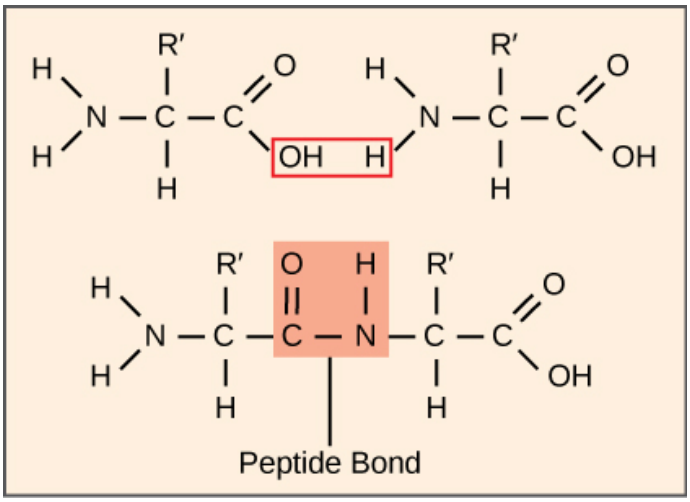

Figure 1. A peptide bond links the carboxyl end of one amino acid with the amino end of another, expelling one water molecule. For simplicity in this image, only the functional groups involved in the peptide bond are shown. The R and R′ designations refer to the rest of each amino acid structure.

The Protein Synthesis Machinery

In addition to the mRNA template, many molecules and macromolecules contribute to the process of translation. Translation requires the input of an **mRNA template**, **ribosomes**, **tRNAs**, and various enzymatic factors.

Click through the steps of this PBS interactive to see protein synthesis in action.

Ribosomes

A ribosome is a complex macromolecule composed of structural and catalytic rRNAs, and many distinct polypeptides. Ribosomes exist in the cytoplasm in prokaryotes and in the cytoplasm and rough endoplasmic reticulum in eukaryotes. Ribosomes are made up of two subunits. In *E. coli*, the small subunit is described as 30S, and the large subunit is 50S, for a total of 70S. Mammalian ribosomes have a small 40S subunit and a large 60S subunit, for a total of 80S. The small subunit is responsible for binding the mRNA template, whereas the large subunit sequentially binds tRNAs.

tRNAs

The tRNAs are structural RNA molecules that were transcribed from genes by RNA polymerase III. Serving as adaptors, specific tRNAs bind to sequences on the mRNA template and add the corresponding amino acid to the polypeptide chain. Therefore, tRNAs are the molecules that actually “translate” the language of RNA into the language of proteins.

Of the 64 possible mRNA codons—or triplet combinations of A, U, G, and C—three specify the termination of protein synthesis and 61 specify the addition of amino acids to the polypeptide chain. Of these 61, one codon (AUG) also known as the “start codon” encodes the initiation of translation. Each tRNA anticodon can base pair with one of the mRNA codons and add an amino acid or terminate translation, according to the genetic code. For instance, if the sequence CUA occurred on an mRNA template in the proper reading frame, it would bind a tRNA expressing the complementary sequence, GAU, which would be linked to the amino acid leucine.

Mature tRNAs take on a three-dimensional structure through intramolecular hydrogen bonding to position the amino acid binding site at one end and the **anticodon** at the other end (Figure 2).The anticodon is a three-nucleotide sequence in a tRNA that interacts with an mRNA codon through complementary base pairing.

tRNAs need to interact with three factors:

1. They must be recognized by the correct aminoacyl synthetase.
2. They must be recognized by ribosomes.
3. They must bind to the correct sequence in mRNA.

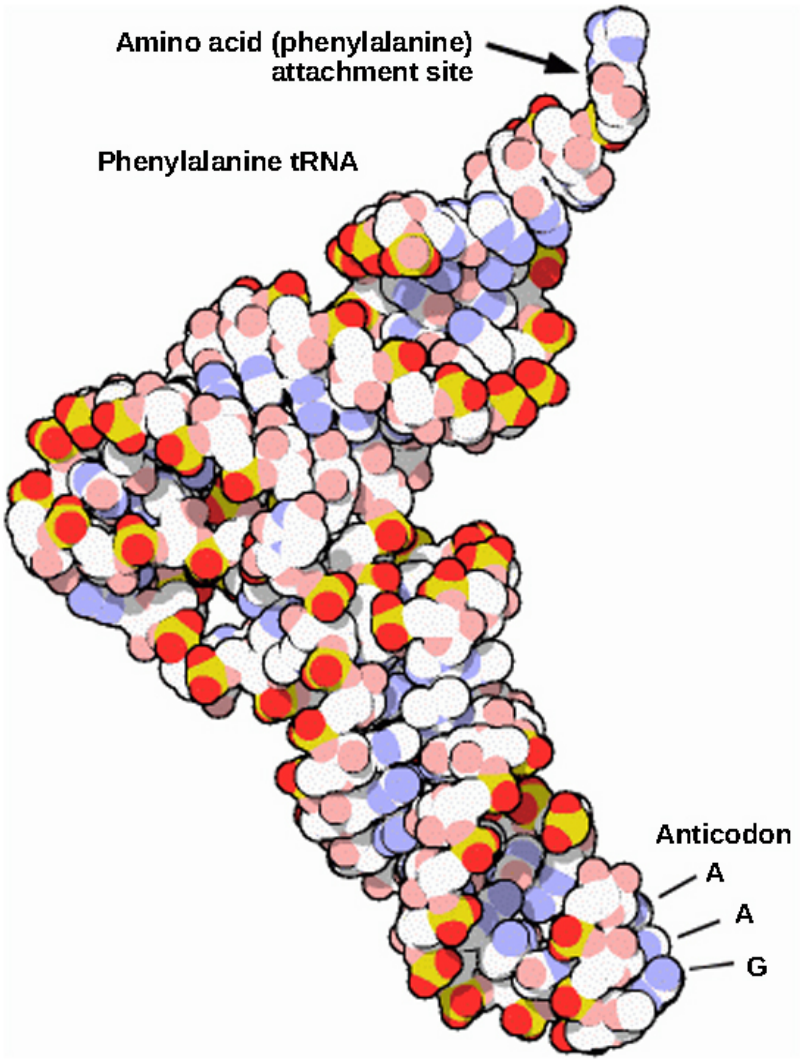

Figure 2. Phenylalanine tRNA

Aminoacyl tRNA Synthetases

Through the process of tRNA “charging,” each tRNA molecule is linked to its correct amino acid by a group of enzymes called aminoacyl tRNA synthetases. At least one type of **aminoacyl tRNA synthetase** exists for each of the 20 amino acids.

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GENETIC CODE

Learning Outcomes

Identify the components of the genetic code

Given the different numbers of “letters” in the mRNA and protein “alphabets,” scientists theorized that combinations of nucleotides corresponded to single amino acids. Scientists theorized that amino acids were encoded by **nucleotide triplets** and that the genetic code was **degenerate**. In other words, a given amino acid could be encoded by more than one nucleotide triplet. These nucleotide triplets are called **codons**. Scientists painstakingly solved the **genetic code** by translating synthetic mRNAs in vitro and sequencing the proteins they specified (Figure 1).

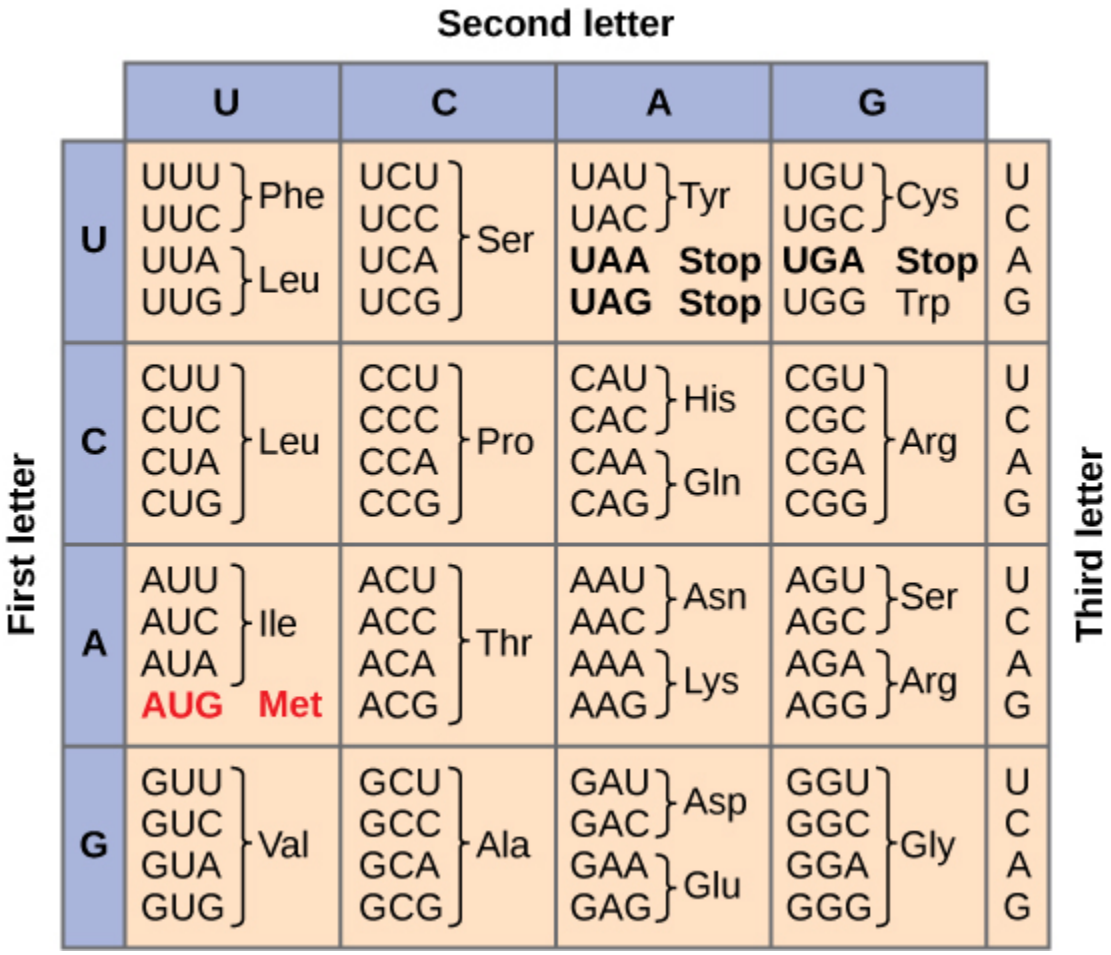

Figure 1. This figure shows the genetic code for translating each nucleotide triplet in mRNA into an amino acid or a termination signal in a nascent protein. (credit: modification of work by NIH)

In addition to instructing the addition of a specific amino acid to a polypeptide chain, three (**UAA**, **UAG**, **UGA**) of the 64 codons terminate protein synthesis and release the polypeptide from the translation machinery. These triplets are called **nonsense codons, or stop codons**. Another codon, **AUG**, also has a special function. In addition to specifying the amino acid methionine, it also serves as the **start codon** to initiate translation. The reading frame for translation is set by the AUG start codon near the 5′ end of the mRNA.

The genetic code is **universal**. With a few exceptions, virtually all species use the same genetic code for protein synthesis. Conservation of codons means that a purified mRNA encoding the globin protein in horses could be transferred to a tulip cell, and the tulip would synthesize horse globin. That there is only one genetic code is powerful evidence that all of life on Earth shares a common origin, especially considering that there are about 1084 possible combinations of 20 amino acids and 64 triplet codons.

Transcribe a gene and translate it to protein using complementary pairing and the genetic code at this site.

Degeneracy is believed to be a cellular mechanism to reduce the negative impact of random mutations. Codons that specify the same amino acid typically only differ by one nucleotide. In addition, amino acids with chemically similar side chains are encoded by similar codons. This nuance of the genetic code ensures that a single-nucleotide substitution mutation might either specify the same amino acid but have no effect or specify a similar amino acid, preventing the protein from being rendered completely nonfunctional.

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STEPS OF TRANSLATION

Learning Outcomes

Outline the basic steps of translation

As with mRNA synthesis, protein synthesis can be divided into three phases: **initiation**, **elongation**, **and termination**. The process of translation is similar in prokaryotes and eukaryotes. Here we'll explore how translation occurs in *E. coli*, a representative prokaryote, and specify any differences between prokaryotic and eukaryotic translation.

Initiation of Translation

Protein synthesis begins with the formation of an initiation complex. In *E. coli*, this complex involves the small 30S **ribosome**, the **mRNA template**, **initiation factors** and a special **initiator tRNA**. The initiator tRNA interacts with the **start codon AUG**. Guanosine triphosphate (GTP), which is a purine nucleotide triphosphate, acts as an energy source during translation—both at the start of elongation and during the ribosome's translocation.

Once the appropriate AUG is identified, the 50S subunit binds to the complex of Met-tRNAi, mRNA, and the 30S subunit. This step completes the initiation of translation.

Elongation of Translation

The 50S ribosomal subunit of *E. coli* consists of three compartments: the **A** (aminoacyl) **site** binds incoming charged aminoacyl tRNAs. The **P** (peptidyl) **site** binds charged tRNAs carrying amino acids that have formed peptide bonds with the growing polypeptide chain but have not yet dissociated from their corresponding tRNA. The **E** (exit) **site** releases dissociated tRNAs so that they can be recharged with free amino acids. this creates an initiation complex with a free A site ready to accept the tRNA corresponding to the first codon after the AUG.

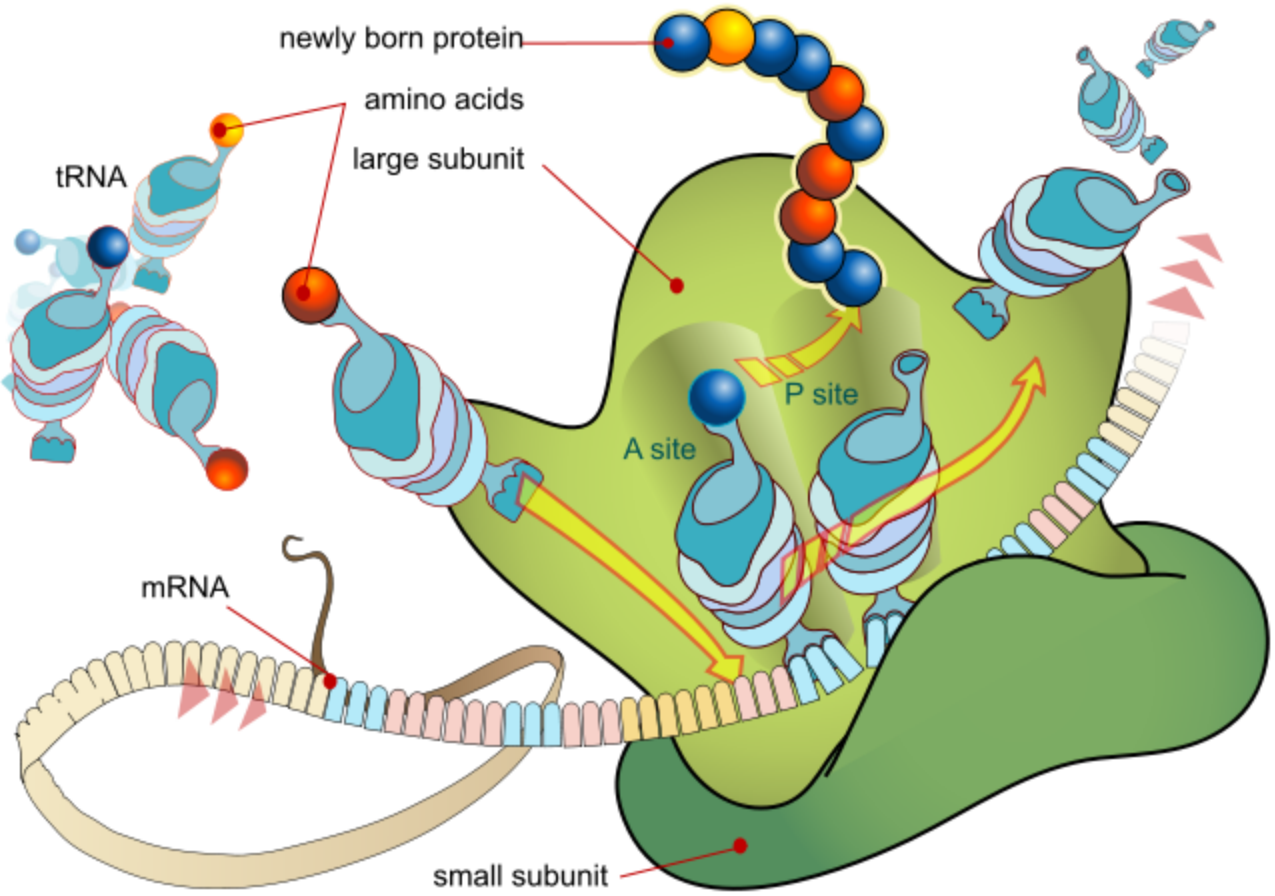

Figure 1. Ribosome mRNA translation

During translation elongation, the mRNA template provides specificity. As the ribosome moves along the mRNA, each mRNA codon comes into register, and specific binding with the corresponding charged tRNA anticodon is ensured. If mRNA were not present in the elongation complex, the ribosome would bind tRNAs nonspecifically.

Elongation proceeds with charged tRNAs entering the A site and then shifting to the P site followed by the E site with each single-codon "step" of the ribosome. Ribosomal steps are induced by conformational changes that advance the ribosome by three bases in the 3′ direction. The energy for each step of the ribosome is donated by an elongation factor that hydrolyzes GTP. Peptide bonds form between the amino group of the amino acid attached to the A-site tRNA and the carboxyl group of the amino acid attached to the P-site tRNA. The formation of each peptide bond is catalyzed by **peptidyl transferase**, an RNA-based enzyme that is integrated into the 50S ribosomal subunit. The energy for each peptide bond formation is derived from GTP hydrolysis, which is catalyzed by a separate elongation factor. The amino acid bound to the P-site tRNA is also linked to the growing polypeptide chain. As the ribosome steps across the mRNA, the former P-site tRNA enters the E site, detaches from the amino acid, and is expelled (Figure 2). Amazingly, the *E. coli* translation apparatus takes only 0.05 seconds to add each amino acid, meaning that a 200-amino acid protein can be translated in just 10 seconds.

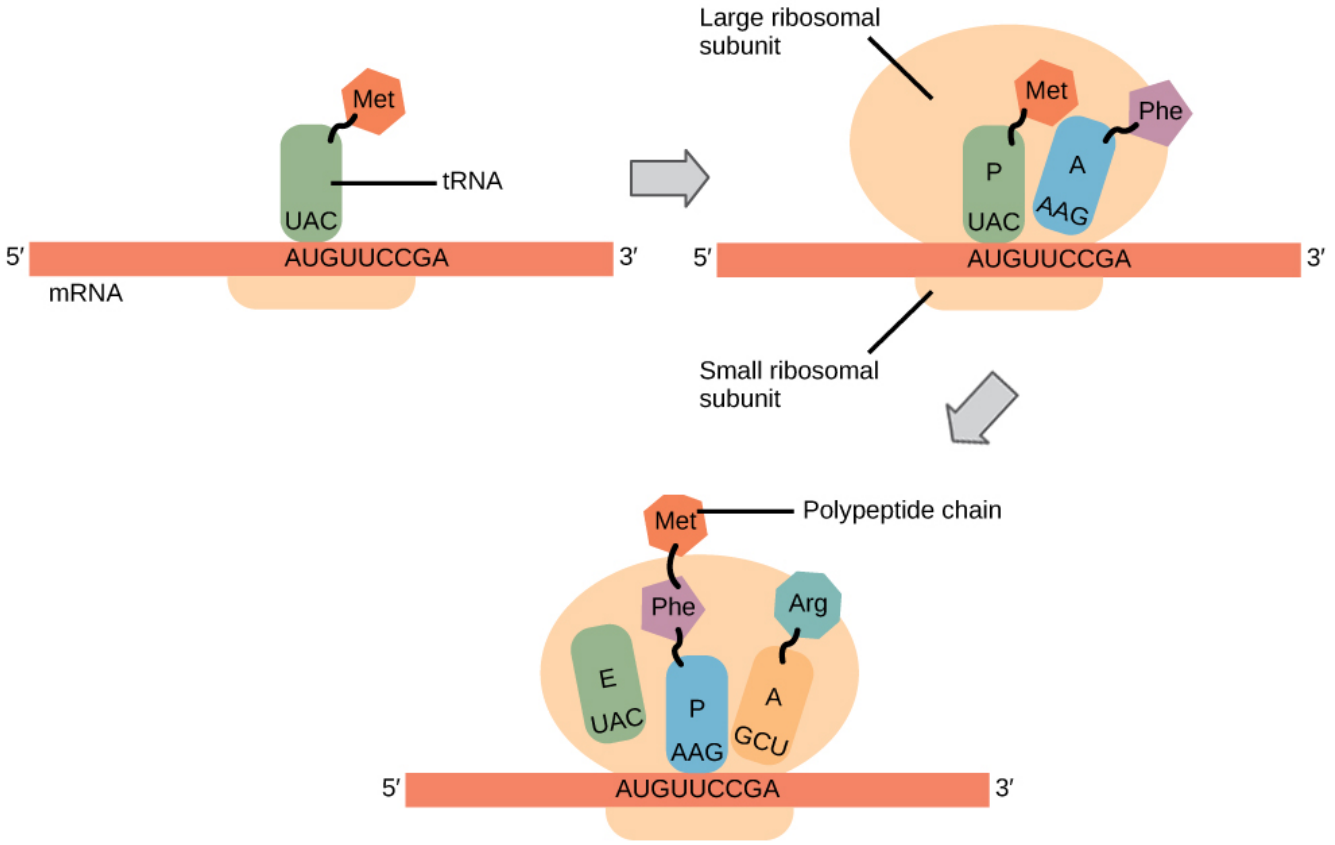

Figure 2. Translation begins when an initiator tRNA anticodon recognizes a codon on mRNA. The large ribosomal subunit joins the small subunit, and a second tRNA is recruited. As the mRNA moves relative to the ribosome, the polypeptide chain is formed. Entry of a release factor into the A site terminates translation and the components dissociate.

Practice Question

Many antibiotics inhibit bacterial protein synthesis. For example, tetracycline blocks the A site on the bacterial ribosome, and chloramphenicol blocks peptidyl transfer. What specific effect would you expect each of these antibiotics to have on protein synthesis?

Tetracycline would directly affect:

a. tRNA binding to the ribosome
b. ribosome assembly
c. growth of the protein chain

Answer

Answer a. Tetracycline would directly affect tRNA binding to the ribosome.

Practice Question

Chloramphenicol would directly affect

a. tRNA binding to the ribosome
b. ribosome assembly
c. growth of the protein chain

Answer

Answer c. Chloramphenicol would directly affect growth of the protein chain.

Termination of Translation

Termination of translation occurs when a nonsense codon (UAA, UAG, or UGA) is encountered. Upon aligning with the A site, these nonsense codons are recognized by release factors in prokaryotes and eukaryotes that instruct peptidyl transferase to add a water molecule to the carboxyl end of the P-site amino acid. This reaction forces the P-site amino acid to detach from its tRNA, and the newly made protein is released. The small and large ribosomal subunits dissociate from the mRNA and from each other; they are recruited almost immediately into another translation initiation complex. After many ribosomes have completed translation, the mRNA is degraded so the nucleotides can be reused in another transcription reaction.

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INTRODUCTION TO THE CENTRAL DOGMA

What you'll learn to do: Identify the central dogma of life

As you have learned, information flow in an organism takes place from DNA to RNA to protein. DNA dictates the structure of mRNA in a process known as transcription, and RNA dictates the structure of protein in a process known as translation. This is known as the **Central Dogma of Life**.

Does the Central Dogma always apply?

Scientists are always experimenting and exploring within their current understanding of the world. As they learn and discover new things, their ideas and understanding change to reflect the new evidence they have before them.

With modern research, it is becoming clear that some aspects of the central dogma are not entirely accurate. In order to flesh out our understanding, current research is focusing on investigating the function of non-coding RNA. Although this molecules does not follow the central dogma it still has a functional role in the cell.

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THE CENTRAL DOGMA

Learning Outcomes

Identify the central dogma of life

As you have learned, information flow in an organism takes place from DNA to RNA to protein:

- DNA is transcribed to RNA via complementary base pairing rules (but with U instead of T in the transcript)
- The RNA transcript, specifically mRNA, is then translated to an amino acid polypeptide
- Final folding and modifications of the polypeptide lead to functional proteins that actually do things in cells

This is known as the **Central Dogma of Life**, which holds true for all organisms.

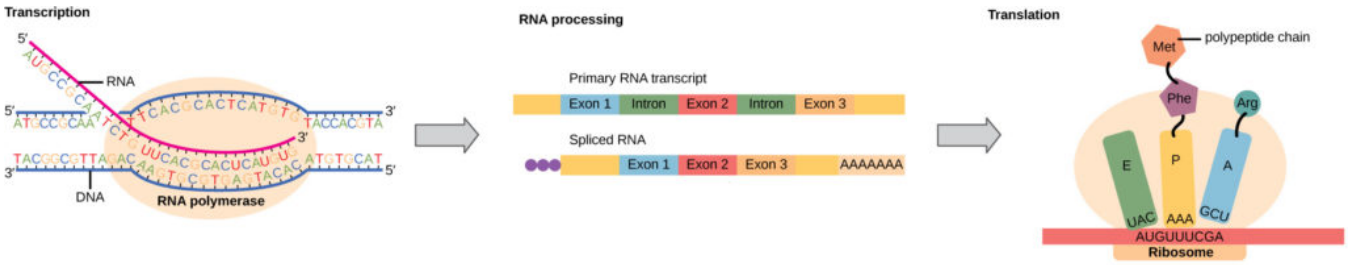

Figure 1. Click for a larger image. Instructions on DNA are transcribed onto messenger RNA. Ribosomes are able to read the genetic information inscribed on a strand of messenger RNA and use this information to string amino acids together into a protein.

Click here for a text-only version of the activity.

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INTRODUCTION TO DNA MUTATIONS

What you'll learn to do: Recognize the impact of DNA mutations

A mutation is a permanent alteration in the DNA sequence that makes up a gene; that is, the sequence differs from what is found in most people. Mutations range in size; they can affect anywhere from a single DNA building block (base pair) to a large segment of a chromosome that includes multiple genes. Gene mutations can be classified in two major ways:

- Hereditary mutations are inherited from a parent and are present throughout a person's life in virtually every cell in the body.

- Acquired (or somatic) mutations occur at some time during a person's life and are present only in certain cells, not in every cell in the body.

Mutations can impact an organism in both negative and positive ways—and sometimes a genetic mutation doesn't impact the organism at all!

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WHAT IS A MUTATION?

Learning Outcomes

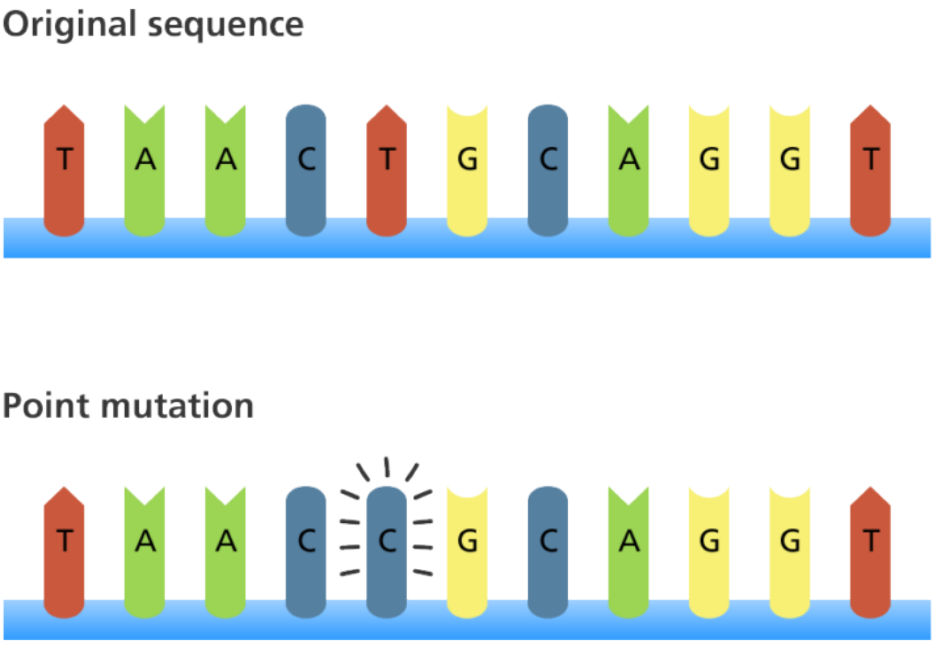
Understand what a mutation is and how one generally occurs

Over a lifetime, our DNA can undergo changes or **mutations** in the sequence of bases: A, C, G and T. This results in changes in the proteins that are made. This can be a bad or a good thing.

A mutation is a change that occurs in our DNA sequence, either due to mistakes when the DNA is copied or as the result of environmental factors such as UV light and cigarette smoke. Mutations can occur during DNA replication if errors are made and not corrected in time. Mutations can also occur as the result of exposure to environmental factors such as smoking, sunlight and radiation. Often cells can recognize any potentially mutation-causing damage and repair it before it becomes a fixed mutation.

Mutations contribute to genetic variation within species. Mutations can also be inherited, particularly if they occur in a germ cell (reproductive egg or sperm). Mutations that have a positive effect are more likely to be continually passed on. For example, the disorder sickle cell anaemia is caused by a mutation in the gene that instructs the building of a protein called hemoglobin. This causes the red blood cells to become an abnormal, rigid, sickle shape. However, in African populations, having this mutation also protects against malaria.

However, mutation can also disrupt normal gene activity and cause diseases, like cancer. Cancer is the most common human genetic disease; it is caused by mutations occurring in a number of growth-controlling genes. Sometimes faulty, cancer-causing genes can exist from birth, increasing a person's chance of getting cancer.



An illustration to show an example of a DNA mutation. Image credit: Genome Research Limited

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MAJOR TYPES OF MUTATIONS

Learning Outcomes

Identify the major types of DNA mutations

A well-studied example of a mutation is seen in people suffering from xeroderma pigmentosa (Figure 1). Affected individuals have skin that is highly sensitive to UV rays from the sun.

Figure 1. Xeroderma pigmentosa is a condition in which thymine dimerization from exposure to UV is not repaired. Exposure to sunlight results in skin lesions. (credit: James Halpern et al.)

When individuals are exposed to UV, pyrimidine dimers, especially those of thymine, are formed; people with xeroderma pigmentosa are not able to repair the damage. These are not repaired because of a defect in the nucleotide excision repair enzymes, whereas in normal individuals, the thymine dimers are excised and the defect is corrected. The thymine dimers distort the structure of the DNA double helix, and this may cause problems during DNA replication. People with xeroderma pigmentosa may have a higher risk of contracting skin cancer than those who don't have the condition.

Errors during DNA replication are not the only reason why mutations arise in DNA. **Mutations**, variations in the nucleotide sequence of a genome, can also occur because of damage to DNA. Such mutations may be of two types: induced or spontaneous. **Induced mutations** are those that result from an exposure to chemicals, UV rays, x-rays, or some other environmental agent. **Spontaneous mutations** occur without any exposure to any environmental agent; they are a result of natural reactions taking place within the body.

Mutations may have a wide range of effects. Some mutations have no impact on an organism; these are known as **silent mutations**. **Point mutations** are those mutations that affect a single base pair. The most common nucleotide mutations are substitutions, in which one base is replaced by another. These can be of two types, either transitions or transversions. **Transition substitution** refers to a purine or pyrimidine being replaced by a base of the same kind; for example, a purine such as adenine may be replaced by the purine guanine. Transversion substitution refers to a purine being replaced by a pyrimidine, or vice versa; for example, cytosine, a pyrimidine, is replaced by adenine, a purine. Mutations can also be the result of the addition of a base, known as an insertion, or the removal of a base, also known as deletion. Sometimes a piece of DNA from one chromosome may get moved to another chromosome or to another region of the same chromosome; this is also known as translocation.

Mutations in repair genes have been known to cause cancer. Many mutated repair genes have been implicated in certain forms of pancreatic cancer, colon cancer, and colorectal cancer. Mutations can affect either somatic cells or germ cells. If many mutations accumulate in a somatic cell, they may lead to problems such as the uncontrolled cell division observed in cancer. If a mutation takes place in germ cells, the mutation will be passed on to the next generation, as in the case of hemophilia and xeroderma pigmentosa.

The Causes of Genetic Mutations

Watch this video online: https://youtu.be/rtUoWyIE1FM

In Summary: Major Types of Mutations

DNA polymerase can make mistakes while adding nucleotides. Most mistakes are corrected, but if they are not, they may result in a mutation defined as a permanent change in the DNA sequence. Mutations can be of many types, such as **substitution**, **deletion**, **insertion**, and **translocation**. Mutations in repair genes may lead to serious consequences such as cancer. Mutations can be induced or may occur spontaneously.

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PUTTING IT TOGETHER: DNA TRANSCRIPTION AND TRANSLATION

As we've seen, transcription and translation are essential processes that enable life. These processes, also called the central dogma, are how our bodies—and all living organisms on earth—are built.

This video compares transcription and translation to something you may be a little more familiar with: creating a Hot Pocket.

Watch this video online: https://youtu.be/itsb2SqR-R0

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MODULE 10: GENE EXPRESSION

WHY IT MATTERS: GENE EXPRESSION

Why explain the regulation of gene expression?

Cancer is one of the top ten causes of death in the United States. It is not a single disease but includes many different diseases. In cancer cells, mutations modify cell-cycle control, and cells don't stop growing as they normally would. Mutations can also alter the growth rate or the progression of the cell through the cell cycle.

Thus, cancer can be described as a disease of altered gene expression. Changes at every level of eukaryotic gene expression can be detected in some form of cancer at some point in time. In order to understand how changes to gene expression can cause cancer, it is critical to understand how each stage of gene regulation works in normal cells. By understanding the mechanisms of control in normal, non-diseased cells, it will be easier for scientists to understand what goes wrong in disease states including complex ones like cancer.

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INTRODUCTION TO REGULATION OF GENE EXPRESSION

What you'll learn to do: Define the term regulation as it applies to genes

For a cell to function properly, necessary proteins must be synthesized at the proper time. All cells control or regulate the synthesis of proteins from information encoded in their DNA. The process of turning on a gene to produce RNA and protein is called **gene expression**. Whether in a simple unicellular organism or a complex multi-cellular organism, each cell controls when and how its genes are expressed. For this to occur, there must be a mechanism to control when a gene is expressed to make RNA and protein, how much of the protein is made, and when it is time to stop making that protein because it is no longer needed.

The regulation of gene expression conserves energy and space. It would require a significant amount of energy for an organism to express every gene at all times, so it is more energy efficient to turn on the genes only when they are required. In addition, only expressing a subset of genes in each cell saves space because DNA must be

unwound from its tightly coiled structure to transcribe and translate the DNA. Cells would have to be enormous if every protein were expressed in every cell all the time.

The control of gene expression is extremely complex. Malfunctions in this process are detrimental to the cell and can lead to the development of many diseases, including cancer.

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EXPRESSION OF GENES

Learning Outcomes

Discuss why every cell does not express all of its genes

Gene regulation makes cells different

Gene regulation is how a cell controls which genes, out of the many genes in its genome, are “turned on” (**expressed**). Thanks to gene regulation, each cell type in your body has a different set of active genes—despite the fact that almost all the cells of your body contain the exact same DNA. These different patterns of gene expression cause your various cell types to have different sets of proteins, making each cell type uniquely specialized to do its job. Ultimately gene expression can involve changes in transcription or translation, but in eukaryotes, most gene expression control occurs at transcription.

For example, one of the jobs of the liver is to remove toxic substances like alcohol from the bloodstream. To do this, liver cells express genes encoding subunits (pieces) of an enzyme called alcohol dehydrogenase. This enzyme breaks alcohol down into a non-toxic molecule. The neurons in a person’s brain don’t remove toxins from the body, so they keep these genes unexpressed, or “turned off.” Similarly, the cells of the liver don’t send signals using neurotransmitters, so they keep neurotransmitter genes turned off (Figure 1).

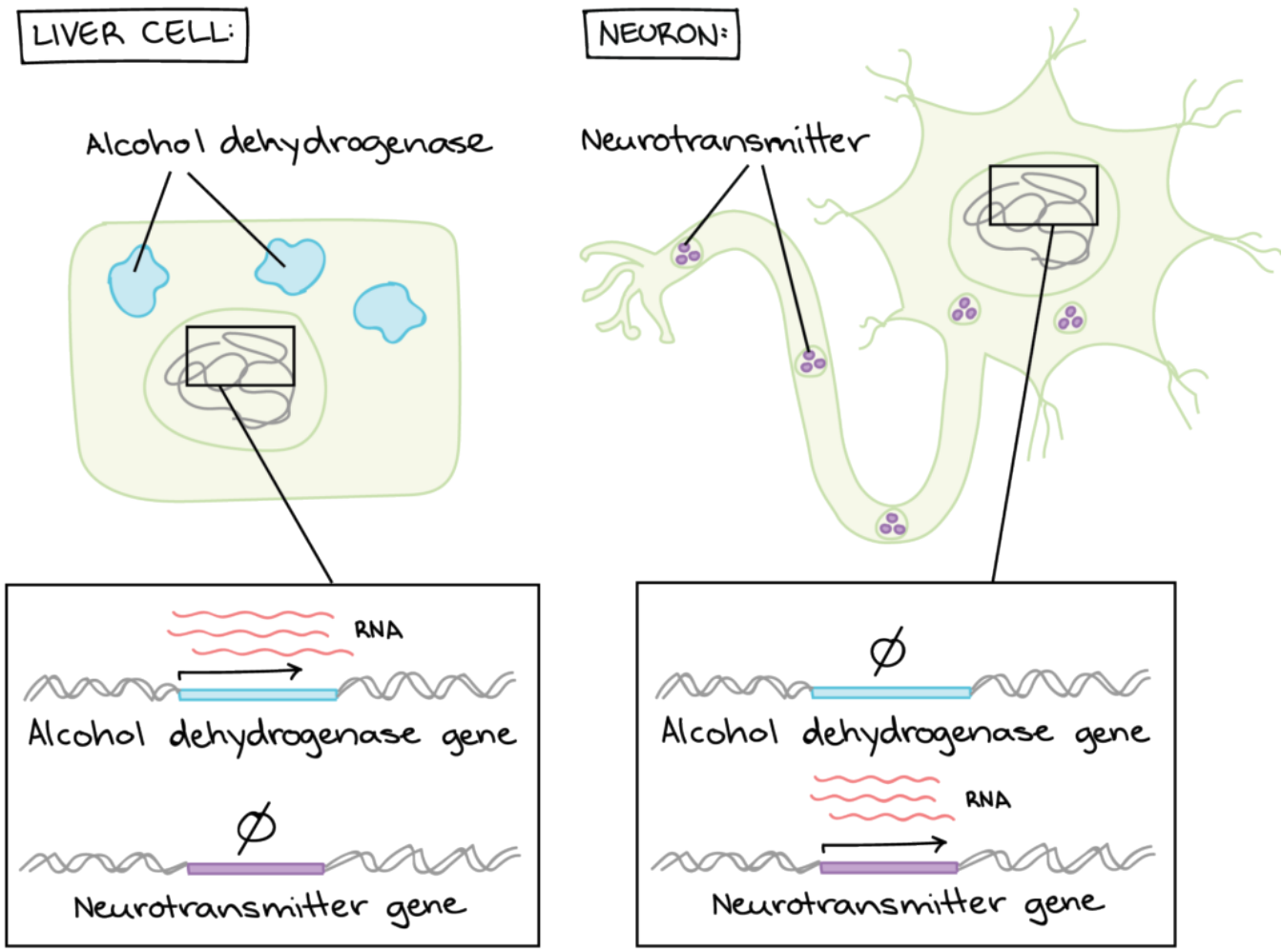

Figure 1. Different cells have different genes "turned on."

There are many other genes that are expressed differently between liver cells and neurons (or any two cell types in a multicellular organism like yourself).

How do cells "decide" which genes to turn on?

Now there's a tricky question! Different cell types express different sets of genes, as we saw above. However, two different cells of the same type may also have different gene expression patterns depending on their environment and internal state.

Broadly speaking, we can say that a cell's gene expression pattern is determined by information from both inside and outside the cell.

- Examples of information from *inside* the cell: the proteins it inherited from its mother cell, whether its DNA is damaged, and how much ATP it has.
- Examples of information from *outside* the cell: chemical signals from other cells, mechanical signals from the extracellular matrix, and nutrient levels.

How do these cues help a cell "decide" what genes to express? Cells don't make decisions in the sense that you or I would. Instead, they have molecular pathways that convert information—such as the binding of a chemical signal to its receptor—into a change in gene expression.

As an example, let's consider how cells respond to growth factors. A growth factor is a chemical signal from a neighboring cell that instructs a target cell to grow and divide. We could say that the cell "notices" the growth factor and "decides" to divide, but how do these processes actually occur?

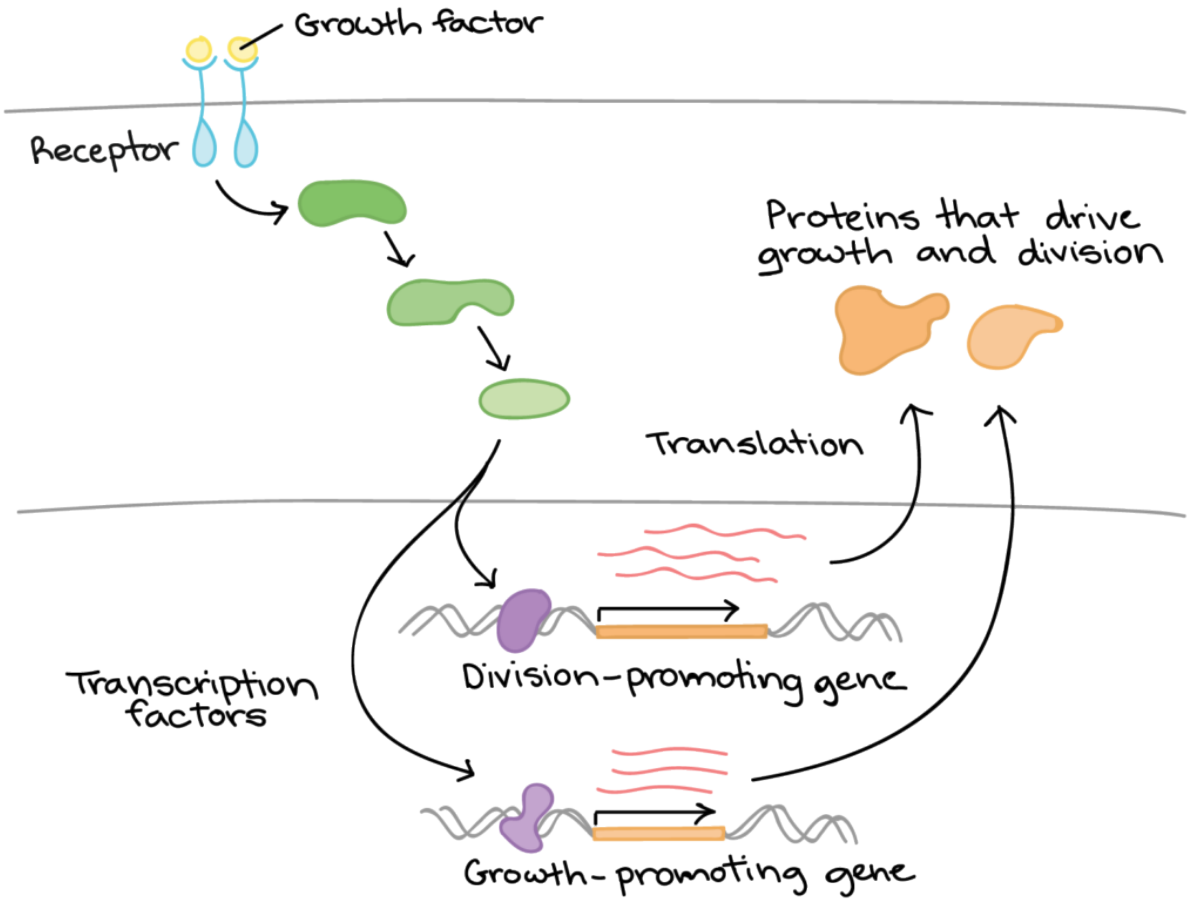

Figure 2. Growth factor prompting cell division

- The cell detects the growth factor through physical binding of the growth factor to a receptor protein on the cell surface.
- Binding of the growth factor causes the receptor to change shape, triggering a series of chemical events in the cell that activate proteins called transcription factors.
- The transcription factors bind to certain sequences of DNA in the nucleus and cause transcription of cell division-related genes.
- The products of these genes are various types of proteins that make the cell divide (drive cell growth and/or push the cell forward in the cell cycle).

This is just one example of how a cell can convert a source of information into a change in gene expression. There are many others, and understanding the logic of gene regulation is an area of ongoing research in biology today.

Growth factor signaling is complex and involves the activation of a variety of targets, including both transcription factors and non-transcription factor proteins.

In Summary: Expression of Genes

- Gene regulation is the process of controlling which genes in a cell's DNA are expressed (used to make a functional product such as a protein).

- Different cells in a multicellular organism may express very different sets of genes, even though they contain the same DNA.
- The set of genes expressed in a cell determines the set of proteins and functional RNAs it contains, giving it its unique properties.
- In eukaryotes like humans, gene expression involves many steps, and gene regulation can occur at any of these steps. However, many genes are regulated primarily at the level of transcription.

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PROKARYOTIC AND EUKARYOTIC GENE REGULATION

Learning Outcomes

Compare prokaryotic and eukaryotic gene regulation

To understand how gene expression is regulated, we must first understand how a gene codes for a functional protein in a cell. The process occurs in both prokaryotic and eukaryotic cells, just in slightly different manners.

Prokaryotic organisms are single-celled organisms that lack a cell nucleus, and their DNA therefore floats freely in the cell cytoplasm. To synthesize a protein, the processes of transcription and translation occur almost simultaneously. When the resulting protein is no longer needed, transcription stops. As a result, the primary method to control what type of protein and how much of each protein is expressed in a prokaryotic cell is the **regulation of DNA transcription**. All of the subsequent steps occur automatically. When more protein is required, more transcription occurs. Therefore, in prokaryotic cells, the control of gene expression is mostly at the transcriptional level.

Eukaryotic cells, in contrast, have intracellular organelles that add to their complexity. In eukaryotic cells, the DNA is contained inside the cell's nucleus and there it is transcribed into RNA. The newly synthesized RNA is then transported out of the nucleus into the cytoplasm, where ribosomes translate the RNA into protein. The processes of transcription and translation are physically separated by the nuclear membrane; transcription occurs only within the nucleus, and translation occurs only outside the nucleus in the cytoplasm. **The regulation of gene expression can occur at all stages of the process** (Figure 1). Regulation may occur when the DNA is uncoiled and loosened from nucleosomes to bind transcription factors (**epigenetic** level), when the RNA is transcribed (transcriptional level), when the RNA is processed and exported to the cytoplasm after it is transcribed (**post-transcriptional** level), when the RNA is translated into protein (translational level), or after the protein has been made (**post-translational** level).

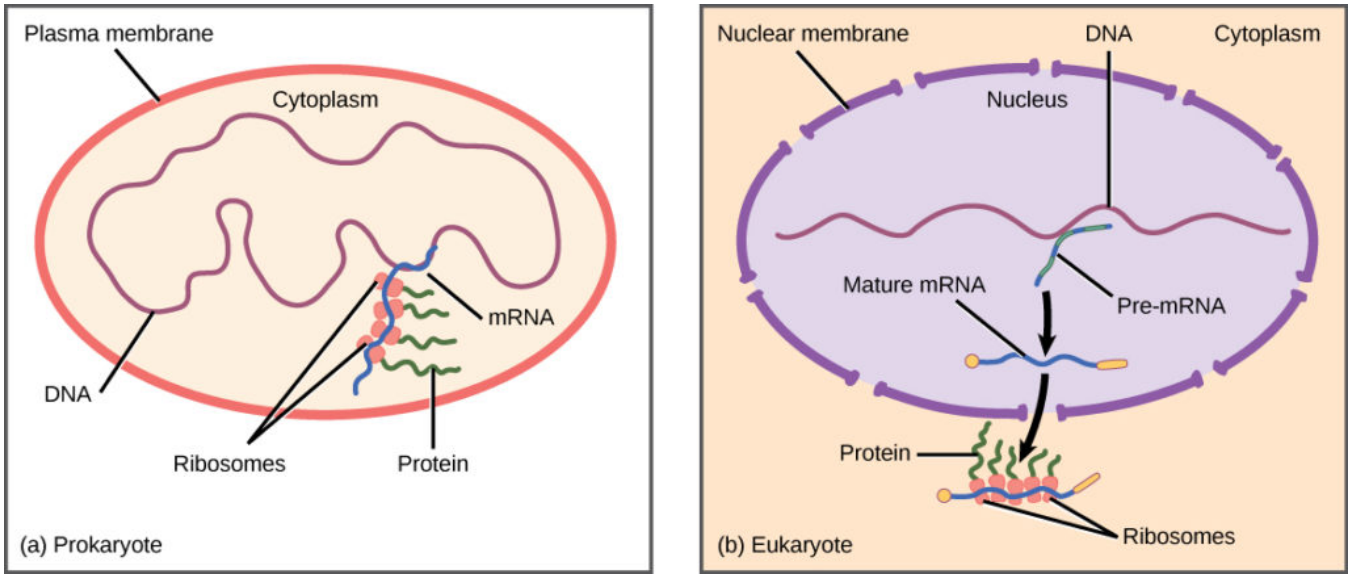

Figure 1. Prokaryotic transcription and translation occur simultaneously in the cytoplasm, and regulation occurs at the transcriptional level. Eukaryotic gene expression is regulated during transcription and RNA processing, which take place in the nucleus, and during protein translation, which takes place in the cytoplasm. Further regulation may occur through post-translational modifications of proteins.

The differences in the regulation of gene expression between prokaryotes and eukaryotes are summarized in Table 1. The regulation of gene expression in these types of organisms is discussed in detail in subsequent sections.

Table 1. Differences in the Regulation of Gene Expression of Prokaryotic and Eukaryotic Organisms

Prokaryotic organisms	Eukaryotic organisms
Lack nucleus	Contain nucleus
DNA is found in the cytoplasm	DNA is confined to the nuclear compartment
RNA transcription and protein formation occur almost simultaneously	RNA transcription occurs prior to protein formation, and it takes place in the nucleus. Translation of RNA to protein occurs in the cytoplasm.

Table 1. Differences in the Regulation of Gene Expression of Prokaryotic and Eukaryotic Organisms	
Prokaryotic organisms	Eukaryotic organisms
Gene expression is regulated primarily at the transcriptional level	Gene expression is regulated at many levels (epigenetic, transcriptional, nuclear shuttling, post-transcriptional, translational, and post-translational)

Practice Questions

Control of gene expression in eukaryotic cells occurs at which level(s)?

a. only the transcriptional level
b. epigenetic and transcriptional levels
c. epigenetic, transcriptional, and translational levels
d. epigenetic, transcriptional, post-transcriptional, translational, and post-translational levels

Answer

Answer d. Control of gene expression in eukaryotic cells occurs at epigenetic, transcriptional, post-transcriptional, translational, and post-translational levels.

Post-translational control refers to the:

a. regulation of gene expression after transcription
b. regulation of gene expression after translation
c. control of epigenetic activation
d. period between transcription and translation

Answer

Answer b. Post-translational control refers to the regulation of gene expression after translation

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INTRODUCTION TO PROKARYOTIC GENE REGULATION

What you'll learn to do: Understand the basic steps in gene regulation in prokaryotic cells

The DNA of prokaryotes is organized into a circular chromosome supercoiled in the nucleoid region of the cell cytoplasm. Proteins that are needed for a specific function are encoded together in blocks called **operons**. For example, all of the genes needed to use lactose as an energy source are coded next to each other in the lactose (or lac) operon.

In prokaryotic cells, there are three types of regulatory molecules that can affect the expression of operons: repressors, activators, and inducers. **Repressors** are proteins that suppress transcription of a gene in response to an external stimulus, whereas **activators** are proteins that increase the transcription of a gene in response to an external stimulus. Finally, **inducers** are small molecules that either activate or repress transcription depending on the needs of the cell and the availability of substrate.

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GENE REGULATION IN PROKARYOTES

Learning Outcomes

Understand the basic steps in gene regulation in prokaryotic cells

In bacteria and archaea, structural proteins with related functions—such as the genes that encode the enzymes that catalyze the many steps in a single biochemical pathway—are usually encoded together within the genome in a block called an **operon** and are transcribed together under the control of a single **promoter**. This forms a polycistronic transcript (Figure 1). The promoter then has simultaneous control over the regulation of the transcription of these structural genes because they will either all be needed at the same time, or none will be needed.

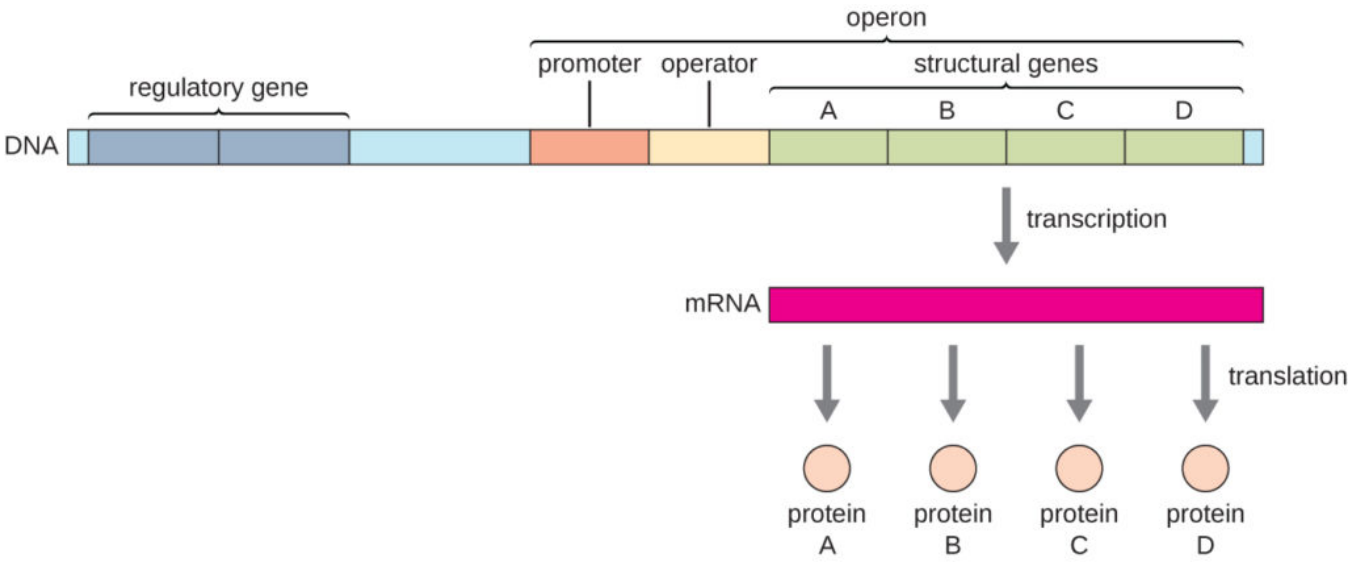

Figure 1. In prokaryotes, structural genes of related function are often organized together on the genome and transcribed together under the control of a single promoter. The operon's regulatory region includes both the promoter and the operator. If a repressor binds to the operator, then the structural genes will not be transcribed. Alternatively, activators may bind to the regulatory region, enhancing transcription.

French scientists François **Jacob** (1920–2013) and Jacques **Monod** at the Pasteur Institute were the first to show the organization of bacterial genes into operons, through their studies on the ***lac* operon** of *E. coli*. They found that in *E. coli*, all of the structural genes that encode enzymes needed to use lactose as an energy source lie next

to each other in the lactose (or *lac*) operon under the control of a single promoter, the *lac* promoter. For this work, they won the Nobel Prize in Physiology or Medicine in 1965.

Each operon includes DNA sequences that influence its own transcription; these are located in a region called the regulatory region. The regulatory region includes the promoter and the region surrounding the promoter, to which **transcription factors**, proteins encoded by regulatory genes, can bind. Transcription factors influence the binding of **RNA polymerase** to the promoter and allow its progression to transcribe structural genes. A **repressor** is a transcription factor that suppresses transcription of a gene in response to an external stimulus by binding to a DNA sequence within the regulatory region called the **operator**, which is located between the RNA polymerase binding site of the promoter and the transcriptional start site of the first structural gene. Repressor binding physically blocks RNA polymerase from transcribing structural genes. Conversely, an **activator** is a transcription factor that increases the transcription of a gene in response to an external stimulus by facilitating RNA polymerase binding to the promoter. An **inducer**, a third type of regulatory molecule, is a small molecule that either activates or represses transcription by interacting with a repressor or an activator.

Other genes in prokaryotic cells are needed all the time. These gene products will be **constitutively expressed**, or turned on continually. Most consitutively expressed genes are “housekeeping” genes responsible for overall maintenance of a cell.

Practice Question

What are the parts in the DNA sequence of an operon?

Answer

An operon is composed of a promoter, an operator, and the structural genes. They must occur in that order.

Practice Question

What types of regulatory molecules are there?

Answer

There are three types of regulatory molecules: repressors, activators, and inducers.

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INTRODUCTION TO EUKARYOTIC GENE REGULATION

What you'll learn to do: Discuss different components and types of epigenetic gene regulation

Your amazing body contains hundreds of different cell types, from immune cells to skin cells to neurons. Almost all of your cells contain the same set of DNA instructions: so why do they look so different, and do such different jobs? The answer: different gene regulation!

Gene regulation is how a cell controls which genes, out of the many genes in its genome, are "turned on" (expressed). Thanks to gene regulation, each cell type in your body has a different set of active genes – despite the fact that almost all the cells of your body contain the exact same DNA. These different patterns of gene expression cause your various cell types to have different sets of proteins, making each cell type uniquely specialized to do its job.

Eukaryotic gene expression is more complex than prokaryotic gene expression because the processes of transcription and translation are physically separated. Unlike prokaryotic cells, eukaryotic cells can regulate gene expression at many different levels. Eukaryotic gene expression begins with control of access to the DNA. This form of regulation, called epigenetic regulation, occurs even before transcription is initiated.

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EUKARYOTIC EPIGENETIC GENE REGULATION

Learning Outcomes

Explain the process of epigenetic regulation

The human genome encodes over 20,000 genes; each of the 23 pairs of human chromosomes encodes thousands of genes. The DNA in the nucleus is precisely wound, folded, and compacted into chromosomes so that it will fit into the nucleus. It is also organized so that specific segments can be accessed as needed by a specific cell type.

The first level of organization, or packing, is the winding of DNA strands around histone proteins. Histones package and order DNA into structural units called nucleosome complexes, which can control the access of proteins to the DNA regions (Figure 1a). Under the electron microscope, this winding of DNA around histone proteins to form nucleosomes looks like small beads on a string (Figure 1b). These beads (histone proteins) can move along the string (DNA) and change the structure of the molecule.

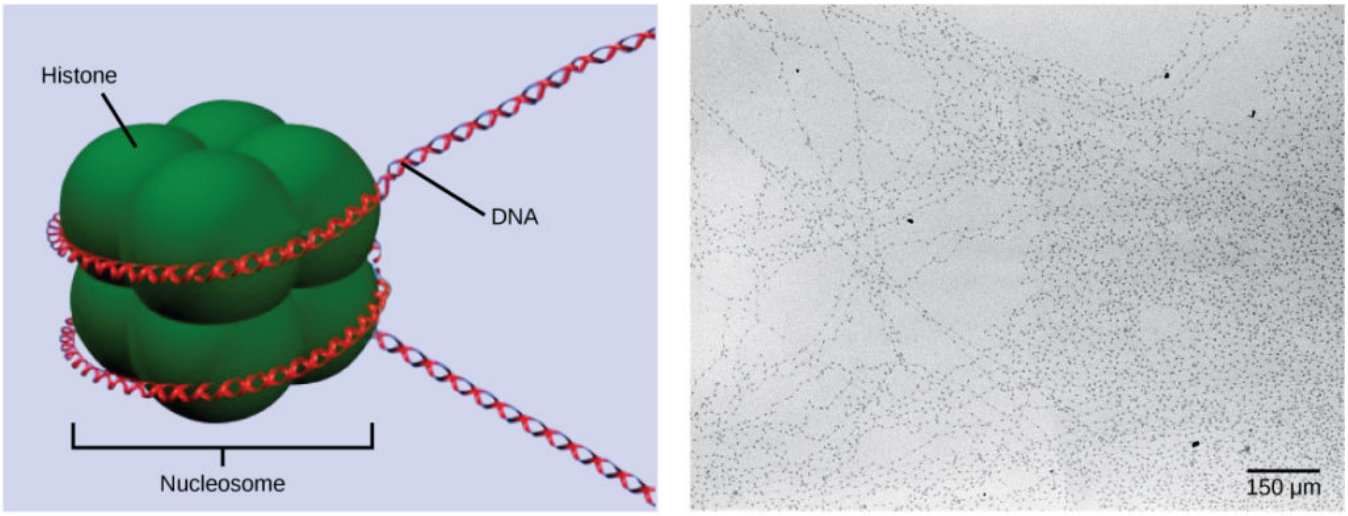
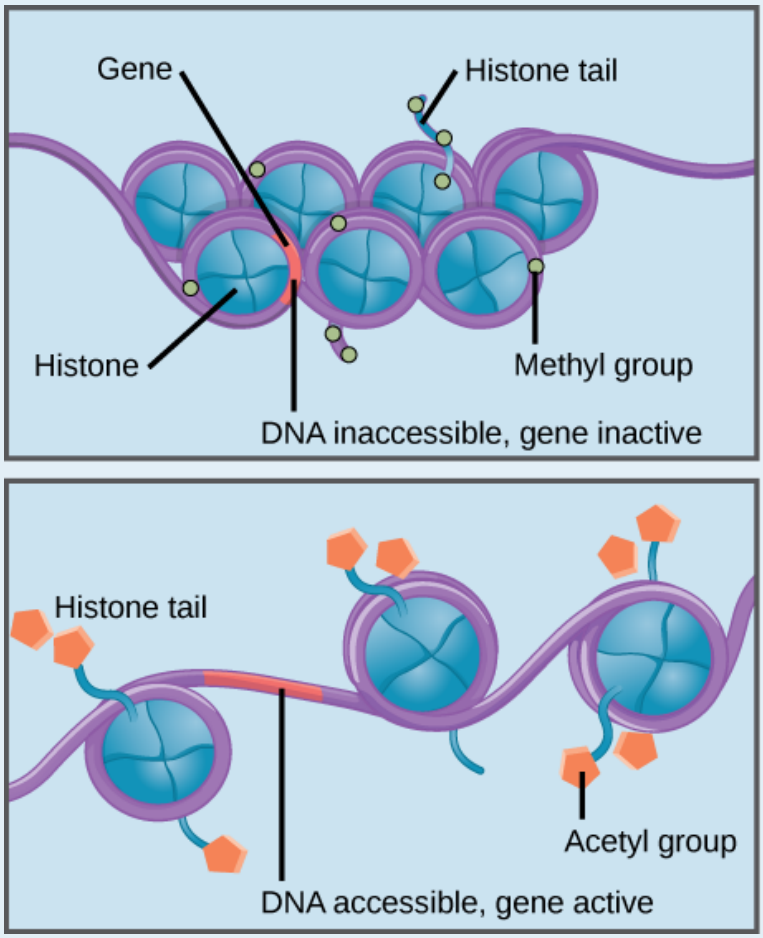


Figure 1. DNA is folded around histone proteins to create (a) nucleosome complexes. These nucleosomes control the access of proteins to the underlying DNA. When viewed through an electron microscope (b), the nucleosomes look like beads on a string. (credit "micrograph": modification of work by Chris Woodcock)

If DNA encoding a specific gene is to be transcribed into RNA, the nucleosomes surrounding that region of DNA can slide down the DNA to open that specific chromosomal region and allow for the transcriptional machinery (RNA polymerase) to initiate transcription (Figure 2). Nucleosomes can move to open the chromosome structure to expose a segment of DNA, but do so in a very controlled manner.

Practice Question



Methylation of DNA and histones causes nucleosomes to pack tightly together. Transcription factors cannot bind the DNA, and genes are not expressed.

Histone acetylation results in loose packing of nucleosomes. Transcription factors can bind the DNA and genes are expressed.

Figure 2. Nucleosomes can slide along DNA. When nucleosomes are spaced closely together (top), transcription factors cannot bind and gene expression is turned off. When the nucleosomes are spaced far apart (bottom), the DNA is exposed. Transcription factors can bind, allowing gene expression to occur. Modifications to the histones and DNA affect nucleosome spacing.

In females, one of the two X chromosomes is inactivated during embryonic development because of epigenetic changes to the chromatin. What impact do you think these changes would have on nucleosome packing?

Answer

The nucleosomes would pack more tightly together.

This type of gene regulation is called epigenetic regulation. Epigenetic means “around genetics.” The changes that occur to the histone proteins and DNA do not alter the nucleotide sequence and are not permanent. Instead, these changes are temporary (although they often persist through multiple rounds of cell division) and alter the chromosomal structure (open or closed) as needed. A gene can be turned on or off depending upon the location and modifications to the histone proteins and DNA.

View this video that describes how epigenetic regulation controls gene expression.

Watch this video online: https://youtu.be/eYrQ0EhVCYA

In Summary: Eukaryotic Epigenetic Gene Regulation

In eukaryotic cells, the first stage of gene expression control occurs at the epigenetic level. Epigenetic mechanisms control access to the chromosomal region to allow genes to be turned on or off. These mechanisms control how DNA is packed into the nucleus by regulating how tightly the DNA is wound around histone proteins. The addition or removal of chemical modifications (or flags) to histone proteins or DNA signals to the cell to open or close a chromosomal region. Therefore, eukaryotic cells can control whether a gene is expressed by controlling accessibility to transcription factors and the binding of RNA polymerase to initiate transcription.

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EUKARYOTIC TRANSCRIPTION GENE REGULATION

Learning Outcomes

Discuss the role of transcription factors in gene regulation

Like prokaryotic cells, the transcription of genes in eukaryotes requires the actions of an RNA polymerase to bind to a sequence upstream of a gene to initiate transcription. However, unlike prokaryotic cells, the eukaryotic RNA polymerase requires other proteins, or transcription factors, to facilitate transcription initiation. **Transcription factors** are proteins that bind to the **promoter** sequence and other regulatory sequences to control the transcription of the target gene. RNA polymerase by itself cannot initiate transcription in eukaryotic cells. Transcription factors must bind to the promoter region first and recruit RNA polymerase to the site for transcription to be established.

View the process of transcription—the making of RNA from a DNA template:
Watch this video online: https://youtu.be/WsofH466lqk

The Promoter and the Transcription Machinery

Genes are organized to make the control of gene expression easier. The **promoter region** is immediately upstream of the coding sequence. The purpose of the promoter is to bind transcription factors that control the initiation of transcription.

Enhancers and Transcription

In some eukaryotic genes, there are regions that help increase or enhance transcription. These regions, called **enhancers**, are not necessarily close to the genes they enhance. They can be located upstream of a gene, within the coding region of the gene, downstream of a gene, or may be thousands of nucleotides away. Enhancer regions are binding sequences, or sites, for transcription factors. When a DNA-bending protein binds, the shape of the DNA changes (Figure 1). This shape change allows for the interaction of the **activators** bound to the enhancers with the transcription factors bound to the promoter region and the RNA polymerase.

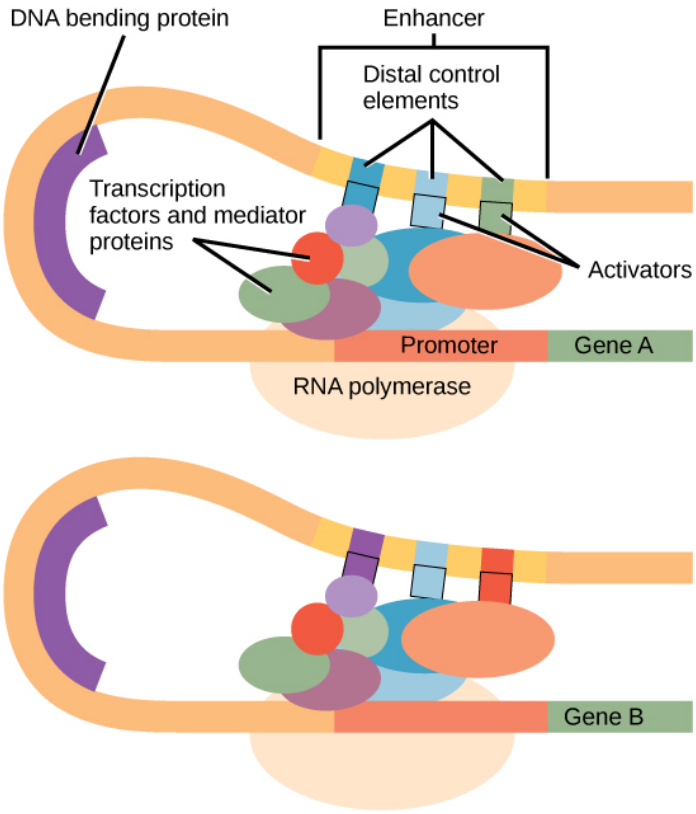

Figure 1. An enhancer is a DNA sequence that promotes transcription. Each enhancer is made up of short DNA sequences called distal control elements. Activators bound to the distal control elements interact with mediator proteins and transcription factors. Two different genes may have the same promoter but different distal control elements, enabling differential gene expression.

Turning Genes Off: Transcriptional Repressors

Like prokaryotic cells, eukaryotic cells also have mechanisms to prevent transcription. Transcriptional **repressors** can bind to promoter or enhancer regions and block transcription. Like the transcriptional activators, repressors respond to external stimuli to prevent the binding of activating transcription factors.

In Summary: Eukaryotic Transcription Gene Regulation

To start transcription, transcription factors, must first bind to the promoter and recruit RNA polymerase to that location. In addition to promoter sequences, enhancer regions help augment transcription. Enhancers can be upstream, downstream, within a gene itself, or on other chromosomes. Transcription factors bind to enhancer regions to increase or prevent transcription.

Practice Question

The binding of _____ is required for transcription to start.

a. a protein
b. DNA polymerase
c. RNA polymerase
d. a transcription factor

Answer

Answer c. The binding of RNA polymerase is required for transcription to start.

Practice Question

What will result from the binding of a transcription factor to an enhancer region?

a. decreased transcription of an adjacent gene
b. increased transcription of a distant gene
c. alteration of the translation of an adjacent gene
d. initiation of the recruitment of RNA polymerase

Answer

Answer b. Increased transcription of a distant gene will result from the binding of a transcription factor to an enhancer region.

Practice Question

A mutation within the promoter region can alter transcription of a gene. Describe how this can happen.

Answer

A mutation in the promoter region can change the binding site for a transcription factor that normally binds to increase transcription. The mutation could either decrease the ability of the transcription factor to bind, thereby decreasing transcription, or it can increase the ability of the transcription factor to bind, thus increasing transcription.

Practice Question

What could happen if a cell had too much of an activating transcription factor present?

Answer

If too much of an activating transcription factor were present, then transcription would be increased in the cell. This could lead to dramatic alterations in cell function.

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POST-TRANSLATIONAL CONTROL OF GENE EXPRESSION

Learning Outcomes

- Understand RNA splicing and explain its role in regulating gene expression
- Describe the importance of RNA stability in gene regulation

RNA is transcribed, but must be processed into a mature form before translation can begin. This processing after an RNA molecule has been transcribed, but before it is translated into a protein, is called post-transcriptional modification. As with the epigenetic and transcriptional stages of processing, this post-transcriptional step can also be regulated to control gene expression in the cell. If the RNA is not processed, shuttled, or translated, then no protein will be synthesized.

RNA splicing, the first stage of post-transcriptional control

In eukaryotic cells, the RNA transcript often contains regions, called **introns**, that are removed prior to translation. The regions of RNA that code for protein are called **exons** (Figure 1). After an RNA molecule has been transcribed, but prior to its departure from the nucleus to be translated, the RNA is processed and the introns are removed by **splicing**.

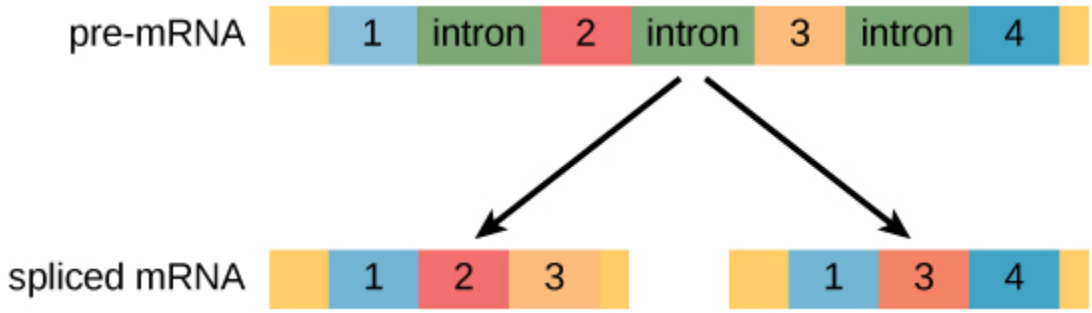

Figure 1. Pre-mRNA can be alternatively spliced to create different proteins.

Alternative RNA Splicing

Alternative RNA splicing is a mechanism that allows different protein products to be produced from one gene when different combinations of introns, and sometimes exons, are removed from the transcript (Figure 2).

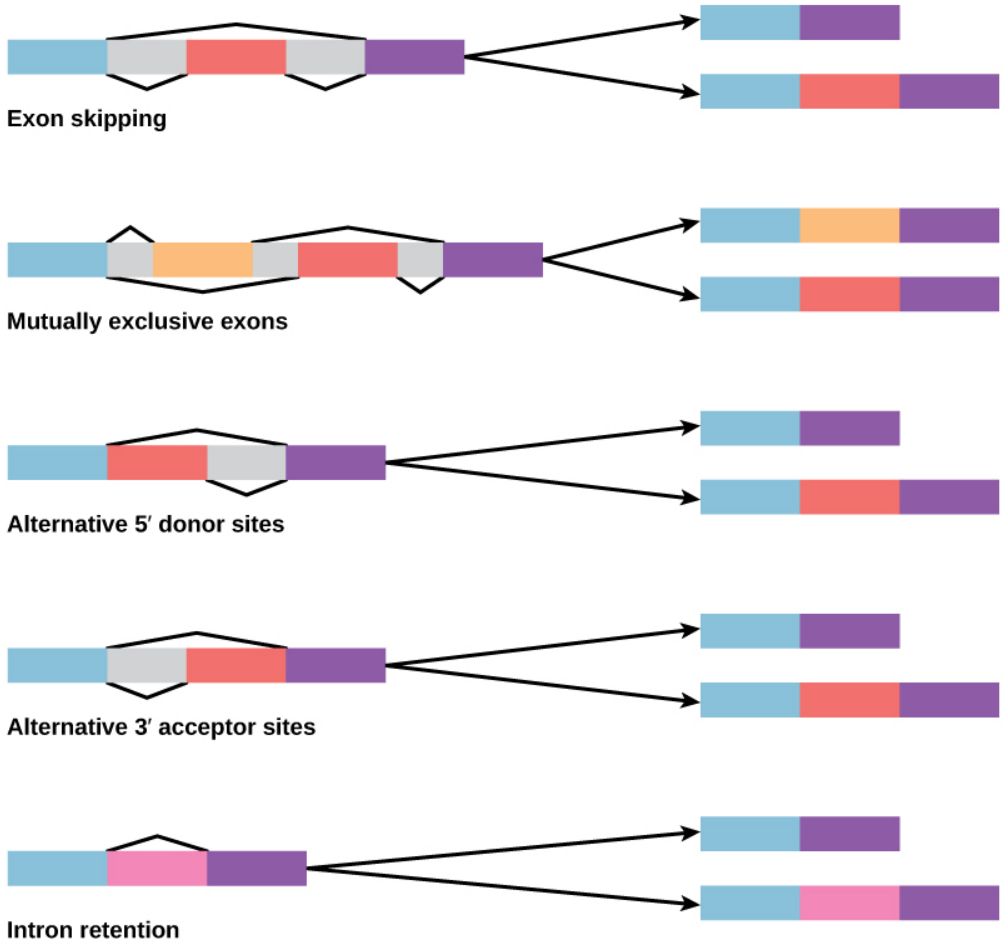

Figure 2. There are five basic modes of alternative splicing.

This alternative splicing can be haphazard, but more often it is controlled and acts as a mechanism of gene regulation.

> Visualize how mRNA splicing happens by watching the process in action in this video:
> Watch this video online: https://youtu.be/FVuAwBGw_pQ

Control of RNA Stability

Before the mRNA leaves the nucleus, it is given two protective "caps" that prevent the end of the strand from degrading during its journey. The **5′ cap**, which is placed on the 5′ end of the mRNA and **poly-A tail**, which is attached to the 3′ end. Once the RNA is transported to the cytoplasm, the length of time that the RNA resides there can be controlled. Each RNA molecule has a defined lifespan and decays at a specific rate. This rate of decay is referred to as the **RNA stability**. If the RNA is stable, it will be detected for longer periods of time in the cytoplasm.

RNA Stability and microRNAs

The **microRNAs**, or miRNAs, are short single-stranded RNA molecules that are only 21–24 nucleotides in length. Like transcription factors and RBPs, mature miRNAs recognize a specific sequence and bind to the RNA to degrade the target mRNA. They rapidly destroy the RNA molecule.

In Summary: Post-Translational Control of Gene Expression

Post-transcriptional control can occur at any stage after transcription, including RNA splicing, nuclear shuttling, and RNA stability. Once RNA is transcribed, it must be processed to create a mature RNA that is ready to be translated. This involves the removal of introns that do not code for protein. Spliceosomes bind to the signals that mark the exon/intron border to remove the introns and ligate the exons together. Once this occurs, the RNA is mature and can be translated. RNA is created and spliced in the nucleus, but needs to be transported to the cytoplasm to be translated. RNA is transported to the cytoplasm through the nuclear pore complex. Once the RNA is in the cytoplasm, the length of time it resides there before being degraded, called RNA stability, can also be altered to control the overall amount of protein that is synthesized. RNA stability is controlled by microRNAs (miRNAs). These miRNAs bind to the 5′ CAP or the 3′ Tail of the RNA to decrease RNA stability and promote decay.

Practice Question

Which of the following are involved in post-transcriptional control?

a. control of RNA splicing
b. control of RNA shuttling
c. control of RNA stability
d. all of the above

Answer

Answer d. All of the above (control of RNA splicing, RNA shuttling, and RNA stability) are involved in post-transcriptional control.

Practice Question

Binding of a miRNAs will _____ the stability of the RNA molecule.

a. increase
b. decrease
c. neither increase nor decrease
d. either increase or decrease

Answer

Answer b. Binding of a miRNAs will decrease the stability of the RNA molecule.

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PUTTING IT TOGETHER: GENE EXPRESSION

Let's return to our earlier look at cancer: One example of a gene modification that alters the growth rate is increased phosphorylation of cyclin B, a protein that controls the progression of a cell through the cell cycle and serves as a cell-cycle checkpoint protein.

For cells to move through each phase of the cell cycle, the cell must pass through checkpoints. This ensures that the cell has properly completed the step and has not encountered any mutation that will alter its function. Many proteins, including cyclin B, control these checkpoints. The phosphorylation of cyclin B, a post-translational event, alters its function. As a result, cells can progress through the cell cycle unimpeded, even if mutations exist in the cell and its growth should be terminated.

Cancer Treatment

Cancer therapies are designed to target actively dividing cells. Besides cancer cells, what other cells actively divide in humans? One of the most physically obvious side effects of cancer treatments is hair loss. This is because the living cells in the hair root continually divide to make hair grow longer. These cells therefore are often impacted by broad scale cancer treatments like chemotherapy drugs and radiation localized to the head. Shortly after cancer treatments, a patient's blood count may also drop. This is due to the rapid division of blood cells throughout a person's life. Unlike hair cells though, blood cells divide fairly often and rapidly. Hence, the blood count often returns to normal much faster than hair regrows.

New Drugs to Combat Cancer: Targeted Therapies

Scientists are using what is known about the regulation of gene expression in disease states, including cancer, to develop new ways to treat and prevent disease development. Many scientists are designing drugs on the basis of the gene expression patterns within individual tumors. This idea, that therapy and medicines can be tailored to an individual, has given rise to the field of personalized medicine. With an increased understanding of gene regulation and gene function, medicines can be designed to specifically target diseased cells without harming healthy cells. Some new medicines, called targeted therapies, have exploited the overexpression of a specific protein or the mutation of a gene to develop a new medication to treat disease. One such example is the use of anti-EGF receptor medications to treat the subset of breast cancer tumors that have very high levels of the EGF protein. Undoubtedly, more targeted therapies will be developed as scientists learn more about how gene expression changes can cause cancer.

As scientists learn more about cell division and the unique ways it malfunctions in cancer cells, they are able to develop targeted therapies. These drugs are still chemotherapies, but they are often focused on a particular feature of different types of cancer cells, making them less likely to target non-cancerous dividing cells. This reduces global side effects. Unfortunately, our understanding of cancer is still incomplete. Therefore, every day cancer researchers and clinicians work to manage and treat these horrible diseases.

Read more at www.cancer.org

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MODULE 11: TRAIT INHERITANCE

WHY IT MATTERS: TRAIT INHERITANCE

Why learn about trait inheritance?

Take a moment to pause and think about yourself and your family. How much do you look like your parents? What about the other members of your family: aunts, uncles, cousins, full siblings, half-siblings? Why do some family members look nearly identical to each other, while other family members seem as if they don't share two traits? The answer to this boils down to *how* traits are inherited.

Parents pass on traits to their children. Traits like blood type, cleft chin, dimples, and widow's peaks are all inherited in a fairly straight-forward, simple fashion. However, the inheritance of other traits is much more complex and harder to understand: these traits include height, skin color, and eye color. These more complex paths of inheritance are what cause some siblings to look so different from each other—despite having the same two parents.

Unfortunately, genetics also plays a role in the inheritance of some diseases such as Huntingdon's, sickle cell anemia, and Tay-Sachs disease. A degree in genetics can be used in careers ranging from a forensic examiner, a genetic counselor, a medical geneticist, a statistical geneticist, to a clinical technician.

Occupation Spotlight: Genetic Counselors

Genetic counselors use their understanding of the rules of heredity to help gauge the risks of genetic diseases. They may advise a couple of their risks for passing on certain genetic disorders, order genetic testing, and then explain the chances that some genetic diseases will show up in any offspring. Genetic counselors are there to help potential parents understand risk factors and the prognosis of genetic disorders, including basic treatment and management plans in the case of certain disorders.

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INTRODUCTION TO THE FATHER OF GENETICS

What you'll learn to do: Identify the impact of Gregor Mendel on the field of genetics and apply Mendel's two laws of genetics

Gregor Mendel is often referred to as the *Father of Genetics*. But just what did he do to earn this honorary title? Though farmers had known for centuries that crossbreeding of animals and plants could favor certain desirable traits, Mendel's pea plant experiments conducted between 1856 and 1863 established many of the rules of heredity.

While Mendel never enjoyed recognition in his lifetime, in the decades following his life, scientists would verify his research and learn more about genes and the special substance called DNA that carried each living thing's specific traits.

In this outcome we'll examine the work he did and how his work still impacts genetics today.

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MENDEL'S EXPERIMENTS AND HEREDITY

Learning Outcomes

Describe Mendel's study of garden peas and hereditary

Genetics is the study of heredity. Johann Gregor Mendel set the framework for genetics long before chromosomes or genes had been identified, at a time when meiosis was not well understood. Mendel selected a simple biological system and conducted methodical, quantitative analyses using large sample sizes. Because of Mendel's work, the fundamental principles of heredity were revealed. We now know that genes, carried on chromosomes, are the basic functional units of heredity with the capability to be replicated, expressed, or mutated. Today, the postulates put forth by Mendel form the basis of classical, or Mendelian, genetics. Not all genes are transmitted from parents to offspring

Figure 1. Experimenting with thousands of garden peas, Mendel uncovered the fundamentals of genetics. (credit: modification of work by Jerry Kirkhart)

according to Mendelian genetics, but Mendel's experiments serve as an excellent starting point for thinking about inheritance.

Mendel's Experiments and the Laws of Probability

Johann Gregor Mendel (1822–1884) (Figure 2) was a lifelong learner, teacher, scientist, and man of faith. As a young adult, he joined the Augustinian Abbey of St. Thomas in Brno in what is now the Czech Republic. Supported by the monastery, he taught physics, botany, and natural science courses at the secondary and university levels. In 1856, he began a decade-long research pursuit involving inheritance patterns in honeybees and plants, ultimately settling on pea plants as his primary **model system** (a system with convenient characteristics used to study a specific biological phenomenon to be applied to other systems). In 1865, Mendel presented the results of his experiments with nearly 30,000 pea plants to the local Natural History Society. He demonstrated that traits are transmitted faithfully from parents to offspring independently of other traits and in dominant and recessive patterns. In 1866, he published his work, *Experiments in Plant Hybridization,* in the proceedings of the Natural History Society of Brünn.

Figure 2. Johann Gregor Mendel is considered the father of genetics.

Mendel's work went virtually unnoticed by the scientific community that believed, incorrectly, that the process of inheritance involved a blending of parental traits that produced an intermediate physical appearance in offspring; this hypothetical process appeared to be correct because of what we know now as continuous variation. **Continuous variation** results from the action of many genes to determine a characteristic like human height. Offspring appear to be a "blend" of their parents' traits when we look at characteristics that exhibit continuous variation. The **blending theory of inheritance** asserted that the original parental traits were lost or absorbed by the blending in the offspring, but we now know that this is not the case. Mendel was the first researcher to see it. Instead of continuous characteristics, Mendel worked with traits that were inherited in distinct classes (specifically, violet versus white flowers); this is referred to as **discontinuous variation**. Mendel's choice of these kinds of traits allowed him to see experimentally that the traits were not blended in the offspring, nor were they absorbed, but rather that they kept their distinctness and could be passed on. In 1868, Mendel became abbot of the monastery and exchanged his scientific pursuits for his pastoral duties. He was not recognized for his extraordinary scientific contributions during his lifetime. In fact, it was not until 1900 that his work was rediscovered, reproduced, and revitalized by scientists on the brink of discovering the chromosomal basis of heredity.

Mendel's Model System

Mendel's seminal work was accomplished using the garden **pea, *Pisum sativum***, to study inheritance. This species naturally self-fertilizes, such that pollen encounters ova within individual flowers. The flower petals remain sealed tightly until after pollination, preventing pollination from other plants. The result is highly inbred, or "true-breeding," pea plants. These are plants that always produce offspring that look like the parent. By experimenting with true-breeding pea plants, Mendel avoided the appearance of unexpected traits in offspring that might occur if the plants were not true breeding. The garden pea also grows to maturity within one season, meaning that several generations could be evaluated over a relatively short time. Finally, large quantities of garden peas could be cultivated simultaneously, allowing Mendel to conclude that his results did not come about simply by chance.

Mendelian Crosses

Mendel performed **hybridizations**, which involve mating two true-breeding individuals that have different traits. In the pea, which is naturally self-pollinating, this is done by manually transferring pollen from the anther of a mature pea plant of one variety to the stigma of a separate mature pea plant of the second variety. In plants, pollen carries the male gametes (sperm) to the stigma, a sticky organ that traps pollen and allows the sperm to move down the pistil to the female gametes (ova) below. To prevent the pea plant that was receiving pollen from self-fertilizing and confounding his results, Mendel painstakingly removed all of the anthers from the plant's flowers before they had a chance to mature.

Plants used in first-generation crosses were called P_0, or parental generation one, plants (Figure 3). Mendel collected the seeds belonging to the P_0 plants that resulted from each cross and grew them the following season. These offspring were called the **F_1**, or the first filial (*filial* = offspring, daughter or son), generation. Once Mendel examined the characteristics in the F_1 generation of plants, he allowed them to self-fertilize naturally. He then collected and grew the seeds from the F_1 plants to produce the **F_2**, or second filial, generation. Mendel's experiments extended beyond the F_2 generation to the F_3 and F_4generations, and so on, but it was the ratio of characteristics in the P_0–F_1–F_2 generations that were the most intriguing and became the basis for Mendel's postulates.

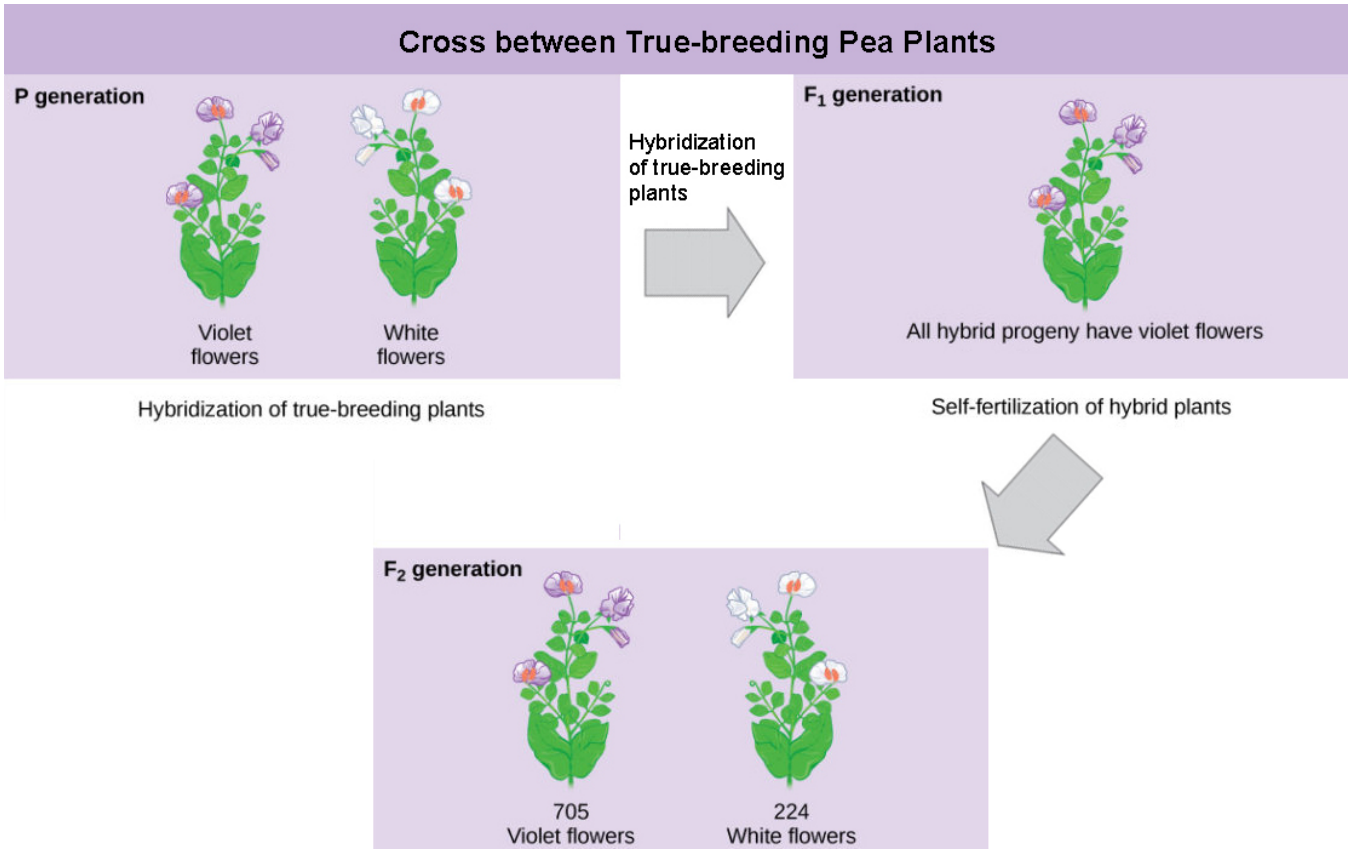

Figure 3. In one of his experiments on inheritance patterns, Mendel crossed plants that were true-breeding for violet flower color with plants true-breeding for white flower color (the P_0 generation). The resulting hybrids in the F_1 generation all had violet flowers. In the F_2 generation, approximately three quarters of the plants had violet flowers, and one quarter had white flowers.

Garden Pea Characteristics Revealed the Basics of Heredity

In his 1865 publication, Mendel reported the results of his crosses involving seven different characteristics, each with two contrasting traits. A **trait** is defined as a variation in the physical appearance of a heritable characteristic. The characteristics included plant height, seed texture, seed color, flower color, pea pod size, pea pod color, and

flower position. For the characteristic of flower color, for example, the two contrasting traits were white versus violet. To fully examine each characteristic, Mendel generated large numbers of F_1 and F_2 plants, reporting results from 19,959 F_2 plants alone. His findings were consistent.

What results did Mendel find in his crosses for flower color? First, Mendel confirmed that he had plants that bred true for white or violet flower color. Regardless of how many generations Mendel examined, all self-crossed offspring of parents with white flowers had white flowers, and all self-crossed offspring of parents with violet flowers had violet flowers. In addition, Mendel confirmed that, other than flower color, the pea plants were physically identical.

Once these validations were complete, Mendel applied the pollen from a plant with violet flowers to the stigma of a plant with white flowers. After gathering and sowing the seeds that resulted from this cross, Mendel found that 100 percent of the F_1 hybrid generation had violet flowers. Conventional wisdom at that time would have predicted the hybrid flowers to be pale violet or for hybrid plants to have equal numbers of white and violet flowers. In other words, the contrasting parental traits were expected to blend in the offspring. Instead, Mendel's results demonstrated that the white flower trait in the F_1 generation had completely disappeared.

Importantly, Mendel did not stop his experimentation there. He allowed the F_1 plants to self-fertilize and found that, of F_2-generation plants, 705 had violet flowers and 224 had white flowers. This was a ratio of 3.15 violet flowers per one white flower, or approximately **3:1**. When Mendel transferred pollen from a plant with violet flowers to the stigma of a plant with white flowers and vice versa, he obtained about the same ratio regardless of which parent, male or female, contributed which trait. This is called a **reciprocal cross**—a paired cross in which the respective traits of the male and female in one cross become the respective traits of the female and male in the other cross. For the other six characteristics Mendel examined, the F_1 and F_2 generations behaved in the same way as they had for flower color. One of the two traits would disappear completely from the F_1 generation only to reappear in the F_2 generation at a ratio of approximately 3:1 (Table 1).

Table 1. The Results of Mendel's Garden Pea Hybridizations

Characteristic	Contrasting P_0 Traits	F_1 Offspring Traits	F_2 Offspring Traits	F_2 Trait Ratios
Flower color	Violet vs. white	100 percent violet	• 705 violet • 224 white	3.15:1
Flower position	Axial vs. terminal	100 percent axial	• 651 axial • 207 terminal	3.14:1
Plant height	Tall vs. dwarf	100 percent tall	• 787 tall • 277 dwarf	2.84:1
Seed texture	Round vs. wrinkled	100 percent round	• 5,474 round • 1,850 wrinkled	2.96:1
Seed color	Yellow vs. green	100 percent yellow	• 6,022 yellow • 2,001 green	3.01:1
Pea pod texture	Inflated vs. constricted	100 percent inflated	• 882 inflated • 299 constricted	2.95:1

Table 1. The Results of Mendel's Garden Pea Hybridizations				
Characteristic	Contrasting P_0 Traits	F_1 Offspring Traits	F_2 Offspring Traits	F_2 Trait Ratios
Pea pod color	Green vs. yellow	100 percent green	• 428 green • 152 yellow	2.82:1

Upon compiling his results for many thousands of plants, Mendel concluded that the characteristics could be divided into expressed and latent traits. He called these, respectively, dominant and recessive traits. **Dominant traits** are those that are inherited unchanged in a hybridization. **Recessive traits** become latent, or disappear, in the offspring of a hybridization. The recessive trait does, however, reappear in the progeny of the hybrid offspring. An example of a dominant trait is the violet-flower trait. For this same characteristic (flower color), white-colored flowers are a recessive trait. The fact that the recessive trait reappeared in the F_2 generation meant that the traits remained separate (not blended) in the plants of the F_1 generation. Mendel also proposed that plants possessed two copies of the trait for the flower-color characteristic, and that each parent transmitted one of its two copies to its offspring, where they came together. Moreover, the physical observation of a dominant trait could mean that the genetic composition of the organism included two dominant versions of the characteristic or that it included one dominant and one recessive version. Conversely, the observation of a recessive trait meant that the organism lacked any dominant versions of this characteristic.

In Summary: Mendel's Experiments and Heredity

Working with garden pea plants, Mendel found that crosses between parents that differed by one trait produced F_1 offspring that all expressed the traits of one parent. Observable traits are referred to as dominant, and non-expressed traits are described as recessive. When the offspring in Mendel's experiment were self-crossed, the F_2 offspring exhibited the dominant trait or the recessive trait in a 3:1 ratio, confirming that the recessive trait had been transmitted faithfully from the original P_0 parent. Reciprocal crosses generated identical F_1 and F_2 offspring ratios. By examining large sample sizes, Mendel showed that his crosses behaved reproducibly according to the laws of probability, and that the traits were inherited as independent events.

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CHARACTERISTICS AND TRAITS

Learning Outcomes

Understand how the inheritance of a genotype generates a phenotype

The seven characteristics that Mendel evaluated in his pea plants were each expressed as one of two versions, or **traits**. The physical expression of characteristics is accomplished through the expression of genes carried on chromosomes. The genetic makeup of peas consists of two similar or homologous copies of each chromosome, one from each parent. Each pair of homologous chromosomes has the same linear order of genes. In other words, peas are diploid organisms in that they have two copies of each chromosome. The same is true for many

other plants and for virtually all animals. Diploid organisms utilize meiosis to produce haploid gametes, which contain one copy of each homologous chromosome that unite at fertilization to create a diploid zygote.

For cases in which a single gene controls a single characteristic, a diploid organism has two genetic copies that may or may not encode the same version of that characteristic. Gene variants that arise by mutation and exist at the same relative locations on homologous chromosomes are called **alleles**. Mendel examined the inheritance of genes with just two allele forms, but it is common to encounter more than two alleles for any given gene in a natural population.

Phenotypes and Genotypes

Two alleles for a given gene in a diploid organism are expressed and interact to produce physical characteristics. The observable traits expressed by an organism are referred to as its **phenotype**. An organism's underlying genetic makeup, consisting of both physically visible and non-expressed alleles, is called its **genotype**. Mendel's hybridization experiments demonstrate the difference between phenotype and genotype. When true-breeding plants in which one parent had yellow pods and one had green pods were cross-fertilized, all of the F_1 hybrid offspring had yellow pods. That is, the hybrid offspring were phenotypically identical to the true-breeding parent with yellow pods. However, we know that the allele donated by the parent with green pods was not simply lost because it reappeared in some of the F_2 offspring. Therefore, the F_1 plants must have been genotypically different from the parent with yellow pods.

The P_0 plants that Mendel used in his experiments were each homozygous for the trait he was studying: that is what made them 'true breeding'. Diploid organisms that are **homozygous** at a given gene, or locus, have two identical alleles for that gene on their homologous chromosomes. Mendel's parental pea plants always bred true because both of the gametes produced carried the same trait. When P_0 plants with contrasting traits were cross-fertilized, all of the offspring were heterozygous for the contrasting trait, meaning that their genotype reflected that they had different alleles for the gene being examined.

Dominant and Recessive Alleles

Our discussion of homozygous and heterozygous organisms brings us to why the F_1 heterozygous offspring were identical to one of the parents, rather than expressing both alleles. In all seven pea-plant characteristics, one of the two contrasting alleles was dominant, and the other was recessive. Mendel called the dominant allele the expressed unit factor; the recessive allele was referred to as the latent unit factor. We now know that these so-called unit factors are actually genes on homologous chromosome pairs. For a gene that is expressed in a dominant and recessive pattern, homozygous dominant and heterozygous organisms will look identical (that is, they will have different genotypes but the same phenotype). The recessive allele will only be observed in homozygous recessive individuals (Table 1).

Table 1. Human Inheritance in Dominant and Recessive Patterns

Dominant Traits	Recessive Traits
Achondroplasia	Albinism
Brachydactyly	Cystic fibrosis
Huntington's disease	Duchenne muscular dystrophy
Marfan syndrome	Galactosemia
Neurofibromatosis	Phenylketonuria
Widow's peak	Sickle-cell anemia

Table 1. Human Inheritance in Dominant and Recessive Patterns	
Dominant Traits	Recessive Traits
Wooly hair	Tay-Sachs disease

Several conventions exist for referring to genes and alleles. For the purposes of this chapter, we will abbreviate genes using the first letter of the gene's corresponding dominant trait. For example, violet is the dominant trait for a pea plant's flower color, so the flower-color gene would be abbreviated as *V* (note that it is customary to italicize gene designations). Furthermore, we will use uppercase and lowercase letters to represent dominant and recessive alleles, respectively.

Therefore, we would refer to the genotype of a homozygous dominant pea plant with violet flowers as *VV*, a homozygous recessive pea plant with white flowers as *vv*, and a heterozygous pea plant with violet flowers as *Vv*.

The Punnett Square Approach for a Monohybrid Cross

When fertilization occurs between two true-breeding parents that differ in only one characteristic, the process is called a **monohybrid** cross, and the resulting offspring are monohybrids. Mendel performed seven monohybrid crosses involving contrasting traits for each characteristic. On the basis of his results in F_1 and F_2 generations, Mendel postulated that each parent in the monohybrid cross contributed one of two paired unit factors to each offspring, and every possible combination of unit factors was equally likely.

To demonstrate a monohybrid cross, consider the case of true-breeding pea plants with yellow versus green pea seeds. The dominant seed color is yellow; therefore, the parental genotypes were *YY* for the plants with yellow seeds and *yy* for the plants with green seeds, respectively. A **Punnett square**, devised by the British geneticist Reginald Punnett, can be drawn that applies the rules of probability to predict the possible outcomes of a genetic cross or mating and their expected frequencies. To prepare a Punnett square, all possible combinations of the parental alleles are listed along the top (for one parent) and side (for the other parent) of a grid, representing their meiotic segregation into haploid gametes. Then the combinations of egg and sperm are made in the boxes in the table to show which alleles are combining. Each box then represents the diploid genotype of a zygote, or fertilized egg, that could result from this mating. Because each possibility is equally likely, genotypic ratios can be determined from a Punnett square. If the pattern of inheritance (dominant or recessive) is known, the phenotypic ratios can be inferred as well. For a monohybrid cross of two true-breeding parents, each parent contributes one type of allele. In this case, only one genotype is possible. All offspring are *Yy* and have yellow seeds (Figure 1).

A self-cross of one of the *Yy* heterozygous offspring can be represented in a 2 × 2 Punnett square because each parent can donate one of two different alleles. Therefore, the offspring can potentially have one of four allele combinations: *YY*, *Yy*, *yY*, or *yy* (Figure 1). Notice that there are two ways to obtain the *Yy* genotype: a *Y* from the egg and a *y* from the sperm, or a *y* from the egg and a *Y* from the sperm. Both of these possibilities must be counted. Recall that Mendel's pea-plant characteristics behaved in the same way in reciprocal crosses. Therefore, the two possible heterozygous combinations produce offspring that are genotypically and phenotypically identical despite their dominant and recessive alleles deriving from different parents. They are grouped together. Because fertilization is a random event, we expect each combination to be equally likely and for the offspring to exhibit a ratio of *YY*:*Yy*:*yy* genotypes of 1:2:1 (Figure 1). Furthermore, because the *YY* and *Yy* offspring have yellow seeds and are phenotypically identical, applying the sum rule of probability, we expect the offspring to exhibit a phenotypic ratio of 3 yellow:1 green. Indeed, working with large sample sizes, Mendel observed approximately this ratio in every F_2 generation resulting from crosses for individual traits.

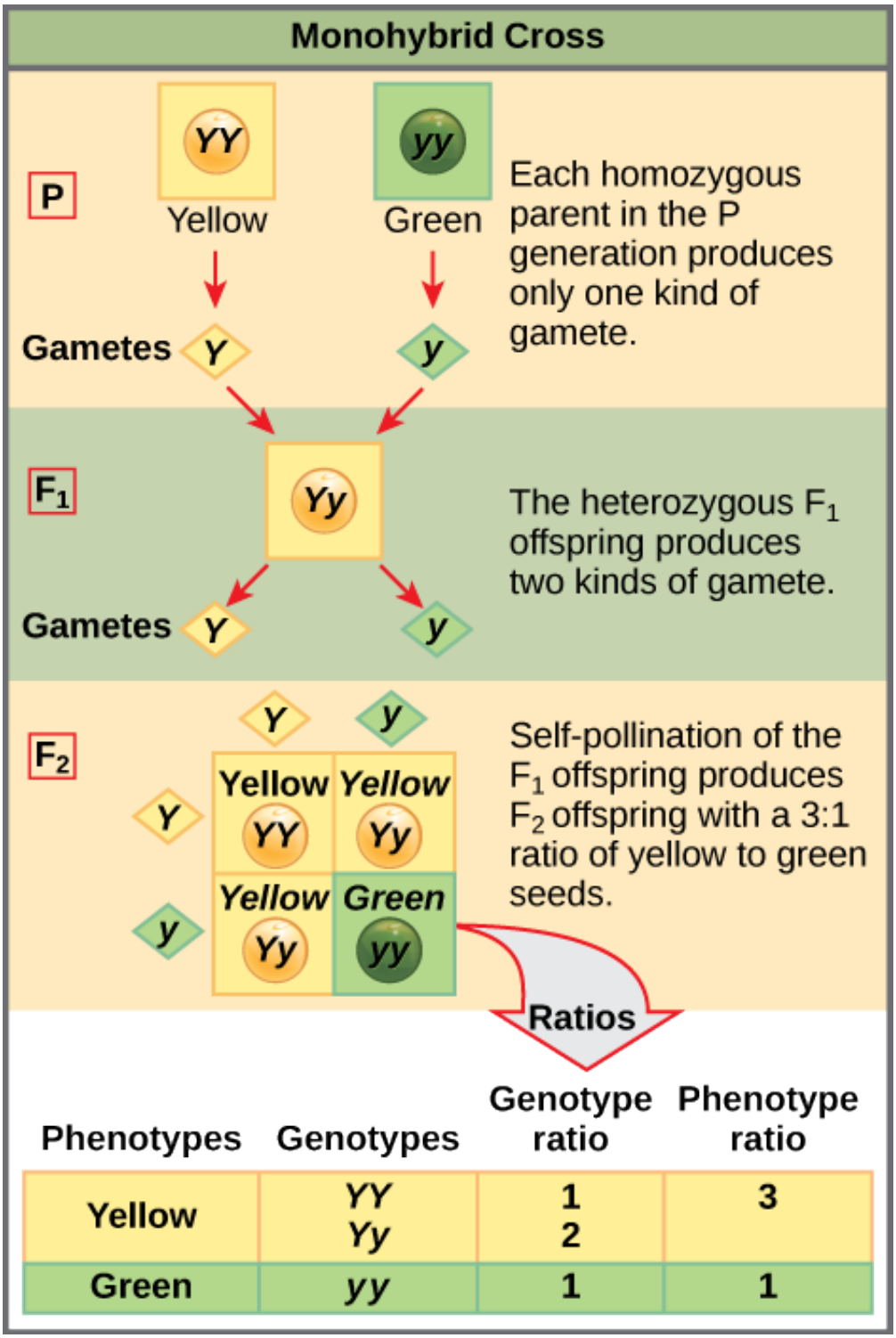

Figure 1. In the P_0 generation, pea plants that are true-breeding for the dominant yellow phenotype are crossed with plants with the recessive green phenotype. This cross produces F_1 heterozygotes with a yellow phenotype. Punnett square analysis can be used to predict the genotypes of the F_2 generation.

Mendel validated these results by performing an F_3 cross in which he self-crossed the dominant- and recessive-expressing F_2 plants. When he self-crossed the plants expressing green seeds, all of the offspring had green seeds, confirming that all green seeds had homozygous genotypes of *yy*. When he self-crossed the F_2 plants expressing yellow seeds, he found that one-third of the plants bred true, and two-thirds of the plants segregated

at a 3:1 ratio of yellow:green seeds. In this case, the true-breeding plants had homozygous (*YY*) genotypes, whereas the segregating plants corresponded to the heterozygous (*Yy*) genotype. When these plants self-fertilized, the outcome was just like the F_1 self-fertilizing cross.

The Test Cross Distinguishes the Dominant Phenotype

Beyond predicting the offspring of a cross between known homozygous or heterozygous parents, Mendel also developed a way to determine whether an organism that expressed a dominant trait was a heterozygote or a homozygote. Called the **test cross**, this technique is still used by plant and animal breeders. In a test cross, the dominant-expressing organism is crossed with an organism that is homozygous recessive for the same characteristic. If the dominant-expressing organism is a homozygote, then all F_1 offspring will be heterozygotes expressing the dominant trait (Figure 2). Alternatively, if the dominant expressing organism is a heterozygote, the F_1 offspring will exhibit a 1:1 ratio of heterozygotes and recessive homozygotes (Figure 2). The test cross further validates Mendel's postulate that pairs of unit factors segregate equally.

Practice Question

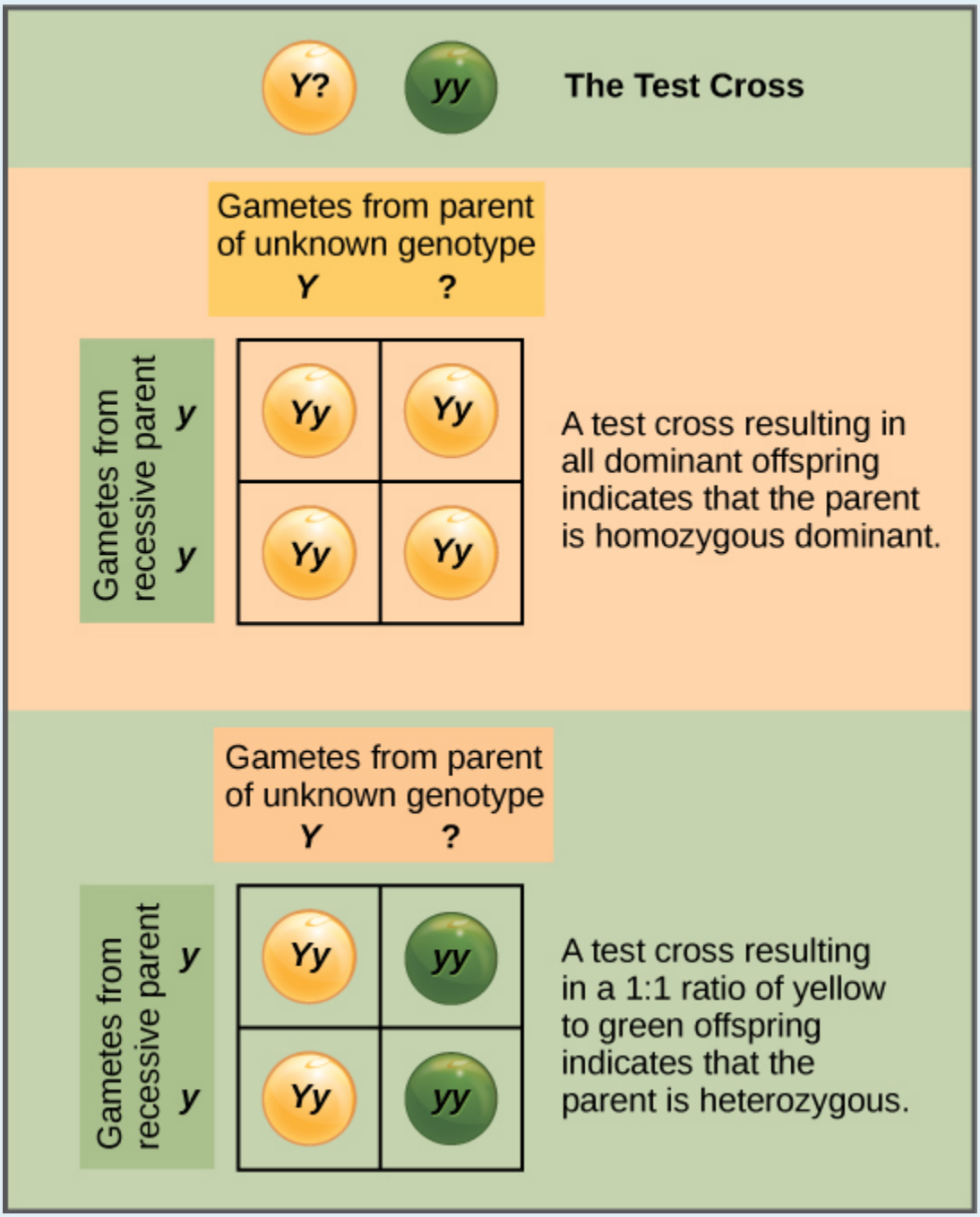

Figure 2. A test cross can be performed to determine whether an organism expressing a dominant trait is a homozygote or a heterozygote.

In pea plants, round peas (*R*) are dominant to wrinkled peas (*r*). You do a test cross between a pea plant with wrinkled peas (genotype *rr*) and a plant of unknown genotype that has round peas. You end up with three plants, all which have round peas. From this data, can you tell if the round pea parent plant is homozygous dominant or heterozygous? If the round pea parent plant is heterozygous, what is the probability that a random sample of 3 progeny peas will all be round?

Answer

You cannot be sure if the plant is homozygous or heterozygous as the data set is too small: by random chance, all three plants might have acquired only the dominant gene even if the recessive one is present. If the round pea parent is heterozygous, there is a one-eighth probability that a random sample of three progeny peas will all be round.

Many human diseases are genetically inherited. A healthy person in a family in which some members suffer from a recessive genetic disorder may want to know if he or she has the disease-causing gene and what risk exists of passing the disorder on to his or her offspring. Of course, doing a test cross in humans is unethical and impractical. Instead, geneticists use pedigree analysis to study the inheritance pattern of human genetic diseases (Figure 3).

Practice Question

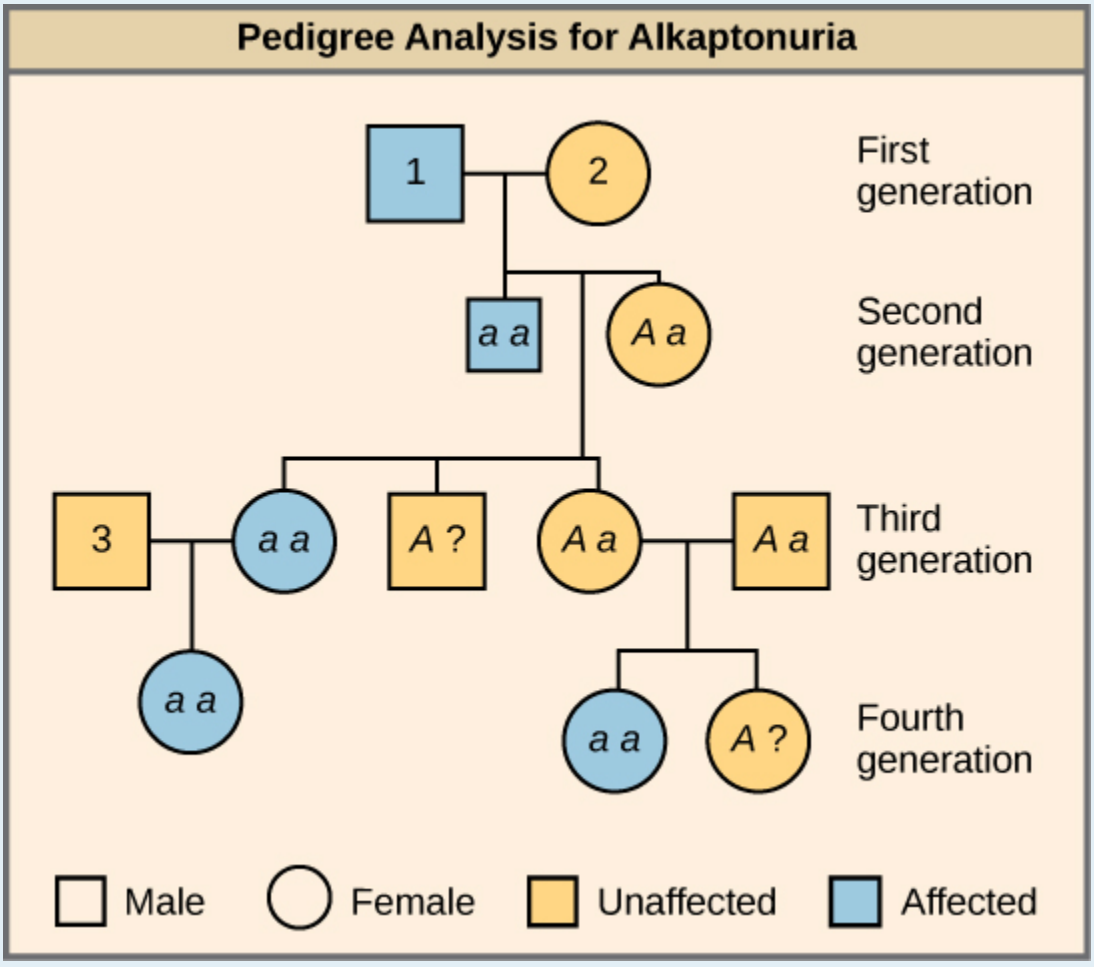

Figure 3. Pedigree Analysis for Alkaptonuria

Alkaptonuria is a recessive genetic disorder in which two amino acids, phenylalanine and tyrosine, are not properly metabolized. Affected individuals may have darkened skin and brown urine, and may suffer joint damage and other complications. In this pedigree, individuals with the disorder are indicated in blue and have the genotype aa. Unaffected individuals are indicated in yellow and have the genotype AA or Aa. Note that it is often possible to determine a person's genotype from the genotype of their offspring. For example, if neither

parent has the disorder but their child does, they must be heterozygous. Two individuals on the pedigree have an unaffected phenotype but unknown genotype. Because they do not have the disorder, they must have at least one normal allele, so their genotype gets the “A?” designation.

What are the genotypes of the individuals labeled 1, 2 and 3?

Answer

Individual 1 has the genotype *aa*. Individual 2 has the genotype *Aa*. Individual 3 has the genotype *Aa*.

In Summary: Characteristics and Traits

When true-breeding or homozygous individuals that differ for a certain trait are crossed, all of the offspring will be heterozygotes for that trait. If the traits are inherited as dominant and recessive, the F_1 offspring will all exhibit the same phenotype as the parent homozygous for the dominant trait. If these heterozygous offspring are self-crossed, the resulting F_2 offspring will be equally likely to inherit gametes carrying the dominant or recessive trait, giving rise to offspring of which one quarter are homozygous dominant, half are heterozygous, and one quarter are homozygous recessive. Because homozygous dominant and heterozygous individuals are phenotypically identical, the observed traits in the F_2 offspring will exhibit a ratio of three dominant to one recessive.

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LAWS OF INHERITANCE

Learning Outcomes

- Apply the law of segregation
- Apply the law of independent assortment

Mendel generalized the results of his pea-plant experiments into four postulates, some of which are sometimes called “laws,” that describe the basis of dominant and recessive inheritance in diploid organisms. As you have learned, more complex extensions of Mendelism exist that do not exhibit the same F_2 phenotypic ratios (3:1). Nevertheless, these laws summarize the basics of classical genetics.

Pairs of Unit Factors, or Genes

Mendel proposed first that paired unit factors of heredity were transmitted faithfully from generation to generation by the dissociation and reassociation of paired factors during gametogenesis and fertilization, respectively. After he crossed peas with contrasting traits and found that the recessive trait resurfaced in the F_2 generation, Mendel

deduced that hereditary factors must be inherited as discrete units. This finding contradicted the belief at that time that parental traits were blended in the offspring.

Alleles Can Be Dominant or Recessive

Mendel's **law of dominance** states that in a heterozygote, one trait will conceal the presence of another trait for the same characteristic. Rather than both alleles contributing to a phenotype, the dominant allele will be expressed exclusively. The recessive allele will remain "latent" but will be transmitted to offspring by the same manner in which the dominant allele is transmitted. The recessive trait will only be expressed by offspring that have two copies of this allele (Figure 1), and these offspring will breed true when self-crossed.

Since Mendel's experiments with pea plants, other researchers have found that the law of dominance does not always hold true. Instead, several different patterns of inheritance have been found to exist.

Figure 1. The child in the photo expresses albinism, a recessive trait.

Equal Segregation of Alleles

Observing that true-breeding pea plants with contrasting traits gave rise to F_1 generations that all expressed the dominant trait and F_2 generations that expressed the dominant and recessive traits in a 3:1 ratio, Mendel proposed the **law of segregation**. This law states that paired unit factors (genes) must segregate equally into gametes such that offspring have an equal likelihood of inheriting either factor. For the F_2 generation of a monohybrid cross, the following three possible combinations of genotypes could result: homozygous dominant, heterozygous, or homozygous recessive. Because heterozygotes could arise from two different pathways (receiving one dominant and one recessive allele from either parent), and because heterozygotes and homozygous dominant individuals are phenotypically identical, the law supports Mendel's observed 3:1 phenotypic ratio. The equal segregation of alleles is the reason we can apply the Punnett square to accurately predict the offspring of parents with known genotypes. The physical basis of Mendel's law of segregation is the first division of meiosis, in which the homologous chromosomes with their different versions of each gene are segregated into daughter nuclei. The role of the meiotic segregation of chromosomes in sexual reproduction was not understood by the scientific community during Mendel's lifetime.

Independent Assortment

Mendel's **law of independent assortment** states that genes do not influence each other with regard to the sorting of alleles into gametes, and every possible combination of alleles for every gene is equally likely to occur. The independent assortment of genes can be illustrated by the **dihybrid** cross, a cross between two true-breeding parents that express different traits for two characteristics. Consider the characteristics of seed color and seed texture for two pea plants, one that has green, wrinkled seeds (*yyrr*) and another that has yellow, round seeds (*YYRR*). Because each parent is homozygous, the law of segregation indicates that the gametes for the green/wrinkled plant all are *yr*, and the gametes for the yellow/round plant are all *YR*. Therefore, the F_1 generation of offspring all are *YyRr* (Figure 2).

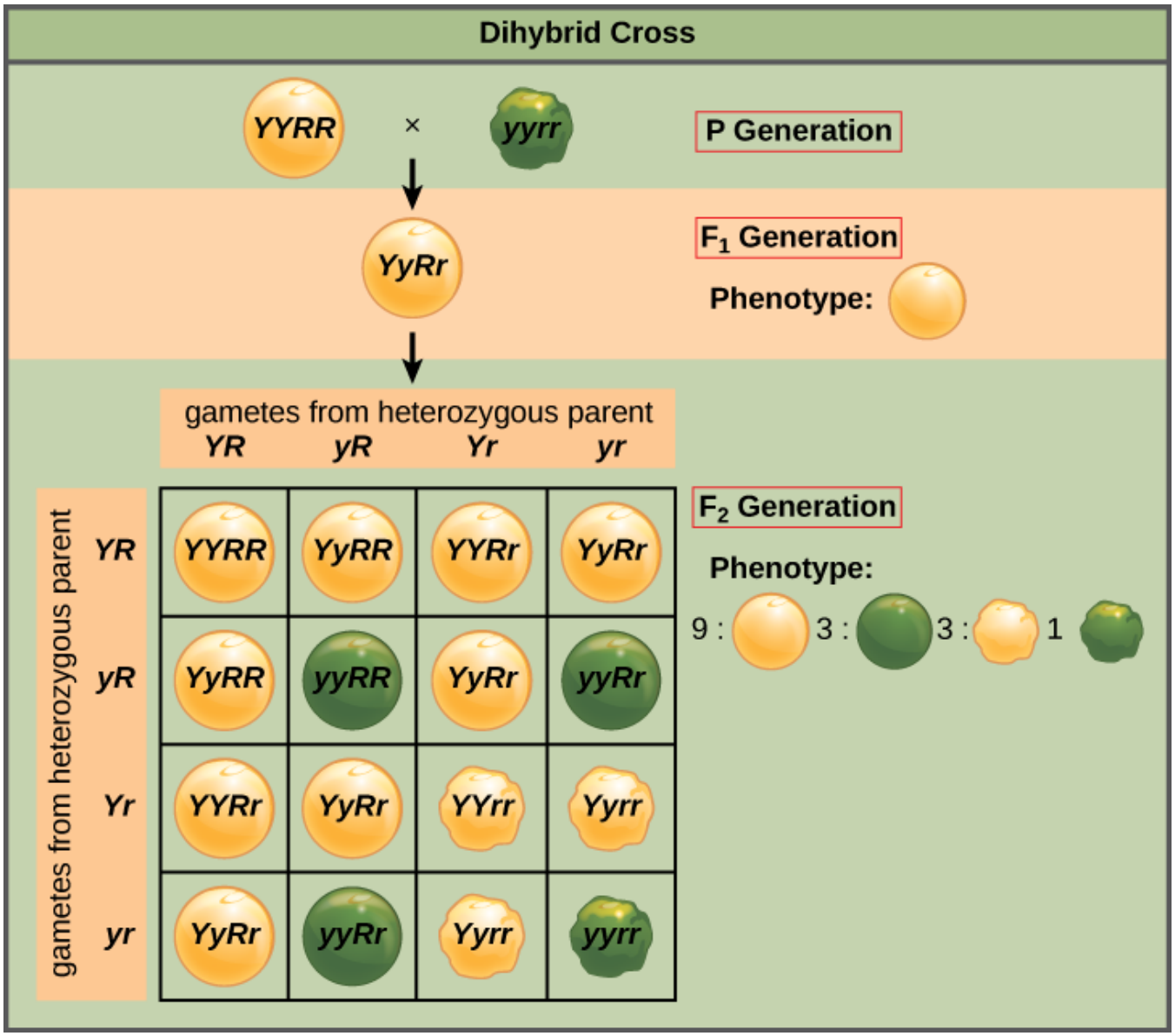

Figure 2. This dihybrid cross of pea plants involves the genes for seed color and texture.

Practice Question

In pea plants, purple flowers (P) are dominant to white flowers (p) and yellow peas (Y) are dominant to green peas (y). What are the possible genotypes and phenotypes for a cross between PpYY and ppYy pea plants? How many squares do you need to do a Punnett square analysis of this cross?

Answer

The possible genotypes are PpYY, PpYy, ppYY, and ppYy. The former two genotypes would result in plants with purple flowers and yellow peas, while the latter two genotypes would result in plants with white flowers with yellow peas, for a 1:1 ratio of each phenotype. You only need a 2 × 2 Punnett square (four squares total) to do this analysis because two of the alleles are homozygous.

For the F_2 generation, the law of segregation requires that each gamete receive either an *R* allele or an *r* allele along with either a *Y* allele or a *y* allele. The law of independent assortment states that a gamete into which an *r* allele sorted would be equally likely to contain either a *Y* allele or a *y* allele. Thus, there are four equally likely gametes that can be formed when the *YyRr* heterozygote is self-crossed, as follows: *YR*, *Yr*, *yR*, and *yr*. Arranging these gametes along the top and left of a 4 × 4 Punnett square (Figure 2) gives us 16 equally likely genotypic combinations. From these genotypes, we infer a phenotypic ratio of 9 round/yellow:3 round/green:3 wrinkled/yellow:1 wrinkled/green (Figure 2). These are the offspring ratios we would expect, assuming we performed the crosses with a large enough sample size.

The law of independent assortment also indicates that a cross between yellow, wrinkled (*YYrr*) and green, round (*yyRR*) parents would yield the same F_1 and F_2 offspring as in the *YYRR* x *yyrr* cross.

The physical basis for the law of independent assortment also lies in meiosis I, in which the different homologous pairs line up in random orientations. Each gamete can contain any combination of paternal and maternal chromosomes (and therefore the genes on them) because the orientation of tetrads on the metaphase plane is random.

In pea plants, purple flowers (P) are dominant to white flowers (p) and yellow peas (Y) are dominant to green peas (y). What are the possible genotypes and phenotypes for a cross between PpYY and ppYy pea plants? How many squares do you need to do a Punnett square analysis of this cross? The possible genotypes are PpYY, PpYy, ppYY, and ppYy. The former two genotypes would result in plants with purple flowers and yellow peas, while the latter two genotypes would result in plants with white flowers with yellow peas, for a 1:1 ratio of each phenotype. You only need a 2 × 2 Punnett square (four squares total) to do this analysis because two of the alleles are homozygous.

Linked Genes Violate the Law of Independent Assortment

Although all of Mendel's pea characteristics behaved according to the law of independent assortment, we now know that some allele combinations are not inherited independently of each other. Genes that are located on separate non-homologous chromosomes will always sort independently. However, each chromosome contains hundreds or thousands of genes, organized linearly on chromosomes like beads on a string. The segregation of alleles into gametes can be influenced by linkage, in which genes that are located physically close to each other on the same chromosome are more likely to be inherited as a pair. However, because of the process of recombination, or "crossover," it is possible for two genes on the same chromosome to behave independently, or as if they are not linked. To understand this, let's consider the biological basis of gene linkage and recombination.

Homologous chromosomes possess the same genes in the same linear order. The alleles may differ on homologous chromosome pairs, but the genes to which they correspond do not. In preparation for the first division of meiosis, homologous chromosomes replicate and synapse. Like genes on the homologs align with each other. At this stage, segments of homologous chromosomes exchange linear segments of genetic material (Figure 3). This process is called recombination, or crossover, and it is a common genetic process. Because the genes are aligned during recombination, the gene order is not altered. Instead, the result of recombination is that maternal and paternal alleles are combined onto the same chromosome. Across a given chromosome, several recombination events may occur, causing extensive shuffling of alleles.

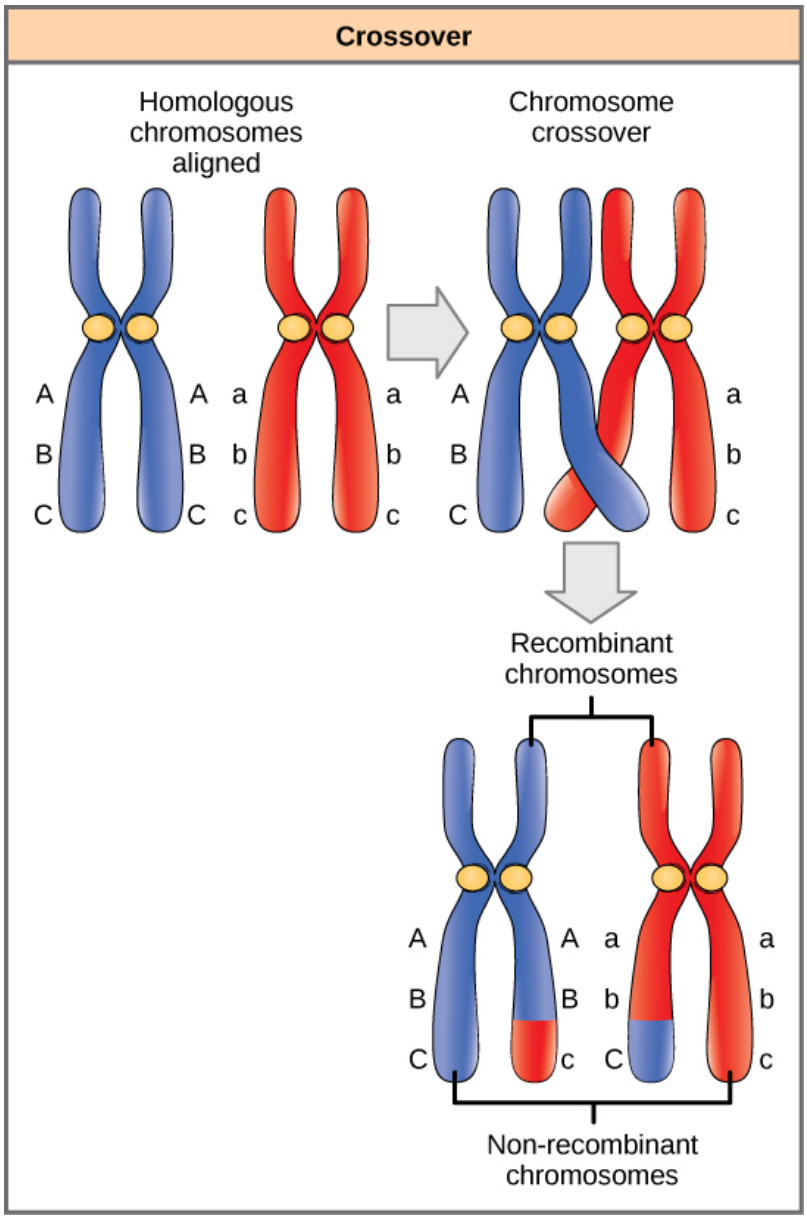

Figure 3. The process of crossover, or recombination, occurs when two homologous chromosomes align during meiosis and exchange a segment of genetic material. Here, the alleles for gene C were exchanged. The result is two recombinant and two non-recombinant chromosomes.

When two genes are located in close proximity on the same chromosome, they are considered **linked**, and their alleles tend to be transmitted through meiosis together. To exemplify this, imagine a dihybrid cross involving flower color and plant height in which the genes are next to each other on the chromosome. If one homologous chromosome has alleles for tall plants and red flowers, and the other chromosome has genes for short plants and yellow flowers, then when the gametes are formed, the tall and red alleles will go together into a gamete and the short and yellow alleles will go into other gametes. These are called the parental genotypes because they have been inherited intact from the parents of the individual producing gametes. But unlike if the genes were on different chromosomes, there will be no gametes with tall and yellow alleles and no gametes with short and red alleles. If you create the Punnett square with these gametes, you will see that the classical Mendelian prediction of a 9:3:3:1 outcome of a dihybrid cross would not apply.

As the distance between two genes increases, the probability of one or more crossovers between them increases, and the genes behave more like they are on separate chromosomes. Geneticists have used the proportion of recombinant gametes (the ones not like the parents) as a measure of how far apart genes are on a chromosome. Using this information, they have constructed elaborate maps of genes on chromosomes for well-studied organisms, including humans.

Testing the Hypothesis of Independent Assortment

To better appreciate the amount of labor and ingenuity that went into Mendel's experiments, proceed through one of Mendel's dihybrid crosses.

Question: What will be the offspring of a dihybrid cross?

Background: Consider that pea plants mature in one growing season, and you have access to a large garden in which you can cultivate thousands of pea plants. There are several true-breeding plants with the following pairs of traits: tall plants with inflated pods, and dwarf plants with constricted pods. Before the plants have matured, you remove the pollen-producing organs from the tall/inflated plants in your crosses to prevent self-fertilization. Upon plant maturation, the plants are manually crossed by transferring pollen from the dwarf/constricted plants to the stigmata of the tall/inflated plants.

Hypothesis: Both trait pairs will sort independently according to Mendelian laws. When the true-breeding parents are crossed, all of the F_1 offspring are tall and have inflated pods, which indicates that the tall and inflated traits are dominant over the dwarf and constricted traits, respectively. A self-cross of the F_1 heterozygotes results in 2,000 F_2 progeny.

Test the hypothesis: Because each trait pair sorts independently, the ratios of tall:dwarf and inflated:constricted are each expected to be 3:1. The tall/dwarf trait pair is called *T/t*, and the inflated/constricted trait pair is designated *I/i*. Each member of the F_1 generation therefore has a genotype of *TtIi*. Construct a grid analogous to Figure 4, in which you cross two *TtIi* individuals. Each individual can donate four combinations of two traits: *TI*, *Ti*, *tI*, or *ti*, meaning that there are 16 possibilities of offspring genotypes. Because the *T* and *I* alleles are dominant, any individual having one or two of those alleles will express the tall or inflated phenotypes, respectively, regardless if they also have a *t* or *i* allele. Only individuals that are *tt* or *ii* will express the dwarf and constricted alleles, respectively. As shown in Figure 4, you predict that you will observe the following offspring proportions: tall/inflated : tall/constricted : dwarf/inflated : dwarf/constricted in a 9:3:3:1 ratio. Notice from the grid that when considering the tall/dwarf and inflated/constricted trait pairs in isolation, they are each inherited in 3:1 ratios.

Table 1 shows all possible combinations of offspring resulting from a dihybrid cross of pea plants that are heterozygous for the tall/dwarf and inflated/constricted alleles.

Table 1. Dihybrid Cross

		TtIi			
		TI	*Ti*	*tI*	*ti*
TtIi	*TI*	TTII	TTIi	TtII	TtIi
	Ti	TTIi	TTii	TtIi	Ttii
	tI	TtII	TtIi	ttII	ttIi
	ti	TtIi	Ttii	ttIi	ttii

Test the hypothesis: You cross the dwarf and tall plants and then self-cross the offspring. For best results, this is repeated with hundreds or even thousands of pea plants. What special precautions should be taken in the crosses and in growing the plants?

Analyze your data: You observe the following plant phenotypes in the F_2 generation: 2706 tall/inflated, 930 tall/constricted, 888 dwarf/inflated, and 300 dwarf/constricted. Reduce these findings to a ratio and determine if they are consistent with Mendelian laws.

Form a conclusion: Were the results close to the expected 9:3:3:1 phenotypic ratio? Do the results support the prediction? What might be observed if far fewer plants were used, given that alleles segregate randomly into gametes? Try to imagine growing that many pea plants, and consider the potential for experimental error. For instance, what would happen if it was extremely windy one day?

In Summary: Laws of Inheritance

Mendel postulated that genes (characteristics) are inherited as pairs of alleles (traits) that behave in a dominant and recessive pattern. Alleles segregate into gametes such that each gamete is equally likely to receive either one of the two alleles present in a diploid individual. In addition, genes are assorted into gametes independently of one another. That is, alleles are generally not more likely to segregate into a gamete with a particular allele of another gene. A dihybrid cross demonstrates independent assortment when the genes in question are on different chromosomes or distant from each other on the same chromosome. For crosses involving more than two genes, use the forked line or probability methods to predict offspring genotypes and phenotypes rather than a Punnett square.

Although chromosomes sort independently into gametes during meiosis, Mendel's law of independent assortment refers to genes, not chromosomes, and a single chromosome may carry more than 1,000 genes. When genes are located in close proximity on the same chromosome, their alleles tend to be inherited together. This results in offspring ratios that violate Mendel's law of independent assortment. However, recombination serves to exchange genetic material on homologous chromosomes such that maternal and paternal alleles may be recombined on the same chromosome. This is why alleles on a given chromosome are not always inherited together. Recombination is a random event occurring anywhere on a chromosome. Therefore, genes that are far apart on the same chromosome are likely to still assort independently because of recombination events that occurred in the intervening chromosomal space.

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HEREDITY

Learning Outcomes

- Describe Mendel's study of garden peas and hereditary
- Understand how the inheritance of a genotype generates a phenotype
- Apply the law of segregation
- Apply the law of independent assortment

Watch this video for a nice summary of Mendel's contribution to the field of genetics and how a genotype leads to a phenotype.

Watch this video online: https://youtu.be/CBezq1fFUEA

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INTRODUCTION TO BEYOND DOMINANCE AND RECESSIVENESS

What you'll learn to do: Explain complications to the phenotypic expression of genotype, including mutations

Mendel's experiments with pea plants suggested the following:

1. Two "units" or alleles exist for every gene.
2. Alleles maintain their integrity in each generation (no blending).
3. In the presence of the dominant allele, the recessive allele is hidden and makes no contribution to the phenotype.

Therefore, recessive alleles can be "carried" and not expressed by individuals. Such heterozygous individuals are sometimes referred to as "carriers." Further genetic studies in other plants and animals have shown that much more complexity exists, but that the fundamental principles of Mendelian genetics still hold true. In the sections to follow, we consider some of the extensions of Mendelism. If Mendel had chosen an experimental system that exhibited these genetic complexities, it's possible that he would not have understood what his results meant.

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NON-MENDELIAN INHERITANCE

Learning Outcomes

- Explain how a trait with incomplete dominance will appear in a population
- Explain how a trait with codominant inheritance will appear in a population
- Explain how a trait with sex-linkage will appear in a population

Incomplete Dominance

Mendel's results, that traits are inherited as dominant and recessive pairs, contradicted the view at that time that offspring exhibited a blend of their parents' traits. However, the heterozygote phenotype occasionally does appear to be intermediate between the two parents.

For example, in the snapdragon, *Antirrhinum majus* (Figure 1), a cross between a homozygous parent with white flowers (C^WC^W) and a homozygous parent with red flowers (C^RC^R) will produce offspring with pink flowers (C^RC^W). (Note that different genotypic abbreviations are used for Mendelian extensions to distinguish these patterns from simple dominance and recessiveness.)

This pattern of inheritance is described as **incomplete dominance**, denoting the expression of two contrasting alleles such that the individual displays an intermediate phenotype. The allele for red flowers is incompletely dominant over the allele for white flowers. However, the results of a heterozygote self-cross can still be predicted, just as with Mendelian dominant and recessive crosses. In this case, the genotypic ratio would be 1 C^RC^R:2 C^RC^W:1 C^WC^W, and the phenotypic ratio would be 1:2:1 for red:pink:white.

Figure 1. These pink flowers of a heterozygote snapdragon result from incomplete dominance. (credit: "storebukkebruse"/Flickr)

Codominant Inheritance

A variation on incomplete dominance is **codominance**, in which both alleles for the same characteristic are simultaneously expressed in the heterozygote.

Codominance can also be seen in human blood types: the AB blood type is a result of both the I^A allele and the I^B allele being codominant. (An A blood type would only have the I^A allele, and a B blood type would only have I^B allele.)

The roan coat color in horses is also an example of codominance. A "red" roan results from the mating of a chestnut parent and a white parent (Figure 2). We know this is codominance because individual hairs are either chestnut or they are white, leading to the red roan overall appearance.

Figure 2. Red Roan Horse

Practice Question

So what's the difference between incomplete dominance and codominant inheritance? While they are very similar, the key difference is this: in incomplete dominance, the two traits are blended together, whereas in codominance, both traits are expressed.

We've already discussed incomplete dominance in flowers (Figure 1). What do you think a flower would look like if the red and white phenotypes were codominant instead?

Answer

The flower would have both red and white petals, like this Rhododendron:

Codominance is shown in this rhododendron.

Sex-Linked Traits

In humans, as well as in many other animals and some plants, the sex of the individual is determined by sex chromosomes. The sex chromosomes are one pair of non-homologous chromosomes. Until now, we have only considered inheritance patterns among non-sex chromosomes, or **autosomes**. In addition to 22 homologous pairs of autosomes, human females have a homologous pair of X chromosomes, whereas human males have an XY chromosome pair. Although the Y chromosome contains a small region of similarity to the X chromosome so that they can pair during meiosis, the Y chromosome is much shorter and contains many fewer genes. When a gene being examined is present on the X chromosome, but not on the Y chromosome, it is said to be **X-linked**.

Eye color in *Drosophila* was one of the first X-linked traits to be identified. Thomas Hunt Morgan mapped this trait to the X chromosome in 1910. Like humans, *Drosophila* males have an XY chromosome pair, and females are XX. In flies, the wild-type eye color is red (X^W) and it is dominant to white eye color (X^w) (Figure 3). Because of the location of the eye-color gene, reciprocal crosses do not produce the same offspring ratios. Males are said to be **hemizygous**, because they have only one allele for any X-linked characteristic. Hemizygosity makes the descriptions of dominance and recessiveness irrelevant for XY males. *Drosophila* males lack a second allele copy on the Y chromosome; that is, their genotype can only be X^WY or X^wY. In contrast, females have two allele copies of this gene and can be X^WX^W, X^WX^w, or X^wX^w.

Figure 3. In Drosophila, the gene for eye color is located on the X chromosome. Clockwise from top left are brown, cinnabar, sepia, vermilion, white, and red. Red eye color is wild-type and is dominant to white eye color.

Practice Question

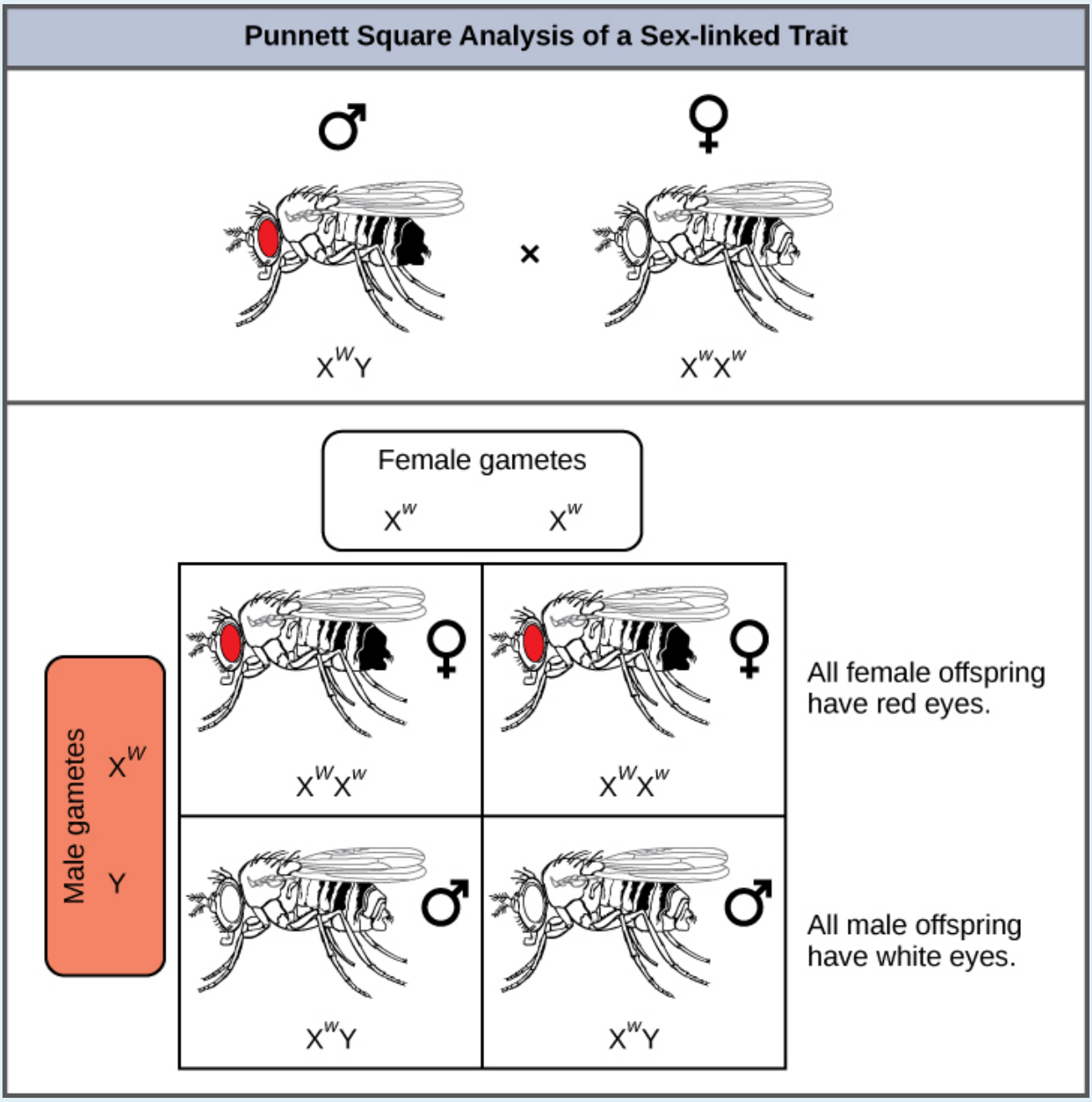

Figure 4. Punnett square analysis is used to determine the ratio of offspring from a cross between a red-eyed male fruit fly and a white-eyed female fruit fly.

What ratio of offspring would result from a cross between a white-eyed male and a female that is heterozygous for red eye color?

Answer

Half of the female offspring would be heterozygous (X^WX^w) with red eyes, and half would be homozygous recessive (X^wX^w) with white eyes. Half of the male offspring would be hemizygous dominant (X^WY) with red yes, and half would be hemizygous recessive (X^wY) with white eyes.

Discoveries in fruit fly genetics can be applied to human genetics. When a female parent is homozygous for a recessive X-linked trait, she will pass the trait on to 100 percent of her offspring. Her male offspring are, therefore, destined to express the trait, as they will inherit their father's Y chromosome. In humans, the alleles for certain conditions (some forms of **color blindness, hemophilia, and muscular dystrophy**) are X-linked. Females who are heterozygous for these diseases are said to be carriers and may not exhibit any phenotypic effects. These females will pass the disease to half of their sons and will pass carrier status to half of their daughters; therefore, recessive X-linked traits appear more frequently in males than females.

In some groups of organisms with sex chromosomes, the gender with the non-homologous sex chromosomes is the female rather than the male. This is the case for all birds. In this case, sex-linked traits will be more likely to appear in the female, in which they are hemizygous.

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NON-MENDELIAN PUNNETT SQUARES

Learning Outcomes

- Explain how a trait with incomplete dominance will appear in a population
- Explain how a trait with codominant inheritance will appear in a population
- Explain how a trait with sex-linkage will appear in a population

This practice activity will help you remember the difference between types of non-Mendelian inheritance and remember just how they work.

Click here for a text-only version of the activity.

Video Review

Watch this video for a summary of the three "special" cases of non-Mendelian inheritance you just practiced.
Watch this video online: https://youtu.be/fQvER3MyI2c

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MULTIPLE ALLELES

Learning Outcomes

Explain how mutli-allele inheritance will impact a trait within in a population

Mendel implied that only two alleles, one dominant and one recessive, could exist for a given gene. We now know that this is an oversimplification. Although individual humans (and all diploid organisms) can only have two alleles for a given gene, multiple alleles may exist at the population level such that many combinations of two alleles are observed. Note that when many alleles exist for the same gene, the convention is to denote the most common phenotype or genotype among wild animals as the **wild type** (often abbreviated "+"); this is considered the standard or norm. All other phenotypes or genotypes are considered **variants** of this standard, meaning that they deviate from the wild type. The variant may be recessive or dominant to the wild-type allele.

ABO blood grouping is an example of multiple alleles.

Multiple Alleles (ABO Blood Types) and Punnett Squares

Watch this video online: https://youtu.be/9O5JQqlngFY

Multiple Alleles Confer Drug Resistance in the Malaria Parasite

Malaria is a parasitic disease in humans that is transmitted by infected female mosquitoes, including *Anopheles gambiae* (Figure 1a), and is characterized by cyclic high fevers, chills, flu-like symptoms, and severe anemia. *Plasmodium falciparum* and *P. vivax* are the most common causative agents of malaria, and *P. falciparum* is the most deadly (Figure 1b). When promptly and correctly treated, *P. falciparum*malaria has a mortality rate of 0.1 percent. However, in some parts of the world, the parasite has evolved resistance to commonly used malaria treatments, so the most effective malarial treatments can vary by geographic region.

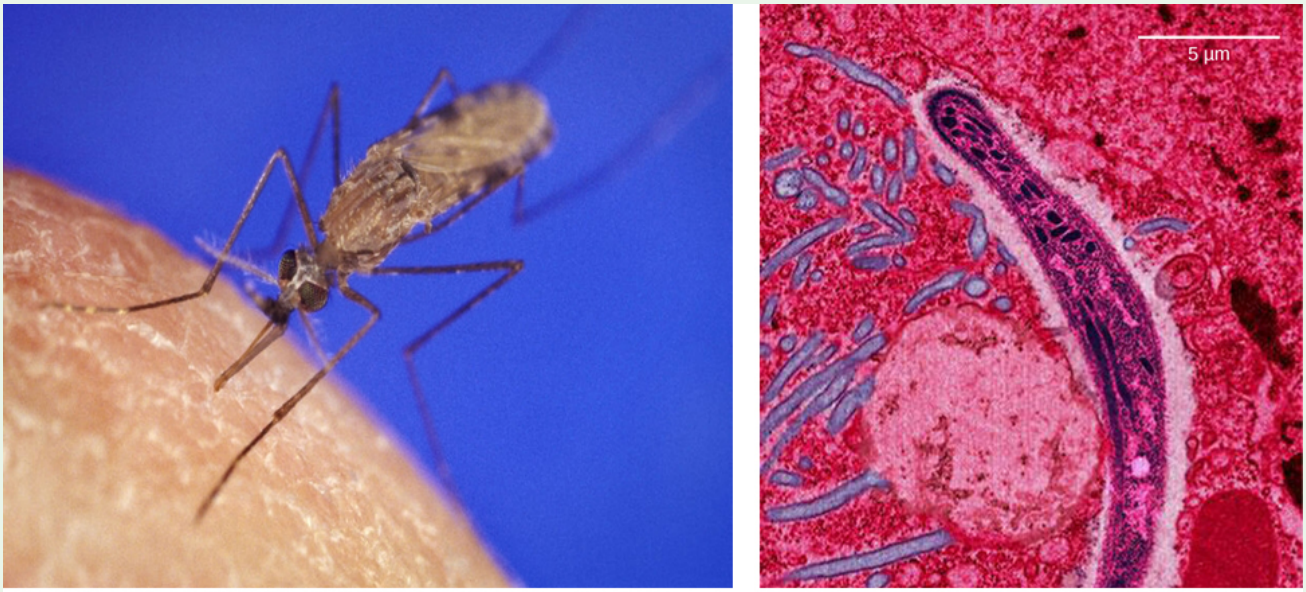

Figure 1. The (a) Anopheles gambiae, or African malaria mosquito, acts as a vector in the transmission to humans of the malaria-causing parasite (b) Plasmodium falciparum, here visualized using false-color transmission electron microscopy. (credit a: James D. Gathany; credit b: Ute Frevert; false color by Margaret Shear; scale-bar data from Matt Russell)

In Southeast Asia, Africa, and South America, *P. falciparum* has developed resistance to the anti-malarial drugs chloroquine, mefloquine, and sulfadoxine-pyrimethamine. *P. falciparum*, which is haploid during the life stage in which it is infectious to humans, has evolved multiple drug-resistant mutant alleles of the *dhps* gene. Varying degrees of sulfadoxine resistance are associated with each of these alleles. Being haploid, *P. falciparum* needs only one drug-resistant allele to express this trait.

In Southeast Asia, different sulfadoxine-resistant alleles of the *dhps* gene are localized to different geographic regions. This is a common evolutionary phenomenon that occurs because drug-resistant mutants arise in a population and interbreed with other *P. falciparum* isolates in close proximity. Sulfadoxine-resistant parasites cause considerable human hardship in regions where this drug is widely used as an over-the-counter malaria remedy. As is common with pathogens that multiply to large numbers within an infection cycle, *P. falciparum* evolves relatively rapidly (over a decade or so) in response to the selective pressure of commonly used anti-malarial drugs. For this reason, scientists must constantly work to develop new drugs or drug combinations to combat the worldwide malaria burden. (Note: Sumiti Vinayak, et al., "Origin and Evolution of Sulfadoxine Resistant *Plasmodium falciparum*," *Public Library of Science Pathogens* 6, no. 3 (2010): e1000830, doi:10.1371/journal.ppat.1000830.)

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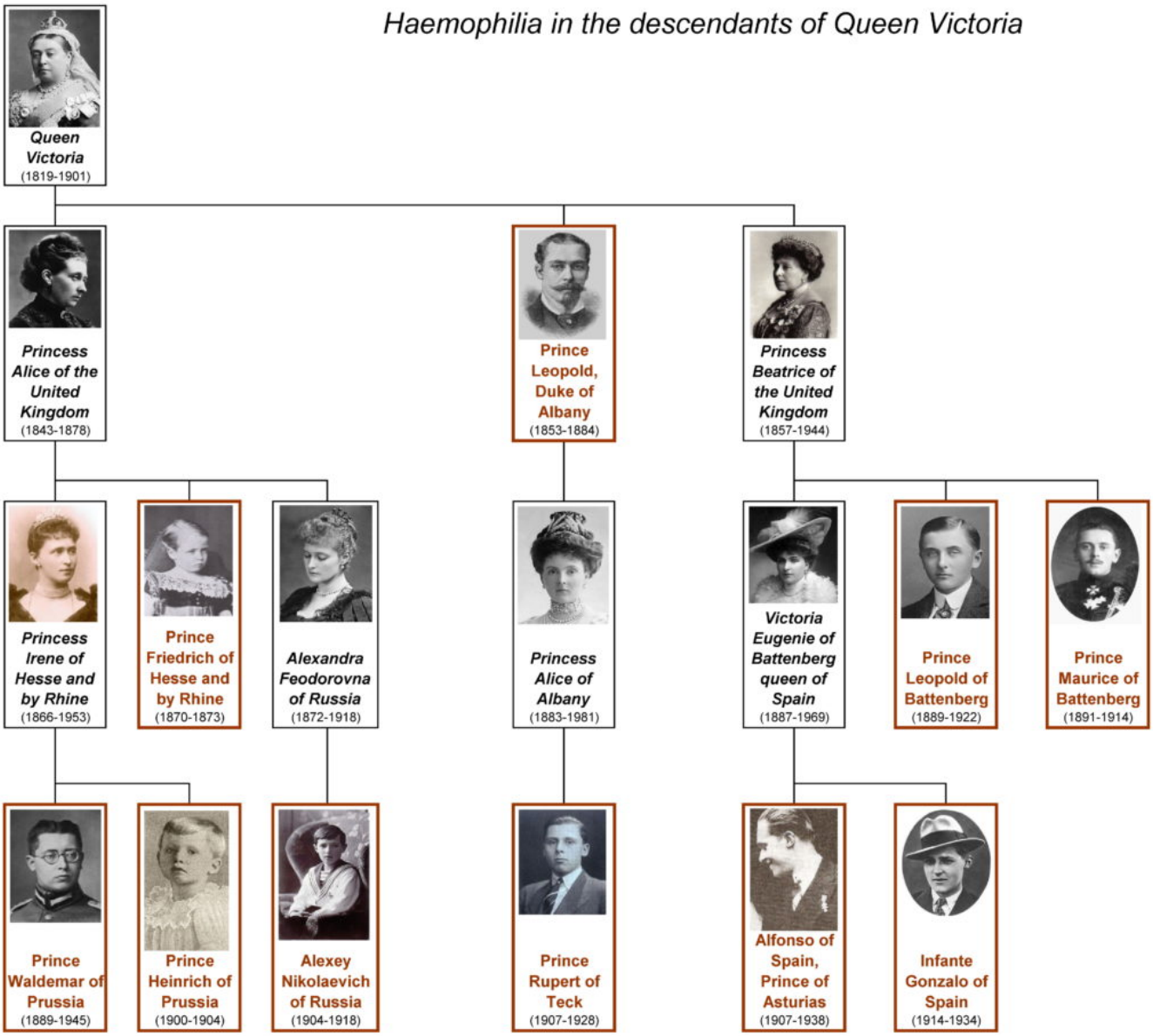
INTRODUCTION TO HEREDITY AND DISEASE

What you'll learn to do: Explain the conventions of a family pedigree and predict whether a disease will be passed through a family in one of three modes

Over the years, we've seen that some diseases are hereditary. In this outcome, we'll learn just how some diseases can be passed through a family line.

Family pedigrees and diseases can also be used to solve "mysteries," such as the case of the Tsar of Russia and the missing Princess Anastasia. In this case, haemophilia within European royalty figured prominently in identifying the bodies of the murdered Russian royals (but not Princess Anastasia).



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PEDIGREES AND DISEASE

Learning Outcomes

Understand why geneticists use pedigrees to track human diseases

Health care professionals have known for a long time that common diseases—like heart disease, cancer, and diabetes—and even rare diseases—like hemophilia, cystic fibrosis, and sickle cell anemia—can run in families. If one generation of a family has high blood pressure, it is not unusual for the next generation to have similarly high blood pressure. Therefore, family history can be a powerful screening tool and has often been referred to as the best “genetic test.”

Both common diseases and rare diseases can run in families. Therefore, family history can be a powerful screening tool. Family history should be updated on each visit and patients should be made aware of its significance to their health.

Importance of Family History

Family history holds important information about an individual’s past and future life. Family history can be used as a diagnostic tool and help guide decisions about genetic testing for the patient and at-risk family members. If a family is affected by a disease, an accurate family history will be important to establish a pattern of transmission. In addition, a family history can even help to exclude genetic diseases, particularly for common diseases in which lifestyle and environment play strong roles. Lastly, a family history can identify potential health problems that an individual may be at increased risk for in the future. Early identification of increased risk can allow the individual and health professional to take steps to reduce risk by implementing lifestyle changes and increasing disease surveillance.

Notwithstanding the importance of family history to help define occurrence of a genetic disorder within a family, it should be noted that some genetic diseases are caused by spontaneous mutations, such as for single gene disorders like Duchenne muscular dystrophy and hemophilia A, as well as for most cases of Down syndrome, chromosomal deletion syndromes, and other chromosomal disorders. Therefore, a genetic disorder cannot be ruled out in the absence of a family history.

How to Take a Family Medical History

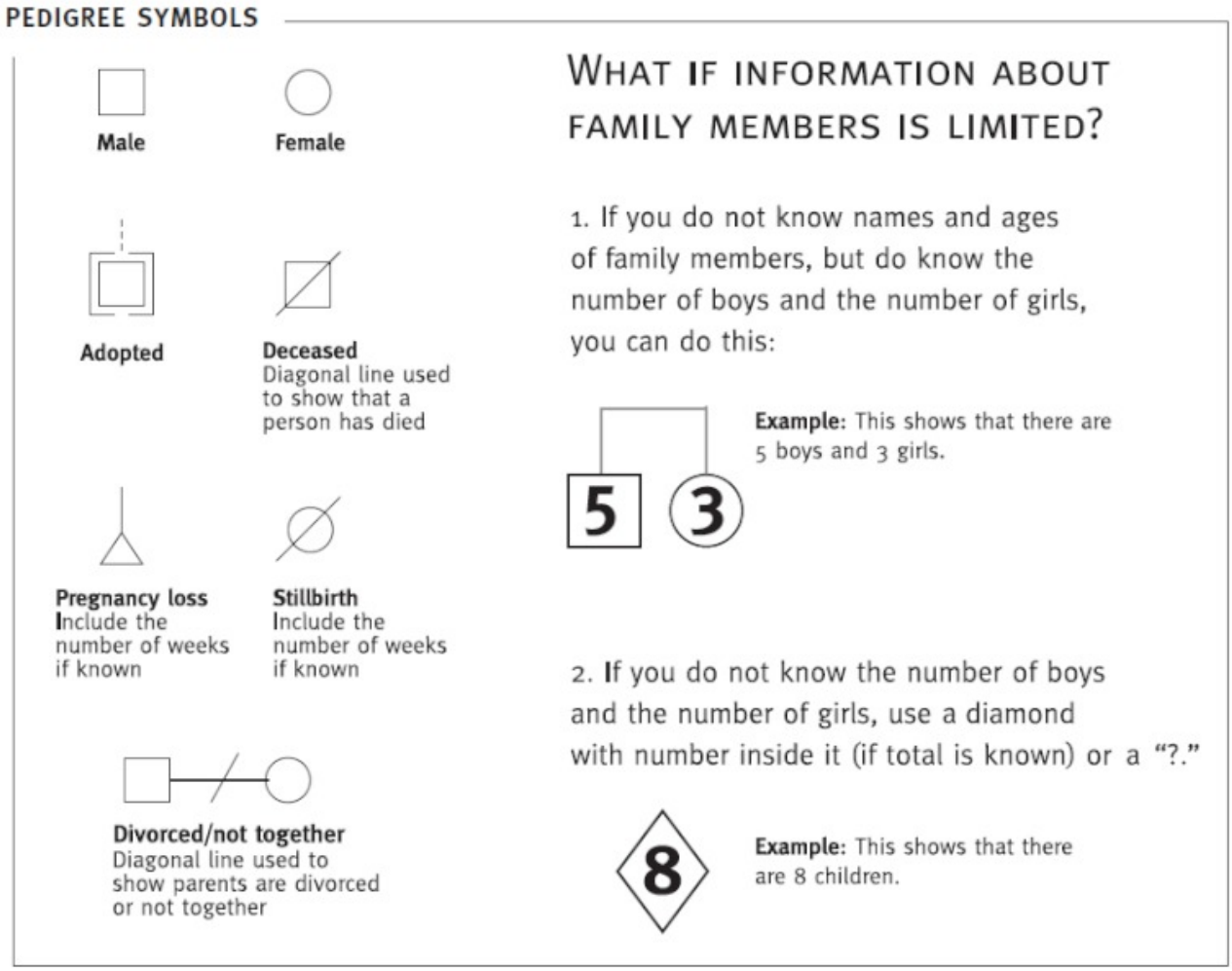
A basic family history should include three generations. To begin taking a family history, start by asking the patient about his/her health history and then ask about siblings and parents. Questions should include:

1. General information such as names and birthdates
2. Family’s origin or racial/ethnic background
3. Health status
4. Age at death and cause of death of each family member
5. Pregnancy outcomes of the patient and genetically-related relatives It may be easier to list all the members of the nuclear family first and then go back and ask about the health status of each one. After

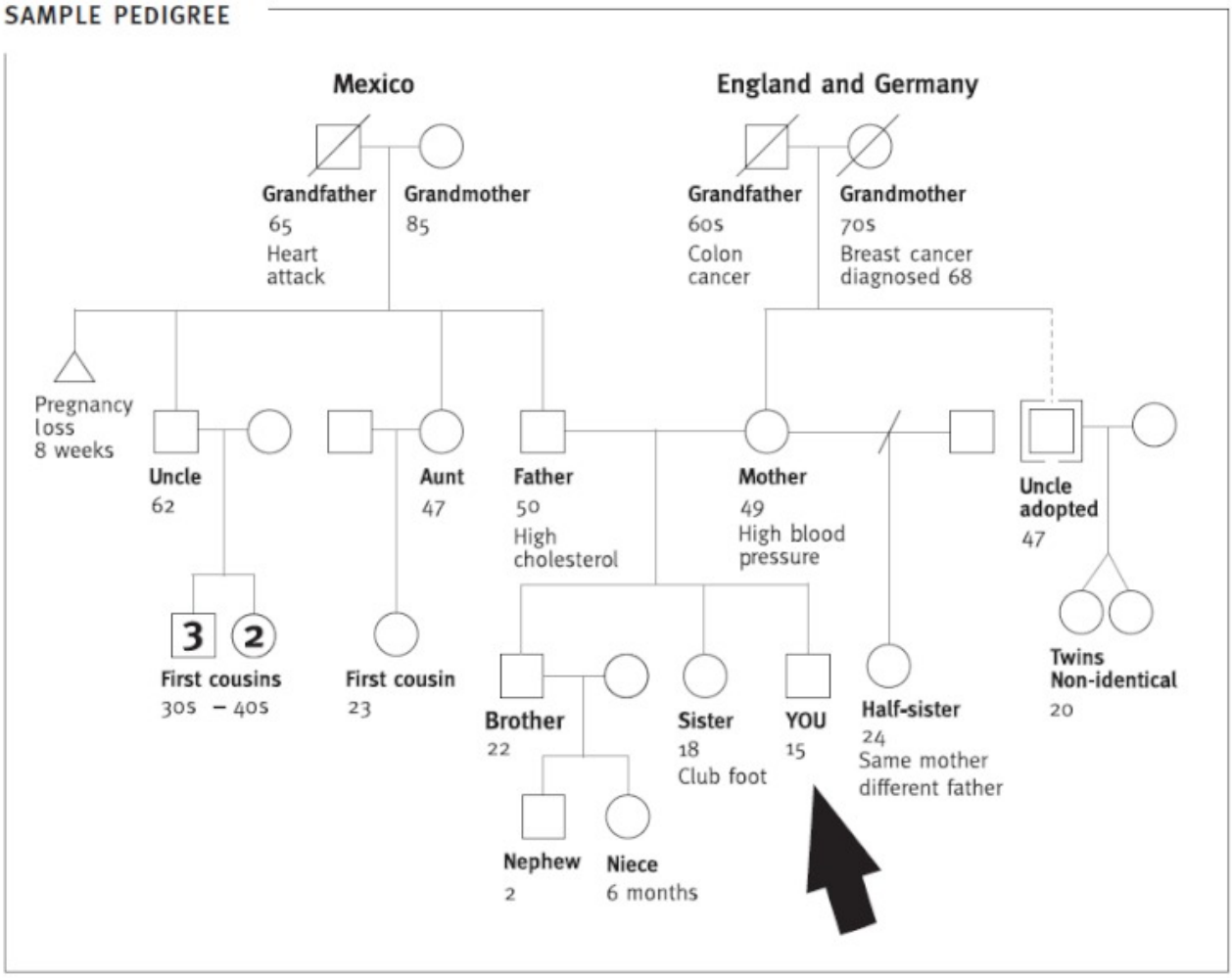
you have taken the family history of the patient's closest relatives, go back one generation at a time and ask about aunts, uncles, grandparents, and first cousins.

Pedigrees

One way to record a family history is by drawing a family tree called a "**pedigree**." A pedigree represents family members and relationships using standardized symbols (see below). As patients relate information to you about their family history, a pedigree can be drawn much quicker than recording the information in writing and allows patterns of disease to emerge as the pedigree is drawn. Since the family history is continually changing, the pedigree can be easily updated on future visits. Patients should be encouraged to record information and update their family history regularly.



The sample pedigree below contains information such as age or date of birth (and, for all deceased family members, age at death and cause of death), major medical problems with age of onset, birth defects, learning problems and mental retardation, and vision loss/hearing loss at a young age. For family members with known medical problems, ask whether they smoke, what their diet and exercise habits are if known, and if they are overweight.



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GENETIC DISORDER AND PEDIGREES

Learning Outcomes

- Understand why geneticists use pedigrees to track human diseases
- Understand how a pedigree analysis is used to identify the inheritance pattern of a genetic disorder

Watch this video to understand the basics of conducting a pedigree analysis of a human genetic disorder. At the end, you should feel comfortable with the basic steps used to conduct a pedigree investigation.

Watch this video online: https://youtu.be/z0cwhooDZ0M

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POLYGENIC INHERITANCE AND ENVIRONMENTAL EFFECTS

Learning Outcomes

- Describe polygenic inheritance and how to recognize it
- Describe continuous variation and how to recognize it

How is Height Inherited?

Many heritable human characteristics don't seem to follow Mendelian rules in their inheritance patterns. For example, consider human height. Unlike a simple Mendelian characteristic, human height displays:

- **Continuous variation.** Unlike Mendel's pea plants, humans don't come in two clear-cut "tall" and "short" varieties. In fact, they don't even come in four heights, or eight, or sixteen. Instead, it's possible to get humans of many different heights, and height can vary in increments of inches or fractions of inches. As an example, consider the bell curve-shaped graph in Figure 1, which shows the heights of a group of male high school seniors.
- **A complex inheritance pattern.** If you've paid attention to the heights of your friends and family, you may have noticed that many different patterns of inheritance are possible. Tall parents can have a short child, short parents can have a tall child, and two parents of different heights may or may not have a child of intermediate height. In addition, siblings with the same two parents may have a range of heights, ones that don't fall into clear, distinct categories. Simple models involving one or two genes can't accurately predict all of these inheritance patterns.

Some human characteristics, such as height, eye color, and hair color, don't come in just a few distinct forms. Instead, they vary in small gradations, forming a spectrum or continuum of possible phenotypes.

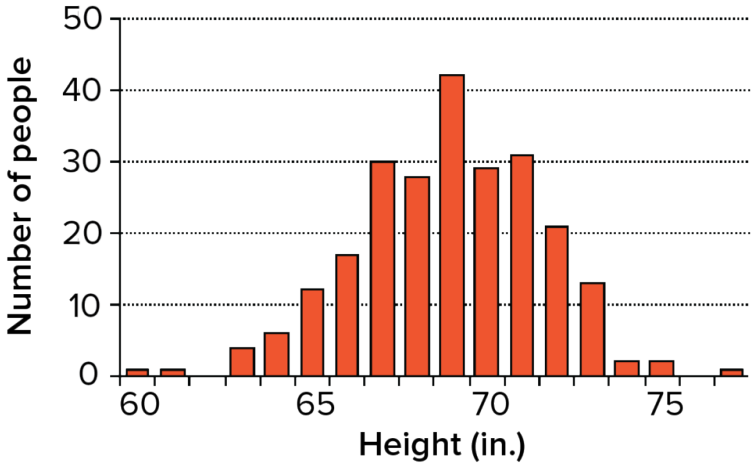

Figure 1. Heights of male high school seniors. Image modified from "Continuous variation: Quantitative traits," by J. W. Kimball (CC BY 3.0).

How, then, is height inherited? Height and other similar features are controlled not just by one gene, but rather, by multiple (often many) genes that each make a small contribution to the overall outcome. This inheritance pattern is called **polygenic inheritance** (*poly–* = many). For instance, a recent study found over 400 genes linked to variation in height (Note: Wood, A. R., Esko, T., Yang, J., Vedantam, S., Pers, T. H., Gustafsson, S., ... Frayling, T. M. (2014). Defining the role of common variation in the genomic and biological architecture of adult human height. *Nature Genetics*, *46*, 1173–1186. http://dx.doi.org/10.1038/ng.3097.). When there are large numbers of genes involved, it becomes hard to distinguish the effect of each individual gene, and even harder to see that gene variants (alleles) are inherited according to Mendelian rules. In a further complication, height doesn't just depend on genetics: it also depends a lot on environmental factors, such as a child's overall health and the type of nutrition he or she receives while growing up.

PRactice Questions

We've learned about polygenic inheritance and continuous variation. Just what is the difference between these two types of inheritance?

Answer

Polygenic traits are traits that rely on multiple genes. *Continuous variation* describes traits whose phenotypes occur on a continuum, rather than having a limited number of possible phenotypes. Traits with continuous variation are often also polygenic traits, but not always, and not all polygenic traits have continuous variation.

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INTRODUCTION TO GENETICS AND THE ENVIRONMENT

What you'll learn to do: Discuss the role environment plays on phenotypes

In recent years, scientists have begun to research how our environment can impact our phenotypes: most common diseases are a result of both your genes and your environment. Your environment can include personal choices, such as what foods you eat and how much you exercise, and external factors, such as stress, clean water, and air quality. Most diseases, especially common diseases, are a combination of your genetic risk and your environment. Only a small number of diseases are a result of just a single mutation in a gene. Examples of these single-gene disorders are Huntington disease and Tay Sachs.

It is becoming difficult to group diseases into either purely "genetic" or "environmental" because most diseases are a little bit of both. For example, emphysema can be the result of both smoking and a disorder called alpha-1-AT deficiency.

In this outcome, we'll learn about a few different ways our environment can impact us.

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EFFECT OF THE ENVIRONMENT

Learning Outcomes

Identify gene-environment interaction and how this impacts trait expression

Characteristics that are influenced by environmental as well as genetic factors are called **multifactorial**. The idea of "nature versus nurture" — in other words, the relative influence of genetics versus environmental factors — has been and still is debated. Just looking at the genes of a given organism will not determine how that organism will develop and act. Even identical twins will show different characteristics, depending on the environment in which they live. Everyone is a product of their environment as well as their genetics.

Even when influenced by the environment, phenotypes have a normal range of expression. For instance, human height varies based on nutrition and genetics, but not many people are shorter than 4½ feet or taller than 7 feet. The range of phenotypic possibilities is called the *norm of reaction*. Hydrangeas, for example, may be blue, pink, or purple, but they are never naturally orange. Hydrangeas are blue in acidic soil with available aluminum, and they are pink in alkaline soil without available aluminum.

Figure 1. Taking a newborn blood sample for PKU testing. By Staff Sgt Eric T. Sheler, U.S. Air Force (Phenylketonuria testing) Public Domain

You may have heard about PKU, a pleiotropic disorder caused by defects in a single gene coding for an enzyme that converts the amino acid phenylalanine to tyrosine. Newborns are tested for this defect very early in life (Figure 1), so that if the results are positive, they can be given a diet limiting phenylalanine ingestion. That way, the toxic buildup is prevented and the children can develop normally. PKU is an example in which environmental factors can modify gene expression.

Practice Question

Two identical twins (female) live in different parts of the country. One is very committed to a healthy lifestyle: not smoking, exercising regularly, eating a diet rich in fresh produce, and avoiding red meats and processed foods. The other is not as careful: she smokes, is overweight, and often eats fast and processed foods. They are aware that several women in their family have had breast cancer, and decide to consult a doctor about their odds of developing the disease. Which of the following statements by the doctor sounds most correct?

a. As identical twins, you are genetically the same, so your chances of developing breast cancer are identical.
b. The twin with the healthy lifestyle should not be terribly concerned, while the one with the unhealthy lifestyle is at a higher risk.
c. Breast cancer has a genetic component, and the twins have identical genes, so they have the same genetic risk. However, environmental factors such as smoking, obesity, and consumption of red meat have been shown to increase the risk of cancer. While both twins should monitor themselves closely, the twin who smokes and is overweight may want to consider a healthier lifestyle to decrease her risk of breast cancer.

Answer

Option A is wrong; while it has been shown that certain genes may predispose people to cancer, there are many associations between environmental effects and cancer. Option B is also wrong; familial cancers have a genetic component which may or may not be balanced by a healthy lifestyle. Option C is the most correct answer; lifestyle choices are important, but genetic influences are to be taken seriously, especially if there is a family pattern associated with them.

In Summary: Effect of the Environment

While genes and genetic causes play a large role in health and phenotypes, the environment also plays an important role. Understanding this can enable the treatment of some disorders, such as the case with PKU in

which limiting the intake of phenylalanine can prevent toxic build up of this amino acid. Often the norm of reaction is set by genetic factors but ultimately determined by environmental exposures.

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PLEIOTROPY AND HUMAN DISORDERS

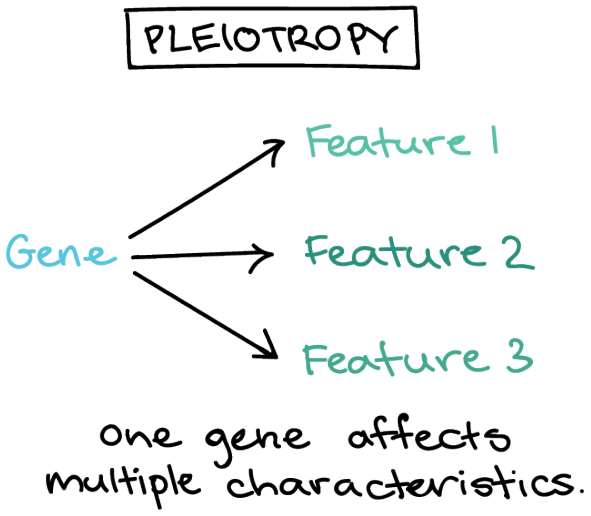
Learning Outcomes

Explain pleiotropy and its impact on traits in a population

Pleiotropy

When we discussed Mendel's experiments with purple-flowered and white-flowered plants, we didn't mention any other phenotypes associated with the two flower colors. However, Mendel noticed that the flower colors were always correlated with two other features: the color of the seed coat (covering of the seed) and the color of the axils (junctions where the leaves met the main stem) (Note: Lobo, I. (2008)).

Genes like this, which affect multiple, seemingly unrelated aspects of an organism's phenotype, are said to be **pleiotropic** (*pleio–* = many, *–tropic* = effects) (Note: *Ibid.*). The seemingly unrelated phenotypes can all be traced back to a defect in a single gene with several jobs.



Based on similar diagram by Ingrid Lobo

Importantly, alleles of pleiotropic genes are transmitted in the same way as alleles of genes that affect single traits. Although the phenotype has multiple elements, these elements are specified as a package, and the dominant and recessive versions of the package would appear in the progeny of a monohybrid cross in a ratio of 3:1.

Pleiotropy in Human Genetic Disorders

Genes affected in human genetic disorders are often pleiotropic. For example, people with the hereditary disorder Marfan syndrome may have a constellation of seemingly unrelated symptoms (Note: Marfan syndrome. (2012). In *Genetics home reference*. Retrieved from http://ghr.nlm.nih.gov/condition/marfan-syndrome.):

- Unusually tall height

- Thin fingers and toes
- Dislocation of the lens of the eye
- Heart problems (in which the aorta, the large blood vessel carrying blood away from the heart, bulges or ruptures).

These symptoms don't appear directly related to one another, but as it turns out, they can all be traced back to the mutation of a single gene.

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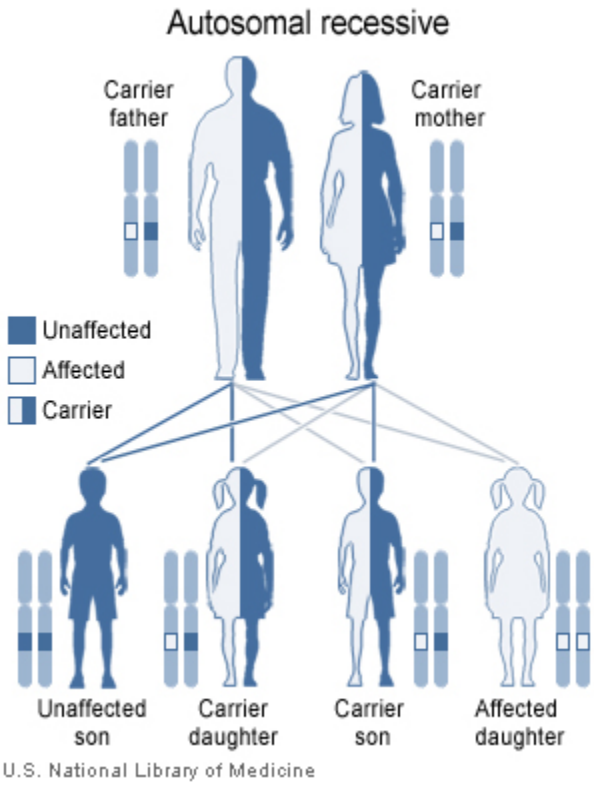
PUTTING IT TOGETHER: TRAIT INHERITANCE

Now that you understand more about genetic inheritance, you can appreciate better the complications associated with predicting traits in children. Unfortunately, these complications apply to inherited disease just as much as they apply to predicting a child's height or eye color.

There are over 6,000 monogenetic disorders and thousands more polygenetic disorders. All of this is in addition to the complications brought about by environmental influences and random genetic mutations or damage. Despite this complexity, we continually learn new things about the genetic root of human diseases. This knowledge can be used not only to predict occurrence, as a genetic counselor does, but also to develop treatments, cures, or preventatives as clinicians and researchers do.

One organization that puts this modern understanding of genetics to use is the Dor Yeshorim, an organization that offers genetic screening to members of the Jewish community worldwide. Its objective is to minimize, and eventually eliminate, the incidence of genetic disorders common to Jewish people, such as Tay–Sachs disease.

In both the Ashkenazi and Sephardi Jewish communities, there is an increased rate of a number of genetic disorders such as Tay–Sachs disease, an autosomal recessive disorder that goes unnoticed in carriers, but is fatal within the first few years of life in almost all homozygotes.



Dor Yeshorim screens only for recessive traits that give rise to lethal or severely debilitating disorders, providing preventative, rather than diagnostic services. They do not screen for disorders arising from dominant gene mutations, as these cannot be prevented by informed mate selection. Orthodox Judaism generally opposes selective abortion, and although preimplantation genetic diagnosis (PGD) is often approved by Halakha, it is a difficult and costly process. By avoiding marriages between carriers of the diseases, the incidence of the disorders decreases without having to resort to these methods.

Dor Yeshorim advocates anonymous testing. Individuals are tested during large sessions in Jewish schools and processed anonymously with only a PIN linking the sample with the candidate. When two members of the system contemplate marriage, they contact the organization and enter both their PINs. When both carry a gene for the same disorder, the risk of affected offspring is 25 percent, and it is considered advisable to discontinue the plans. In the context of shidduchim (a system of

matchmaking in Jewish communities), the “carriership check” is often run within the first three dates, to avoid disappointments and heartbreak.

Figure 1. Tay-Sachs is an autosomal recessive disease. This means that children can only inherit the Tay-Sachs if both of their parents are carriers for the disease.

According to the Dor Yeshorim website, they are “credited with singlehandedly eradicating Tay Sachs from the Jewish community and effectively closing the Tay Sachs ward at Kingsbrook Medical center forever.” (Note: "Our History", *Dor Yeshorim*, https://doryeshorim.org/history-achievements/.)

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MODULE 12: THEORY OF EVOLUTION

WHY IT MATTERS: THEORY OF EVOLUTION

Why learn about the theory of evolution?

Have you ever wondered why doctors say you should get a new flu shot every year? It's just the same old flu virus right? How much could it change from year to year?

Unfortunately, it's not just the same old flu virus—and it often changes a lot from year to year. When the flu vaccine does its work, killing the current iteration of the flu, it typically does a good job and saves its host from illness. However, as it wipes out the majority of flu viruses, the only ones that are left are those that are resistant to the current virus, which means the overall make up of the population of flu viruses has changed: the population has evolved to become resistant to one anti-viral treatment, so another must be found.

Of course, while the theory of evolution can be applied to viruses, it is more often discussed in the context of living things: bacteria, plants, animals, and even humans. The theory of evolution began as a revolutionary idea, but it remains central to the study of biology. Evolution is the unifying concept in biology. This theory documents the change in the genetic makeup of a biological population over time. Evolution helps us understand the development of antibiotic resistance in bacteria and other parasitic organisms. The following are just a few of the antibiotic resistant "bugs" plaguing humans.

- Antibiotic-Resistant Mycobacterium tuberculosis (TB)
- Methicillin-Resistant Staphylococcus aureus (MRSA)
- Vancomycin-Resistant Enterococci (VRE)
- Multidrug-Resistant Neisseria gonorrhoeae (Gonorrhea)

Learn More

You can also check out the NIH's website about antimicrobial resistance for more information.

While evolution is easiest to see in bacteria due to their short life cycles, every living population experiences evolution of one kind or another. Let's see just how the world we live in has guided the evolutionary process.

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INTRODUCTION TO CHARLES DARWIN

What you'll learn to do: Describe the work of Charles Darwin in the Galapagos Islands, especially his discovery of natural selection in finch populations

Charles Robert Darwin, was an English naturalist, geologist and biologist, best known for his contributions to the science of evolution. He established that all species of life have descended over time from common ancestors and, in a joint publication with Alfred Russel Wallace, introduced his scientific theory that this branching pattern of evolution resulted from a process that he called natural selection, in which the struggle for existence has a similar effect to the artificial selection involved in selective breeding.

Darwin published his theory of evolution with compelling evidence in his 1859 book *On the Origin of Species*, overcoming scientific rejection of earlier concepts of transmutation of species. By the 1870s, the scientific community and much of the general public had accepted evolution as a fact. However, many favored competing explanations and it was not until the emergence of the modern evolutionary synthesis from the 1930s to the 1950s that a broad consensus developed in which natural selection was the basic mechanism of evolution. Darwin's scientific discovery is the unifying theory of the life sciences, explaining the diversity of life.

In this outcome we'll learn more about his work and how it helped develop the theory of evolution.

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DARWIN AND DESCENT WITH MODIFICATION

Learning Outcomes

- Outline the work of Charles Darwin as a naturalist aboard the HMS Beagle
- Summarize the prior work and new evidence Darwin used to develop the idea of "descent with modification"

In the mid-nineteenth century, the actual mechanism for evolution was independently conceived of and described by two naturalists: Charles Darwin and Alfred Russel Wallace. Importantly, each naturalist spent time exploring the natural world on expeditions to the tropics. From 1831 to 1836, Darwin traveled around the world on ***H.M.S. Beagle***, including stops in South America, Australia, and the southern tip of Africa. Wallace traveled to Brazil to collect insects in the Amazon rainforest from 1848 to 1852 and to the Malay Archipelago from 1854 to 1862. Darwin's journey, like Wallace's later journeys to the Malay Archipelago, included stops at several island chains, the last being the Galápagos Islands west of Ecuador. On these islands, Darwin observed species of organisms on different islands that were clearly similar, yet had distinct differences. For example, the ground finches inhabiting the Galápagos Islands comprised several species with a unique beak shape (Figure 1).

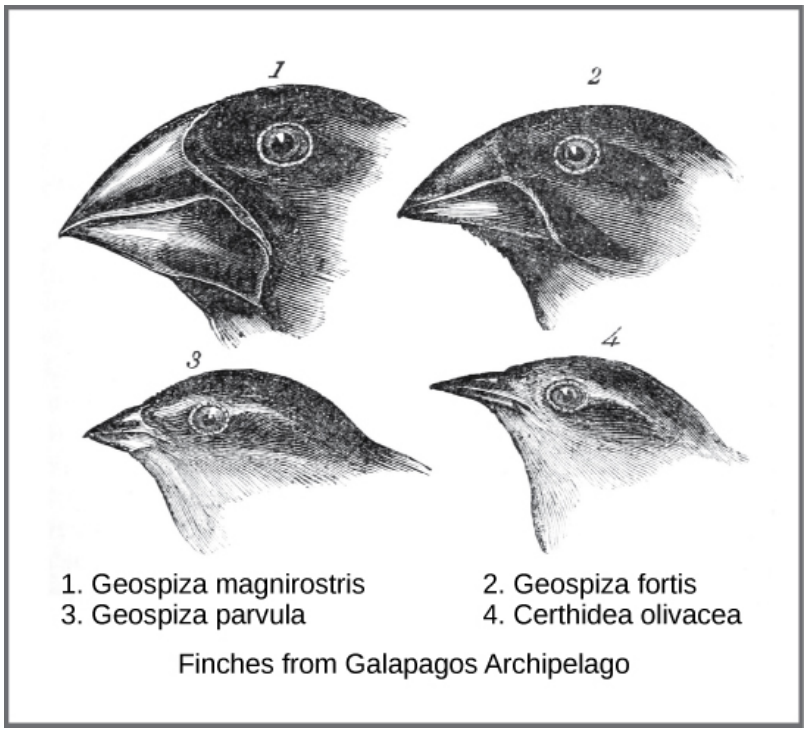


Figure 1. Darwin observed that beak shape varies among finch species. He postulated that the beak of an ancestral species had adapted over time to equip the finches to acquire different food sources.

The species on the islands had a graded series of beak sizes and shapes with very small differences between the most similar. He observed that these finches closely resembled another finch species on the mainland of South America. Darwin imagined that the island species might be species modified from one of the original mainland species. Upon further study, he realized that the varied beaks of each finch helped the birds acquire a specific type of food. For example, seed-eating finches had stronger, thicker beaks for breaking seeds, and insect-eating finches had spear-like beaks for stabbing their prey.

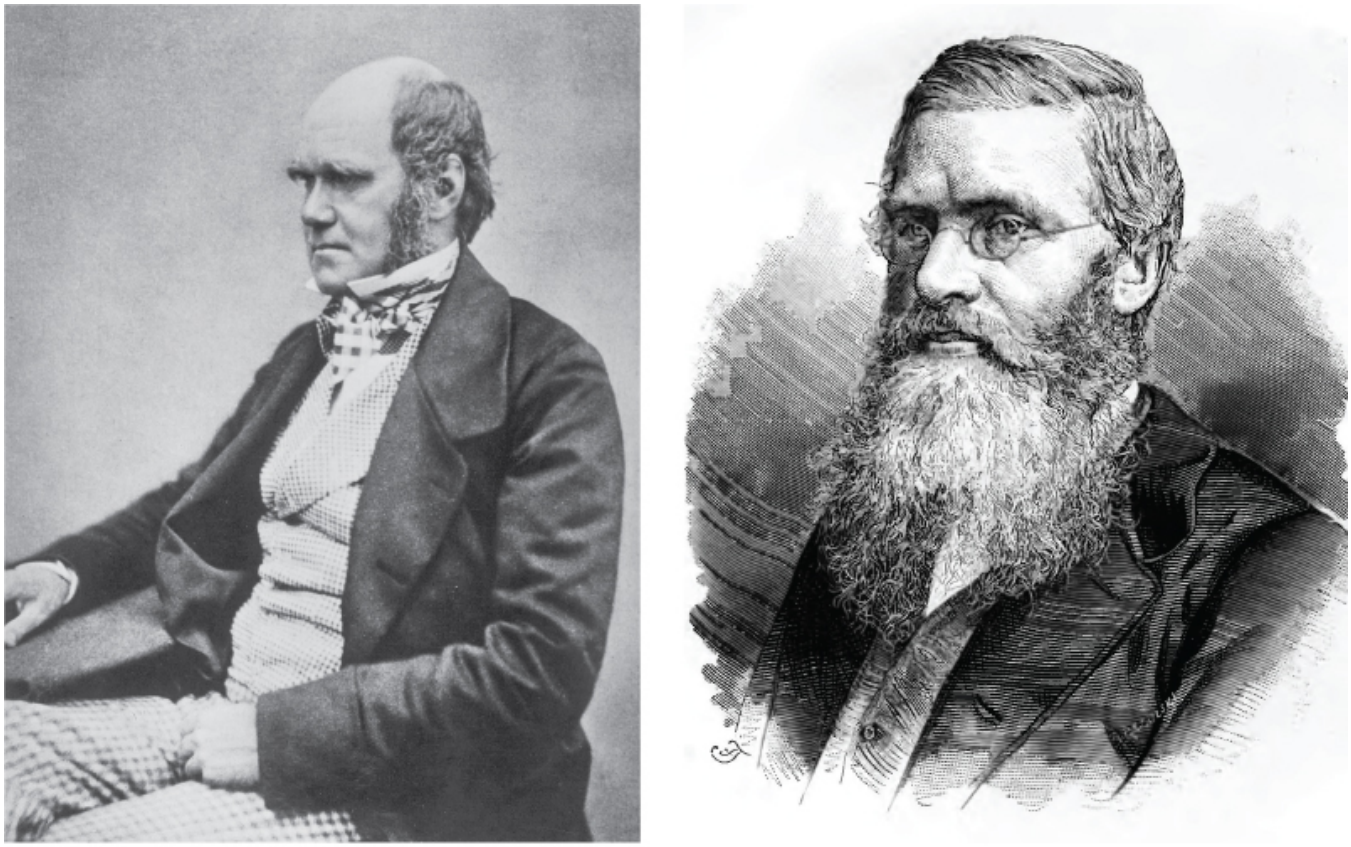
Wallace and Darwin both observed similar patterns in other organisms and they independently developed the same explanation for how and why such changes could take place. Darwin called this mechanism natural selection. Natural selection, also known as "survival of the fittest," is the more prolific reproduction of individuals with favorable traits that survive environmental change because of those traits; this leads to evolutionary change.

For example, a population of giant tortoises found in the Galapagos Archipelago was observed by Darwin to have longer necks than those that lived on other islands with dry lowlands. These tortoises were "selected" because they could reach more leaves and access more food than those with short necks. In times of drought when fewer leaves would be available, those that could reach more leaves had a better chance to eat and survive than those that couldn't reach the food source. Consequently, long-necked tortoises would be more likely to be reproductively successful and pass the long-necked trait to their offspring. Over time, only long-necked tortoises would be present in the population.

Natural selection, Darwin argued, was an inevitable outcome of three principles that operated in nature. First, most characteristics of organisms are inherited, or passed from parent to offspring. Although no one, including Darwin and Wallace, knew how this happened at the time, it was a common understanding. Second, more offspring are produced than are able to survive, so resources for survival and reproduction are limited. The capacity for reproduction in all organisms outstrips the availability of resources to support their numbers. Thus, there is competition for those resources in each generation. Both Darwin and Wallace's understanding of this principle came from reading an essay by the economist Thomas Malthus who discussed this principle in relation to human populations. Third, offspring vary among each other in regard to their characteristics and those variations are inherited. Darwin and Wallace reasoned that offspring with inherited characteristics which allow them to best compete for limited resources will survive and have more offspring than those individuals with variations that are less able to compete. Because characteristics are inherited, these traits will be better represented in the next generation. This will lead to change in populations over generations in a process that Darwin called descent with modification. Ultimately, natural selection leads to greater adaptation of the population

to its local environment; it is the only mechanism known for adaptive evolution. It is important to keep in mind that natural selection leads to adaptation to a *specific* environment, not all environments or even a different environment.

Papers by Darwin and Wallace (Figure 2) presenting the idea of natural selection were read together in 1858 before the Linnean Society in London. The following year Darwin's book, ***On the Origin of Species****,* was published. His book outlined in considerable detail his arguments for gradual changes and adaptive survival by natural selection.



(a) (b)

Figure 2. Both (a) Charles Darwin and (b) Alfred Wallace wrote scientific papers on natural selection that were presented together before the Linnean Society in 1858.

Demonstrations of evolution by natural selection are time consuming and difficult to obtain. One of the best examples has been demonstrated in the very birds that helped to inspire Darwin's theory: the Galápagos finches. Peter and Rosemary Grant and their colleagues have studied Galápagos finch populations every year since 1976 and have provided important demonstrations of natural selection. The Grants found changes from one generation to the next in the distribution of beak shapes with the medium ground finch on the Galápagos island of Daphne Major. The birds have inherited variation in the bill shape with some birds having wide deep bills and others having thinner bills. During a period in which rainfall was higher than normal because of an El Niño, the large hard seeds that large-billed birds ate were reduced in number; however, there was an abundance of the small soft seeds which the small-billed birds ate. Therefore, survival and reproduction were much better in the following years for the small-billed birds. In the years following this El Niño, the Grants measured beak sizes in the population and found that the average bill size was smaller. Since bill size is an inherited trait, parents with smaller bills had more offspring and the size of bills had evolved to be smaller. As conditions improved in 1987 and larger seeds became more available, the trend toward smaller average bill size ceased.

In Summary: Darwin and Descent with Modification

While Charles Darwin is generally called "**the father of evolution**," the basic idea for this concept was actually developed by both Darwin and Alfred Russel Wallace. Both scientists based their hypotheses on observations of diversity among natural populations. Darwin's work in particular focused on animals of the Galapagos islands, especially finches. Over time, the idea that species changed from natural selection pressures through "descent with modification" gave rise to the idea of evolution. Data accumulated over time, for example the long study of the Galapagos finches by the Grant research team, has supported this idea and moved it into the realm of a supported theory of biology.

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DARWIN AND THE THEORY OF EVOLUTION

Learning Outcomes

Describe how Darwin's work developed to the theory of evolution

Natural selection can only take place if there is **variation**, or differences, among individuals in a population. Importantly, these differences must have some genetic basis; otherwise, the selection will not lead to change in the next generation. This is critical because variation among individuals can be caused by non-genetic reasons such as an individual being taller because of better nutrition rather than different genes.

Genetic diversity in a population comes from two main mechanisms: mutation and sexual reproduction. Mutation, a change in DNA, is the ultimate source of new alleles, or new genetic variation in any population. The genetic changes caused by mutation can have one of three outcomes on the phenotype. A mutation affects the phenotype of the organism in a way that gives it reduced fitness—lower likelihood of survival or fewer offspring. A mutation may produce a phenotype with a beneficial effect on fitness. And, many mutations will also have no effect on the fitness of the phenotype; these are called neutral mutations. Mutations may also have a whole range of effect sizes on the fitness of the organism that expresses them in their phenotype, from a small effect to a great effect. Sexual reproduction also leads to genetic diversity: when two parents reproduce, unique combinations of alleles assemble to produce the unique genotypes and thus phenotypes in each of the offspring.

A heritable trait that helps the survival and reproduction of an organism in its present environment is called an **adaptation**. Scientists describe groups of organisms becoming adapted to their environment when a change in the range of genetic variation occurs over time that increases or maintains the "fit" of the population to its environment. The webbed feet of platypuses are an adaptation for swimming. The snow leopards' thick fur is an adaptation for living in the cold. The cheetahs' fast speed is an adaptation for catching prey.

Whether or not a trait is favorable depends on the environmental conditions at the time. The same traits are not always selected because environmental conditions can change. For example, consider a species of plant that grew in a moist climate and did not need to conserve water. Large leaves were selected because they allowed the plant to obtain more energy from the sun. Large leaves require more water to maintain than small leaves, and the moist environment provided favorable conditions to support large leaves. After thousands of years, the climate changed, and the area no longer had excess water. The direction of natural selection shifted so that plants with small leaves were selected because those populations were able to conserve water to survive the new environmental conditions.

The evolution of species has resulted in enormous variation in form and function. Sometimes, evolution gives rise to groups of organisms that become tremendously different from each other. When two species evolve in diverse directions from a common point, it is called divergent evolution. Such **divergent evolution** can be seen in the forms of the reproductive organs of flowering plants which share the same basic anatomies; however, they can look very different as a result of selection in different physical environments and adaptation to different kinds of pollinators (Figure 1).

Figure 1. Flowering plants evolved from a common ancestor. Notice that the (a) dense blazing star (Liatrus spicata) and the (b) purple coneflower (Echinacea purpurea) vary in appearance, yet both share a similar basic morphology. (credit a: modification of work by Drew Avery; credit b: modification of work by Cory Zanker)

In other cases, similar phenotypes evolve independently in distantly related species. For example, flight has evolved in both bats and insects, and they both have structures we refer to as wings, which are adaptations to flight. However, the wings of bats and insects have evolved from very different original structures. This phenomenon is called **convergent evolution**, where similar traits evolve independently in species that do not share a recent common ancestry. The two species came to the same function, flying, but did so separately from each other.

These physical changes occur over enormous spans of time and help explain how evolution occurs. Natural selection acts on individual organisms, which in turn can shape an entire species. Although natural selection may work in a single generation on an individual, it can take thousands or even millions of years for the genotype of an entire species to evolve. It is over these large time spans that life on earth has changed and continues to change.

In Summary: Darwin and the Theory of Evolution

Natural selection, the driving force behind evolution, can only work if variation exists among organisms. Variation arises ultimately from genetic mutations. Diversity is further encouraged through sexual reproduction. As environments change, selective pressures shift and favor different adaptations. In this way, given thousands or millions of years, species evolve.

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INTRODUCTION TO EVIDENCE FOR EVOLUTION

What you'll learn to do: Describe how the theory of evolution by natural selection is supported by evidence

The evidence for evolution is compelling and extensive. Looking at every level of organization in living systems, biologists see the signature of past and present evolution. Darwin dedicated a large portion of his book, *On the Origin of Species*, to identifying patterns in nature that were consistent with evolution, and since Darwin, our understanding has become clearer and broader.

The video below summarizes the major types of evidence supporting evolution; you will read more details in the pages that follow.

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PHYSICAL EVIDENCE

Learning Outcomes

Outline physical evidence that supports the theory of evolution

Fossils

Fossils provide solid evidence that organisms from the past are not the same as those found today, and fossils show a progression of evolution. Scientists determine the age of fossils and categorize them from all over the world to determine when the organisms lived relative to each other. The resulting fossil record tells the story of the past and shows the evolution of form over millions of years (Figure 1a). For example, scientists have recovered highly detailed records showing the evolution of humans and horses (Figure 1b).

Figure 1. In this (a) display, fossil hominids are arranged from oldest (bottom) to newest (top). As hominids evolved, the shape of the skull changed. An artist's rendition of (b) extinct species of the genus Equus reveals that these ancient species resembled the modern horse (Equus ferus) but varied in size.

Anatomy and Embryology

Another type of evidence for evolution is the presence of structures in organisms that share the same basic form. For example, the bones in the appendages of a human, dog, bird, and whale all share the same overall construction (Figure 2) resulting from their origin in the appendages of a common ancestor. Over time, evolution led to changes in the shapes and sizes of these bones in different species, but they have maintained the same overall layout. Scientists call these synonymous parts homologous structures.

Some structures exist in organisms that have no apparent function at all, and appear to be residual parts from a past common ancestor. These unused structures without function are called vestigial structures. Some examples of vestigial structures are wings on flightless birds, leaves on some cacti, and hind leg bones in whales.

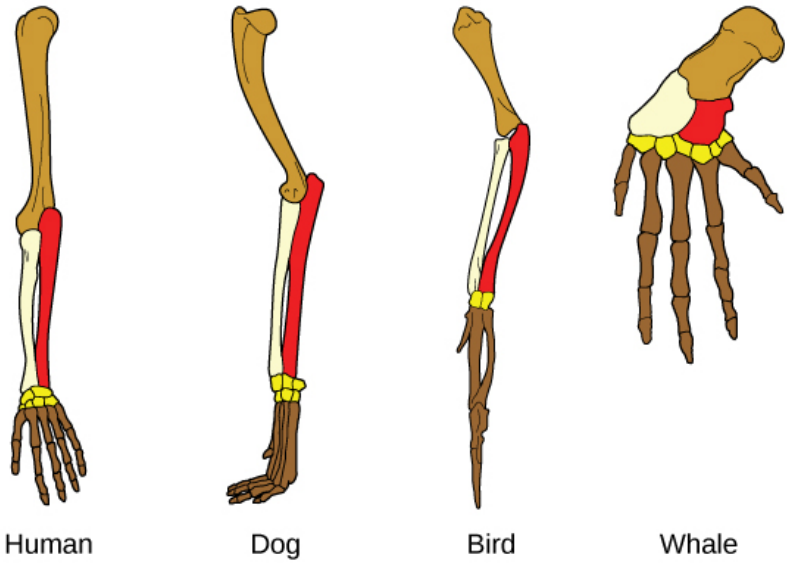

Figure 2. The similar construction of these appendages indicates that these organisms share a common ancestor.

Visit this interactive site to guess which bones structures are homologous and which are analogous, and see examples of evolutionary adaptations to illustrate these concepts.

Another evidence of evolution is the convergence of form in organisms that share similar environments. For example, species of unrelated animals, such as the arctic fox and ptarmigan, living in the arctic region have been selected for seasonal white phenotypes during winter to blend with the snow and ice (Figure 3). These similarities occur not because of common ancestry, but because of similar selection pressures—the benefits of not being seen by predators.

Figure 3. The white winter coat of the (a) arctic fox and the (b) ptarmigan's plumage are adaptations to their environments. (credit a: modification of work by Keith Morehouse)

Embryology, the study of the development of the anatomy of an organism to its adult form, also provides evidence of relatedness between now widely divergent groups of organisms. Mutational tweaking in the embryo can have such magnified consequences in the adult that embryo formation tends to be conserved. As a result, structures that are absent in some groups often appear in their embryonic forms and disappear by the time the adult or juvenile form is reached. For example, all vertebrate embryos, including humans, exhibit gill slits and tails at some point in their early development. These disappear in the adults of terrestrial groups but are maintained in adult forms of aquatic groups such as fish and some amphibians. Great ape embryos, including humans, have a tail structure during their development that is lost by the time of birth.

In Summary: Physical Evidence

Since Darwin developed his ideas on descent with modification and the pressures of natural selection, a variety of evidence has been gathered supporting the theory of evolution. Fossil evidence shows the changes in lineages over millions of years, such as in hominids and horses. Studying anatomy allows scientists to identify homologous structures across diverse groups of related organisms, such as leg bones. Vestigial structures also offer clues to common ancestors. Using embryology, scientists can identify common ancestors through structures present only during development and not in the adult form.

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BIOLOGICAL EVIDENCE

Learning Outcomes

Outline biological evidence that supports the theory of evolution

Biogeography

The geographic distribution of organisms on the planet follows patterns that are best explained by evolution in conjunction with the movement of tectonic plates over geological time. Broad groups that evolved before the breakup of the supercontinent Pangaea (about 200 million years ago) are distributed worldwide. Groups that evolved since the breakup appear uniquely in regions of the planet, such as the unique flora and fauna of northern continents that formed from the supercontinent Laurasia and of the southern continents that formed from the supercontinent Gondwana. The presence of members of the plant family Proteaceae in Australia, southern Africa, and South America is best due to their appearance prior to the southern supercontinent Gondwana breaking up.

The great diversification of marsupials in Australia and the absence of other mammals reflect Australia's long isolation. Australia has an abundance of endemic species—species found nowhere else—which is typical of islands whose isolation by expanses of water prevents species migration. This geographical isolation makes islands "hotbeds" for selective pressures. Due to these pressures, over time these species diverge evolutionarily into new species that look very different from their ancestors that may exist on the mainland. The marsupials of Australia, the finches on the Galápagos, and many species on the Hawaiian Islands are all unique to their one point of origin, yet they display distant relationships to ancestral species on mainlands.

Molecular Biology

Like anatomical structures, the structures of the molecules of life reflect descent with modification. Evidence of a common ancestor for all of life is reflected in the universality of DNA as the genetic material and in the near universality of the genetic code and the machinery of DNA replication and expression. Fundamental divisions in life between the three domains are reflected in major structural differences in otherwise conservative structures such as the components of ribosomes and the structures of membranes. In general, the relatedness of groups of organisms is reflected in the similarity of their DNA sequences—exactly the pattern that would be expected from descent and diversification from a common ancestor.

DNA sequences have also shed light on some of the mechanisms of evolution. For example, it is clear that the evolution of new functions for proteins commonly occurs after gene duplication events that allow the free modification of one copy by mutation, selection, or drift (changes in a population's gene pool resulting from chance), while the other copy continues to produce a functional protein.

In Summary: Biological Evidence

Biogeography offers further clues about evolutionary relationships. The presence of related organisms across continents indicates when these organisms may have evolved. For example, some flora and fauna of the northern continents are similar across these landmasses but distinct from that of the southern continents. Islands such as Australia and the Galapagos chain often have unique species that evolved after these landmasses separated from the mainland. Finally, molecular biology provides data supporting the theory of evolution. In particular, the universality of DNA and near universality of the genetic code for proteins shows

that all life once shared a common ancestor. DNA also provides clues into how evolution may have happened. Gene duplications allow one copy to undergo mutational events without harming an organism, as one copy continues to produce functional protein.

Evolution—It's a Thing

This video defines evolution and discusses several varieties of evidence that support the Theory of Evolution:

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MISCONCEPTIONS OF EVOLUTION

Learning Outcomes

Refute common misconceptions about evolution

Although the theory of evolution generated some controversy when it was first proposed, it was almost universally accepted by biologists, particularly younger biologists, within 20 years after publication of *On the Origin of Species*. Nevertheless, the theory of evolution is a difficult concept and misconceptions about how it works abound.

This site addresses some of the main misconceptions associated with the theory of evolution.

Evolution Is Just a Theory

Critics of the theory of evolution dismiss its importance by purposefully confounding the everyday usage of the word "theory" with the way scientists use the word. In science, a "theory" is understood to be a body of thoroughly tested and verified explanations for a set of observations of the natural world. Scientists have a theory of the atom, a theory of gravity, and the theory of relativity, each of which describes understood facts about the world. In the same way, the theory of evolution describes facts about the living world. As such, a theory in science has survived significant efforts to discredit it by scientists. In contrast, a "theory" in common vernacular is a word meaning a guess or suggested explanation; this meaning is more akin to the scientific concept of "hypothesis." When critics of evolution say evolution is "just a theory," they are implying that there is little evidence supporting it and that it is still in the process of being rigorously tested. This is a mischaracterization.

Individuals Evolve

Evolution is the change in genetic composition of a population over time, specifically over generations, resulting from differential reproduction of individuals with certain alleles. Individuals do change over their lifetime, obviously, but this is called development and involves changes programmed by the set of genes the individual acquired at

birth in coordination with the individual's environment. When thinking about the evolution of a characteristic, it is probably best to think about the change of the average value of the characteristic in the population over time. For example, when natural selection leads to bill-size change in medium-ground finches in the Galápagos, this does not mean that individual bills on the finches are changing. If one measures the average bill size among all individuals in the population at one time and then measures the average bill size in the population several years later, this average value will be different as a result of evolution. Although some individuals may survive from the first time to the second, they will still have the same bill size; however, there will be many new individuals that contribute to the shift in average bill size.

Organisms Evolve on Purpose

Statements such as "organisms evolve in response to a change in an environment" are quite common, but such statements can lead to two types of misunderstandings. First, the statement must not be understood to mean that individual organisms evolve. The statement is shorthand for "a population evolves in response to a changing environment." However, a second misunderstanding may arise by interpreting the statement to mean that the evolution is somehow intentional. A changed environment results in some individuals in the population, those with particular phenotypes, benefiting and therefore producing proportionately more offspring than other phenotypes. This results in change in the population if the characteristics are genetically determined.

It is also important to understand that the variation that natural selection works on is already in a population and does not arise in response to an environmental change. For example, applying antibiotics to a population of bacteria will, over time, select a population of bacteria that are resistant to antibiotics. The resistance, which is caused by a gene, did not arise by mutation because of the application of the antibiotic. The gene for resistance was already present in the gene pool of the bacteria, likely at a low frequency. The antibiotic, which kills the bacterial cells without the resistance gene, strongly selects individuals that are resistant, since these would be the only ones that survived and divided. Experiments have demonstrated that mutations for antibiotic resistance do not arise as a result of antibiotic.

In a larger sense, evolution is not goal directed. Species do not become "better" over time; they simply track their changing environment with adaptations that maximize their reproduction in a particular environment at a particular time. Evolution has no goal of making faster, bigger, more complex, or even smarter species, despite the commonness of this kind of language in popular discourse. What characteristics evolve in a species are a function of the variation present and the environment, both of which are constantly changing in a non-directional way. What trait is fit in one environment at one time may well be fatal at some point in the future. This holds equally well for a species of insect as it does the human species.

Evolution Explains the Origin of Life

It is a common misunderstanding that evolution includes an explanation of life's origins. Conversely, some of the theory's critics believe that it cannot explain the origin of life. The theory does not try to explain the origin of life. The theory of evolution explains how populations change over time and how life diversifies the origin of species. It does not shed light on the beginnings of life including the origins of the first cells, which is how life is defined. The mechanisms of the origin of life on Earth are a particularly difficult problem because it occurred a very long time ago, and presumably it just occurred once. Importantly, biologists believe that the presence of life on Earth precludes the possibility that the events that led to life on Earth can be repeated because the intermediate stages would immediately become food for existing living things.

However, once a mechanism of inheritance was in place in the form of a molecule like DNA either within a cell or pre-cell, these entities would be subject to the principle of natural selection. More effective reproducers would increase in frequency at the expense of inefficient reproducers. So while evolution does not explain the origin of life, it may have something to say about some of the processes operating once pre-living entities acquired certain properties.

In Summary: Misconceptions of Evolution

Many misconceptions exist about the theory of evolution—including some perpetuated by critics of the theory. First, evolution as a scientific theory means that it has years of observation and accumulated data supporting it. It is not “just a theory” as a person may say in common vernacular.

Another misconception is that individuals evolve, though in fact it is populations that evolve over time. Individuals simply carry mutations. Furthermore, these mutations neither arise on purpose nor do they arise in response to an environmental pressure. Instead, mutations in DNA happen spontaneously and are already present in individuals of a population when a selective pressure occurs. Once the environment begins to favor a particular trait, then those individuals already carrying that mutation will have a selective advantage and are likely to survive better and outproduce others without the adaptation.

Finally, the theory of evolution does not in fact address the origins of life on this planet. Scientists believe that we cannot, in fact, repeat the circumstances that led to life on Earth because at this time life already exists. The presence of life has so dramatically changed the environment that the origins cannot be totally produced for study.

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INTRODUCTION TO MUTATIONS AND EVOLUTION

What you’ll learn to do: Recognize that mutations are the basis of microevolution; and that adaptations enhance the survival and reproduction of individuals in a population

We’ve already learned about DNA and mutations, now we’ll learn about how these mutations can drive evolution. This type of evolution falls under the category of microevolution.

Microevolution is the change in allele frequencies that occurs over time within a population. This change is due to five different processes: mutation, selection (natural and artificial), gene flow, gene migration and genetic drift. This change happens over a relatively short (in evolutionary terms) amount of time compared to the changes termed ‘macroevolution’ which is where greater differences in the population occur.

Population genetics is the branch of biology that provides the mathematical structure for the study of the process of microevolution. Ecological genetics concerns itself with observing microevolution in the wild. Typically, observable instances of evolution are examples of microevolution; for example, bacterial strains that have antibiotic resistance.

Microevolution over time leads to speciation or the appearance of novel structure, sometimes classified as macroevolution. Macro and microevolution describe fundamentally identical processes on different scales.

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POPULATION GENETICS

Learning Outcomes

Understand the connection between genetics and evolution

Darwin Meets Mendel—Not Literally

When Darwin came up with his theories of evolution and natural selection, he knew that the processes he was describing depended on heritable variation in populations. That is, they relied on differences in the features of the organisms in a population and on the ability of these different features to be passed on to offspring.

Darwin did not, however, know how traits were inherited. Like other scientists of his time, he thought that traits were passed on via blending inheritance. In this model, parents' traits are supposed to permanently blend in their offspring. The blending model was disproven by Austrian monk Gregor Mendel, who found that traits are specified by non-blending heritable units called genes.

Although Mendel published his work on genetics just a few years after Darwin published his ideas on evolution, Darwin probably never read Mendel's work. Today, we can combine Darwin's and Mendel's ideas to arrive at a clearer understanding of what evolution is and how it takes place.

Microevolution and Population Genetics

Microevolution, or evolution on a small scale, is defined as a change in the frequency of gene variants, alleles, in a population over generations. The field of biology that studies allele frequencies in populations and how they change over time is called **population genetics**.

Microevolution is sometimes contrasted with **macroevolution**, evolution that involves large changes, such as formation of new groups or species, and happens over long time periods. However, most biologists view microevolution and macroevolution as the same process happening on different timescales. Microevolution adds up gradually, over long periods of time to produce macroevolutionary changes. It is important to remember that both these processes are based on changes in DNA sequences, or mutations. Not all mutations are beneficial, just as not all are harmful. Furthermore, the impact of a particular mutation (benefit or harm) may change if the environment changes. This is natural selection in action.

Let's look at three concepts that are core to the definition of microevolution: populations, alleles, and allele frequency.

Populations

A **population** is a group of organisms of the same species that are found in the same area and can interbreed. A population is the smallest unit that can evolve—in other words, an individual can't evolve.

Alleles

An **allele** is a version of a **gene**, a heritable unit that controls a particular feature of an organism.

For instance, Mendel studied a gene that controls flower color in pea plants. This gene comes in a white allele, *w*, and a purple allele, *W*. Each pea plant has two gene copies, which may be the same or different alleles. When the

alleles are different, one—the **dominant** allele, *W*—may hide the other—the **recessive allele**, *w*. A plant's set of alleles, called its **genotype**, determines its **phenotype**, or observable features, in this case flower color.

Allele Frequency

Allele frequency refers to how frequently a particular allele appears in a population. For instance, if all the alleles in a population of pea plants were purple alleles, *W*, the allele frequency of *W* would be 100%, or 1.0. However, if half the alleles were *W* and half were *w*, each allele would have an allele frequency of 50%, or 0.5.

In general, we can define allele frequency as

$$\text{Frequency of allele } A = \frac{\text{Number of copies of allele } A \text{ in population}}{\text{Total number of } A/a \text{ gene copies in population}}$$

Sometimes there are more than two alleles in a population (e.g., there might be *A*, *a*, and A_i alleles of a gene). In that case, you would want to add up *all* of the different alleles to get your denominator.

It's also possible to calculate **genotype frequencies**—the fraction of individuals with a given genotype—and **phenotype frequencies**—the fraction of individuals with a given phenotype. Keep in mind, though, that these are different concepts from allele frequency. We'll see an example of this difference next.

Video Summary

This video talks about population genetics, which helps to explain the evolution of populations over time.
Watch this video online: https://youtu.be/WhFKPaRnTdQ

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SELECTIVE AND ENVIRONMENTAL PRESSURES

Learning Outcomes

Understand how environmental changes and selective pressures impact the spread of mutations, contributing to the process of evolution

Natural selection only acts on the population's heritable traits: selecting for beneficial alleles and thus increasing their frequency in the population, while selecting against deleterious alleles and thereby decreasing their frequency—a process known as **adaptive evolution**. Natural selection does not act on individual alleles, however, but on entire organisms. An individual may carry a very beneficial genotype with a resulting phenotype that, for example, increases the ability to reproduce (fecundity), but if that same individual also carries an allele that results in a fatal childhood disease, that fecundity phenotype will not be passed on to the next generation because the individual will not live to reach reproductive age. Natural selection acts at the level of the individual; it selects for individuals with greater contributions to the gene pool of the next generation, known as an organism's **evolutionary (Darwinian) fitness**.

Fitness is often quantifiable and is measured by scientists in the field. However, it is not the absolute fitness of an individual that counts, but rather how it compares to the other organisms in the population. This concept, called **relative fitness**, allows researchers to determine which individuals are contributing additional offspring to the next generation, and thus, how the population might evolve.

There are several ways selection can affect population variation: stabilizing selection, directional selection, diversifying selection, frequency-dependent selection, and sexual selection. As natural selection influences the allele frequencies in a population, individuals can either become more or less genetically similar and the phenotypes displayed can become more similar or more disparate.

Stabilizing Selection

If natural selection favors an average phenotype, selecting against extreme variation, the population will undergo **stabilizing selection** (Figure 1a). In a population of mice that live in the woods, for example, natural selection is likely to favor individuals that best blend in with the forest floor and are less likely to be spotted by predators. Assuming the ground is a fairly consistent shade of brown, those mice whose fur is most closely matched to that color will be most likely to survive and reproduce, passing on their genes for their brown coat. Mice that carry alleles that make them a bit lighter or a bit darker will stand out against the ground and be more likely to fall victim to predation. As a result of this selection, the population's genetic variance will decrease.

Directional Selection

When the environment changes, populations will often undergo **directional selection** (Figure 1b), which selects for phenotypes at one end of the spectrum of existing variation. A classic example of this type of selection is the evolution of the peppered moth in eighteenth- and nineteenth-century England. Prior to the Industrial Revolution, the moths were predominately light in color, which allowed them to blend in with the light-colored trees and lichens in their environment. But as soot began spewing from factories, the trees became darkened, and the light-colored moths became easier for predatory birds to spot. Over time, the frequency of the melanic form of the moth increased because they had a higher survival rate in habitats affected by air pollution because their darker coloration blended with the sooty trees. Similarly, the hypothetical mouse population may evolve to take on a different coloration if something were to cause the forest floor where they live to change color. The result of this type of selection is a shift in the population's genetic variance toward the new, fit phenotype.

> In science, sometimes things are believed to be true, and then new information comes to light that changes our understanding. The story of the peppered moth is an example: the facts behind the selection toward darker moths have recently been called into question. Read this article to learn more.

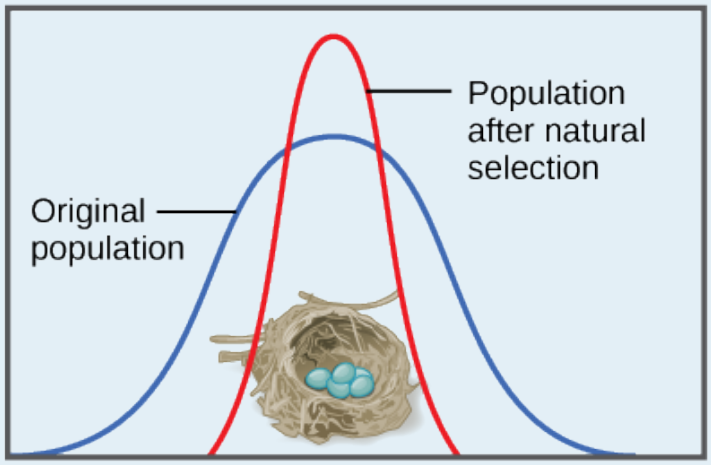
Diversifying Selection

Sometimes two or more distinct phenotypes can each have their advantages and be selected for by natural selection, while the intermediate phenotypes are, on average, less fit. Known as diversifying selection (Figure 1c), this is seen in many populations of animals that have multiple male forms. Large, dominant alpha males obtain mates by brute force, while small males can sneak in for furtive copulations with the females in an alpha male's territory. In this case, both the alpha males and the "sneaking" males will be selected for, but medium-sized males, which can't overtake the alpha males and are too big to sneak copulations, are selected against.

Diversifying selection can also occur when environmental changes favor individuals on either end of the phenotypic spectrum. Imagine a population of mice living at the beach where there is light-colored sand interspersed with patches of tall grass. In this scenario, light-colored mice that blend in with the sand would be favored, as well as dark-colored mice that can hide in the grass. Medium-colored mice, on the other hand, would not blend in with either the grass or the sand, and would thus be more likely to be eaten by predators. The result of this type of selection is increased genetic variance as the population becomes more diverse.

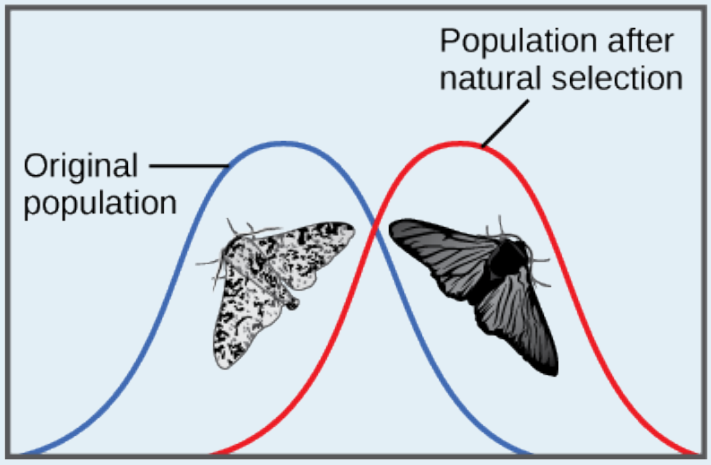
Practice Question

(a) **Stabilizing selection**



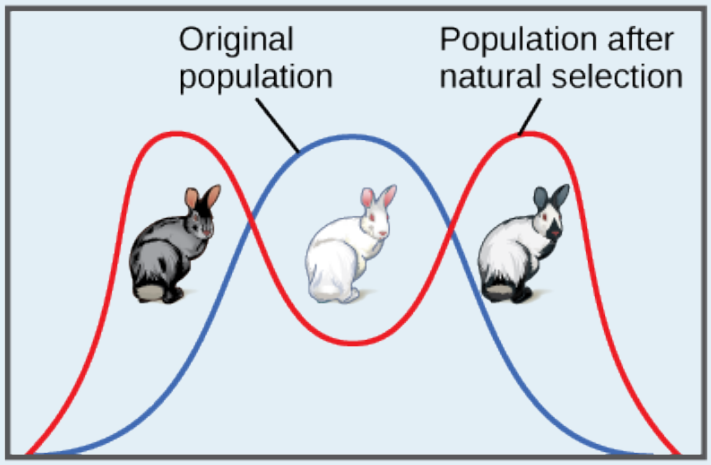
Robins typically lay four eggs, an example of stabilizing selection. Larger clutches may result in malnourished chicks, while smaller clutches may result in no viable offspring.

(b) **Directional selection**



Light-colored peppered moths are better camouflaged against a pristine environment; likewise, dark-colored peppered moths are better camouflaged against a sooty environment. Thus, as the Industrial Revolution progressed in nineteenth-century England, the color of the moth population shifted from light to dark, an example of directional selection.

(c) **Diversifying selection**



In a hyphothetical population, gray and Himalayan (gray and white) rabbits are better able to blend with a rocky environment than white rabbits, resulting in diversifying selection.

Figure 1. Different types of natural selection can impact the distribution of phenotypes within a population. In (a) stabilizing selection, an average phenotype is favored. In (b) directional selection, a change in the environment shifts the spectrum of phenotypes observed. In (c) diversifying selection, two or more extreme phenotypes are selected for, while the average phenotype is selected against.

In recent years, factories have become cleaner, and less soot is released into the environment. What impact do you think this has had on the distribution of moth color in the population?

Answer

Moths have shifted to a lighter color.

Frequency-dependent Selection

Another type of selection, called **frequency-dependent selection**, favors phenotypes that are either common (positive frequency-dependent selection) or rare (negative frequency-dependent selection). An interesting example of this type of selection is seen in a unique group of lizards of the Pacific Northwest. Male common side-blotched lizards come in three throat-color patterns: orange, blue, and yellow. Each of these forms has a different reproductive strategy: orange males are the strongest and can fight other males for access to their females; blue males are medium-sized and form strong pair bonds with their mates; and yellow males (Figure 2) are the smallest, and look a bit like females, which allows them to sneak copulations. Like a game of rock-paper-scissors, orange beats blue, blue beats yellow, and yellow beats orange in the competition for females. That is, the big, strong orange males can fight off the blue males to mate with the blue's pair-bonded females, the blue males are successful at guarding their mates against yellow sneaker males, and the yellow males can sneak copulations from the potential mates of the large, polygynous orange males.

Figure 2. A yellow-throated side-blotched lizard is smaller than either the blue-throated or orange-throated males and appears a bit like the females of the species, allowing it to sneak copulations. (credit: "tinyfroglet"/Flickr)

In this scenario, orange males will be favored by natural selection when the population is dominated by blue males, blue males will thrive when the population is mostly yellow males, and yellow males will be selected for when orange males are the most populous. As a result, populations of side-blotched lizards cycle in the distribution of these phenotypes—in one generation, orange might be predominant, and then yellow males will begin to rise in frequency. Once yellow males make up a majority of the population, blue males will be selected for. Finally, when blue males become common, orange males will once again be favored.

Negative frequency-dependent selection serves to increase the population's genetic variance by selecting for rare phenotypes, whereas positive frequency-dependent selection usually decreases genetic variance by selecting for common phenotypes.

Sexual Selection

Males and females of certain species are often quite different from one another in ways beyond the reproductive organs. Males are often larger, for example, and display many elaborate colors and adornments, like the peacock's tail, while females tend to be smaller and duller in decoration. Such differences are known as **sexual dimorphisms** (Figure 3), which arise from the fact that in many populations, particularly animal populations, there is more variance in the reproductive success of the males than there is of the females. That is, some males—often the bigger, stronger, or more decorated males—get the vast majority of the total matings, while others receive none. This can occur because the males are better at fighting off other males, or because females will choose to mate with the bigger or more decorated males. In either case, this variation in reproductive success generates a strong selection pressure among males to get those matings, resulting in the evolution of bigger body size and elaborate ornaments to get the females' attention. Females, on the other hand, tend to get a handful of selected matings; therefore, they are more likely to select more desirable males.

Sexual dimorphism varies widely among species, of course, and some species are even sex-role reversed. In such cases, females tend to have a greater variance in their reproductive success than males and are correspondingly selected for the bigger body size and elaborate traits usually characteristic of males.

Figure 3. Sexual dimorphism is observed in (a) peacocks and peahens, (b) Argiope appensa spiders (the female spider is the large one), and in (c) wood ducks. (credit "spiders": modification of work by "Sanba38"/Wikimedia Commons; credit "duck": modification of work by Kevin Cole)

The selection pressures on males and females to obtain matings is known as sexual selection; it can result in the development of secondary sexual characteristics that do not benefit the individual's likelihood of survival but help to maximize its reproductive success. Sexual selection can be so strong that it selects for traits that are actually detrimental to the individual's survival. Think, once again, about the peacock's tail. While it is beautiful and the male with the largest, most colorful tail is more likely to win the female, it is not the most practical appendage. In addition to being more visible to predators, it makes the males slower in their attempted escapes. There is some evidence that this risk, in fact, is why females like the big tails in the first place. The speculation is that large tails carry risk, and only the best males survive that risk: the bigger the tail, the more fit the male. This idea is known as the **handicap principle**.

The **good genes hypothesis** states that males develop these impressive ornaments to show off their efficient metabolism or their ability to fight disease. Females then choose males with the most impressive traits because it signals their genetic superiority, which they will then pass on to their offspring. Though it might be argued that females should not be picky because it will likely reduce their number of offspring, if better males father more fit offspring, it may be beneficial. Fewer, healthier offspring may increase the chances of survival more than many, weaker offspring.

In both the handicap principle and the good genes hypothesis, the trait is said to be an **honest signal** of the males' quality, thus giving females a way to find the fittest mates— males that will pass the best genes to their offspring.

No Perfect Organism

Natural selection is a driving force in evolution and can generate populations that are better adapted to survive and successfully reproduce in their environments. But natural selection cannot produce the perfect organism. Natural selection can only select on existing variation in the population; it does not create anything from scratch. Thus, it is limited by a population's existing genetic variance and whatever new alleles arise through mutation and gene flow.

Natural selection is also limited because it works at the level of individuals, not alleles, and some alleles are linked due to their physical proximity in the genome, making them more likely to be passed on together (linkage disequilibrium). Any given individual may carry some beneficial alleles and some unfavorable alleles. It is the net effect of these alleles, or the organism's fitness, upon which natural selection can act. As a result, good alleles can be lost if they are carried by individuals that also have several overwhelmingly bad alleles; likewise, bad alleles can be kept if they are carried by individuals that have enough good alleles to result in an overall fitness benefit.

Furthermore, natural selection can be constrained by the relationships between different polymorphisms. One morph may confer a higher fitness than another, but may not increase in frequency due to the fact that going from the less beneficial to the more beneficial trait would require going through a less beneficial phenotype. Think back to the mice that live at the beach. Some are light-colored and blend in with the sand, while others are dark and blend in with the patches of grass. The dark-colored mice may be, overall, more fit than the light-colored mice, and at first glance, one might expect the light-colored mice be selected for a darker coloration. But remember that the intermediate phenotype, a medium-colored coat, is very bad for the mice—they cannot blend in with either the sand or the grass and are more likely to be eaten by predators. As a result, the light-colored mice would not be selected for a dark coloration because those individuals that began moving in that direction (began being selected for a darker coat) would be less fit than those that stayed light.

Finally, it is important to understand that not all evolution is adaptive. While natural selection selects the fittest individuals and often results in a more fit population overall, other forces of evolution, including genetic drift and gene flow, often do the opposite: introducing deleterious alleles to the population's gene pool. Evolution has no purpose—it is not changing a population into a preconceived ideal. It is simply the sum of the various forces described in this chapter and how they influence the genetic and phenotypic variance of a population.

In Summary: Selective and Environmental Pressures

Because natural selection acts to increase the frequency of beneficial alleles and traits while decreasing the frequency of deleterious qualities, it is adaptive evolution. Natural selection acts at the level of the individual, selecting for those that have a higher overall fitness compared to the rest of the population. If the fit phenotypes are those that are similar, natural selection will result in stabilizing selection, and an overall decrease in the population's variation. Directional selection works to shift a population's variance toward a new, fit phenotype, as environmental conditions change. In contrast, diversifying selection results in increased genetic variance by selecting for two or more distinct phenotypes.

Other types of selection include frequency-dependent selection, in which individuals with either common (positive frequency-dependent selection) or rare (negative frequency-dependent selection) phenotypes are selected for. Finally, sexual selection results from the fact that one sex has more variance in the reproductive success than the other. As a result, males and females experience different selective pressures, which can often lead to the evolution of phenotypic differences, or sexual dimorphisms, between the two.

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GENETIC VARIATION AND DRIFT

Learning Outcomes

Describe the different types of variation in a population

Individuals of a population often display different phenotypes, or express different alleles of a particular gene, referred to as polymorphisms. Populations with two or more variations of particular characteristics are called polymorphic. The distribution of phenotypes among individuals, known as the population variation, is influenced by a number of factors, including the population's genetic structure and the environment (Figure 1). Understanding the sources of a phenotypic variation in a population is important for determining how a population will evolve in response to different evolutionary pressures.

Figure 1. The distribution of phenotypes in this litter of kittens illustrates population variation. (credit: Pieter Lanser)

Genetic Variance

Natural selection and some of the other evolutionary forces can only act on heritable traits, namely an organism's genetic code. Because alleles are passed from parent to offspring, those that confer beneficial traits or behaviors may be selected for, while deleterious alleles may be selected against. Acquired traits, for the most part, are not heritable. For example, if an athlete works out in the gym every day, building up muscle strength, the athlete's offspring will not necessarily grow up to be a body builder. If there is a genetic basis for the ability to run fast, on the other hand, this may be passed to a child. Ultimately, **heritability** tells us how much phenotypic variation in a population is ulimately due to genetic differences as opposed to acquired differences.

The diversity of alleles and genotypes within a population is called **genetic variance**. When scientists are involved in the breeding of a species, such as with animals in zoos and nature preserves, they try to increase a population's genetic variance to preserve as much of the phenotypic diversity as they can. This also helps reduce the risks associated with **inbreeding**, the mating of closely related individuals, which can have the undesirable effect of bringing together deleterious recessive mutations that can cause abnormalities and susceptibility to disease.

In addition to natural selection, there are other evolutionary forces that could be in play: genetic drift, gene flow, mutation, nonrandom mating, and environmental variances.

Genetic Drift

The theory of natural selection stems from the observation that some individuals in a population are more likely to survive longer and have more offspring than others; thus, they will pass on more of their genes to the next generation. A big, powerful male gorilla, for example, is much more likely than a smaller, weaker one to become the population's silverback, the pack's leader who mates far more than the other males of the group. The pack leader will father more offspring, who share half of his genes, and are likely to also grow bigger and stronger like their father. Over time, the genes for bigger size will increase in frequency in the population, and the population

will, as a result, grow larger on average. That is, this would occur if this particular **selection pressure**, or driving selective force, were the only one acting on the population. In other examples, better camouflage or a stronger resistance to drought might pose a selection pressure.

Another way a population's allele and genotype frequencies can change is **genetic drift** (Figure 2), which is simply the effect of chance. By chance, some individuals will have more offspring than others—not due to an advantage conferred by some genetically-encoded trait, but just because one male happened to be in the right place at the right time (when the receptive female walked by) or because the other one happened to be in the wrong place at the wrong time (when a fox was hunting).

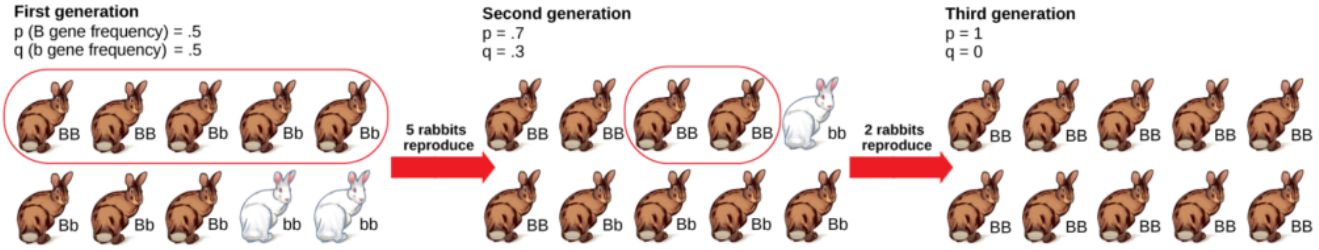

Figure 2. Click for a larger image. Genetic drift in a population can lead to the elimination of an allele from a population by chance. In this example, rabbits with the brown coat color allele (B) are dominant over rabbits with the white coat color allele (b). In the first generation, the two alleles occur with equal frequency in the population, resulting in p and q values of .5. Only half of the individuals reproduce, resulting in a second generation with p and q values of .7 and .3, respectively. Only two individuals in the second generation reproduce, and by chance these individuals are homozygous dominant for brown coat color. As a result, in the third generation the recessive b allele is lost.

Practice Question

Do you think genetic drift would happen more quickly on an island or on the mainland?

Answer

Genetic drift is likely to occur more rapidly on an island where smaller populations are expected to occur.

Small populations are more susceptible to the forces of genetic drift. Large populations, on the other hand, are buffered against the effects of chance. If one individual of a population of 10 individuals happens to die at a young age before it leaves any offspring to the next generation, all of its genes—1/10 of the population's gene pool—will be suddenly lost. In a population of 100, that's only 1 percent of the overall gene pool; therefore, it is much less impactful on the population's genetic structure.

Visit your course online to view an animation of random sampling and genetic drift in action.

Bottleneck Effect

Genetic drift can also be magnified by natural events, such as a natural disaster that kills—at random—a large portion of the population. Known as the **bottleneck effect**, it results in a large portion of the genome suddenly being wiped out (Figure 3). In one fell swoop, the genetic structure of the survivors becomes the genetic structure of the entire population, which may be very different from the pre-disaster population.

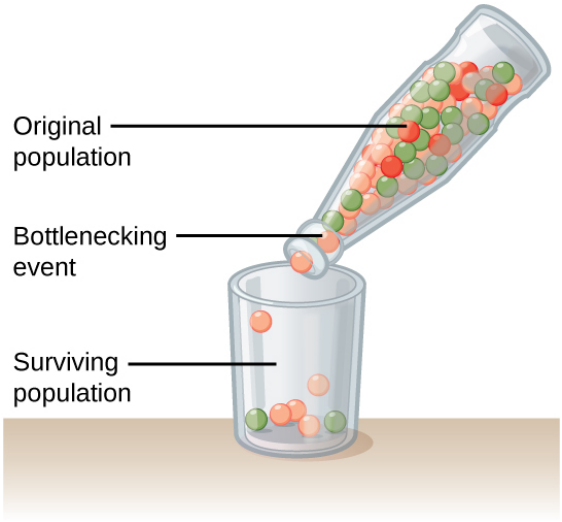

Figure 3. A chance event or catastrophe can reduce the genetic variability within a population.

Founder Effect

Another scenario in which populations might experience a strong influence of genetic drift is if some portion of the population leaves to start a new population in a new location or if a population gets divided by a physical barrier of some kind. In this situation, those individuals are unlikely to be representative of the entire population, which results in the founder effect. The **founder effect** occurs when the genetic structure changes to match that of the new population's founding fathers and mothers. The founder effect is believed to have been a key factor in the genetic history of the Afrikaner population of Dutch settlers in South Africa, as evidenced by mutations that are common in Afrikaners but rare in most other populations. This is likely due to the fact that a higher-than-normal proportion of the founding colonists carried these mutations. As a result, the population expresses unusually high incidences of Huntington's disease (HD) and Fanconi anemia (FA), a genetic disorder known to cause blood marrow and congenital abnormalities—even cancer.

Watch this short video to learn more about the founder and bottleneck effects. Note that the video has no audio.

Watch this video online: https://youtu.be/hEYV9WEvwaI

Gene Flow

Another important evolutionary force is **gene flow**: the flow of alleles in and out of a population due to the migration of individuals or gametes (Figure 4). While some populations are fairly stable, others experience more flux. Many plants, for example, send their pollen far and wide, by wind or by bird, to pollinate other populations of the same species some distance away. Even a population that may initially appear to be stable, such as a pride of lions, can experience its fair share of immigration and emigration as developing males leave their mothers to seek out a new pride with genetically unrelated females. This variable flow of individuals in and out of the group not only changes the gene structure of the population, but it can also introduce new genetic variation to populations in different geological locations and habitats.

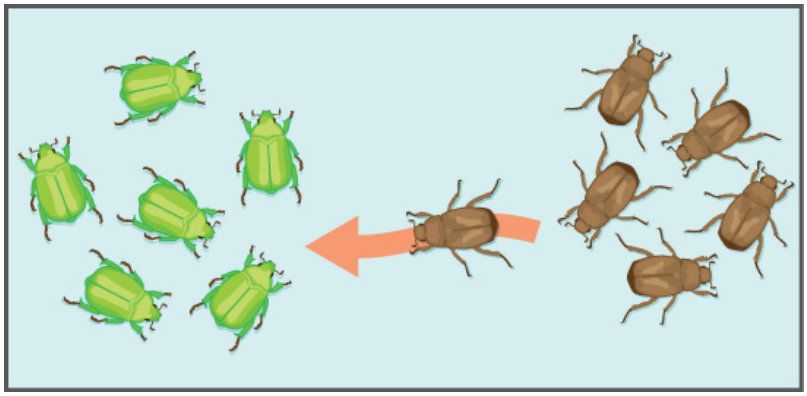

Figure 4. Gene flow can occur when an individual travels from one geographic location to another.

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INTRODUCTION TO PHYLOGENETIC TREES

What you'll learn to do: Read and analyze a phylogenetic tree that documents evolutionary relationships

In scientific terms, the evolutionary history and relationship of an organism or group of organisms is called phylogeny. **Phylogeny** describes the relationships of an organism, such as from which organisms it is thought to have evolved, to which species it is most closely related, and so forth.

Phylogenetic relationships provide information on shared ancestry but not necessarily on how organisms are similar or different. In other words, a "tree of life" can be constructed to illustrate when different organisms evolved and to show the relationships among different organisms (Figure 1).

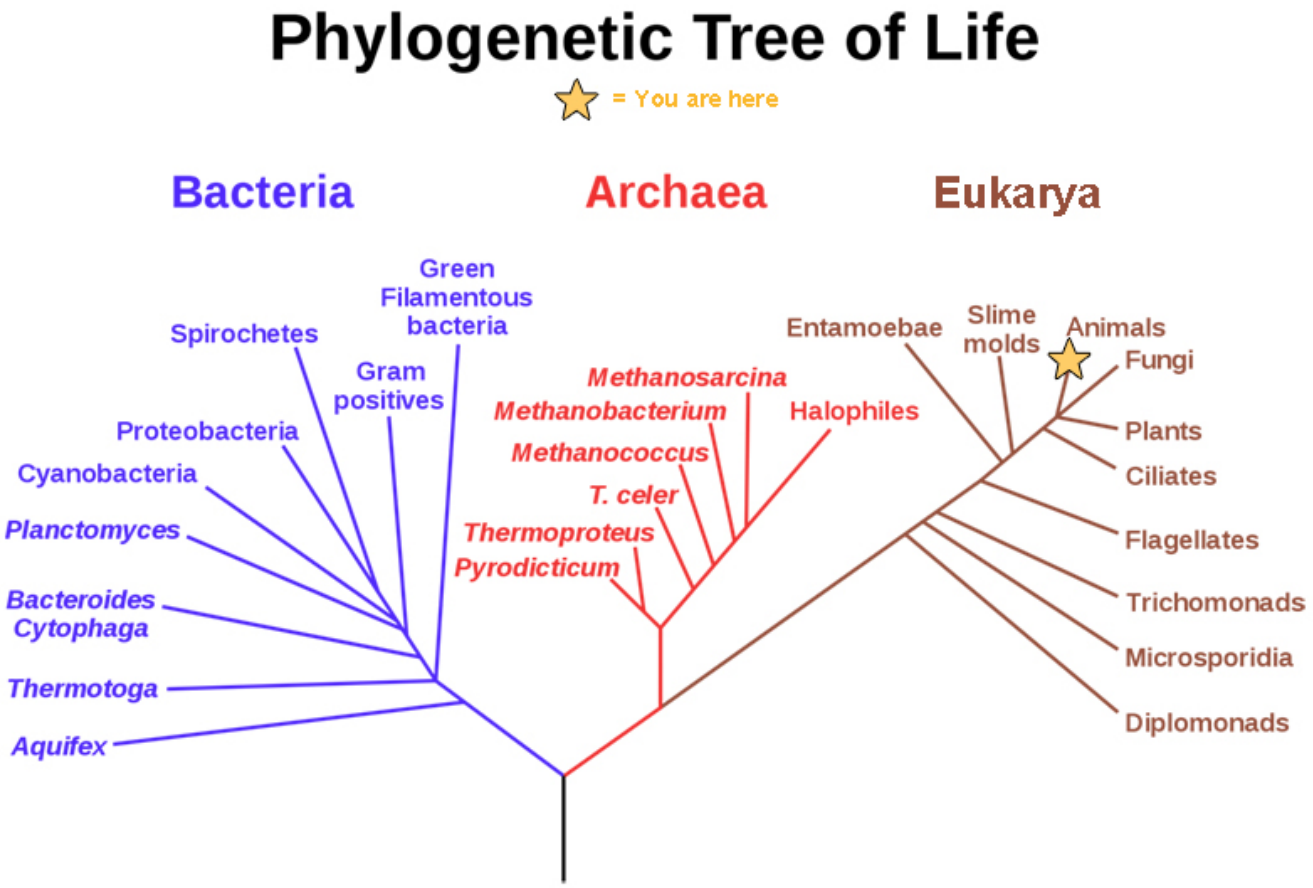

Figure 1. This phylogenetic tree was constructed by microbiologist Carl Woese (See inset below) using genetic relationships. The tree shows the separation of living organisms into three domains: Bacteria, Archaea, and Eukarya. Bacteria and Archaea are

organisms without a nucleus or other organelles surrounded by a membrane and, therefore, are prokaryotes. (credit: modification of work by Eric Gaba)

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SCIENTIFIC CLASSIFICATION

Learning Outcomes

Identify how and why scientists classify the organisms on earth

Why do biologists classify organisms? The major reason is to make sense of the incredible diversity of life on Earth. Scientists have identified millions of different species of organisms. Among animals, the most diverse group of organisms is the insects. More than one million different species of insects have already been described. An estimated nine million insect species have yet to be identified. A tiny fraction of insect species is shown in the beetle collection in Figure 1.

As diverse as insects are, there may be even more species of bacteria, another major group of organisms. Clearly, there is a need to organize the tremendous diversity of life. Classification allows scientists to organize and better understand the basic similarities and differences among organisms. This knowledge is necessary to understand the present diversity and the past evolutionary history of life on Earth.

Figure 1. Only a few of the more than one million known species of insects are represented in this beetle collection. Beetles are a major subgroup of insects. They make up about 40 percent of all insect species and about 25 percent of all known species of organisms.

Phylogenetic Trees

Scientists use a tool called a phylogenetic tree to show the evolutionary pathways and connections among organisms. A **phylogenetic tree** is a diagram used to reflect evolutionary relationships among organisms or groups of organisms. Scientists consider phylogenetic trees to be a hypothesis of the evolutionary past since one cannot go back to confirm the proposed relationships. In other words, a “tree of life” can be constructed to illustrate when different organisms evolved and to show the relationships among different organisms.

Each group of organisms went through its own evolutionary journey, called its phylogeny. Each organism shares relatedness with others, and based on morphologic and genetic evidence, scientists attempt to map the evolutionary pathways of all life on Earth. Many scientists build phylogenetic trees to illustrate evolutionary relationships.

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STRUCTURE OF PHYLOGENETIC TREES

Learning Outcomes

Differentiate between types of phylogenetic trees and what their structures tell us

A **phylogenetic tree** can be read like a map of evolutionary history. Many phylogenetic trees have a single lineage at the base representing a common ancestor. Scientists call such trees rooted, which means there is a single ancestral lineage (typically drawn from the bottom or left) to which all organisms represented in the diagram relate. Notice in the rooted phylogenetic tree that the three domains—Bacteria, Archaea, and Eukarya—diverge from a single point and branch off. The small branch that plants and animals (including humans) occupy in this diagram shows how recent and miniscule these groups are compared with other organisms. Unrooted trees don't show a common ancestor but do show relationships among species.

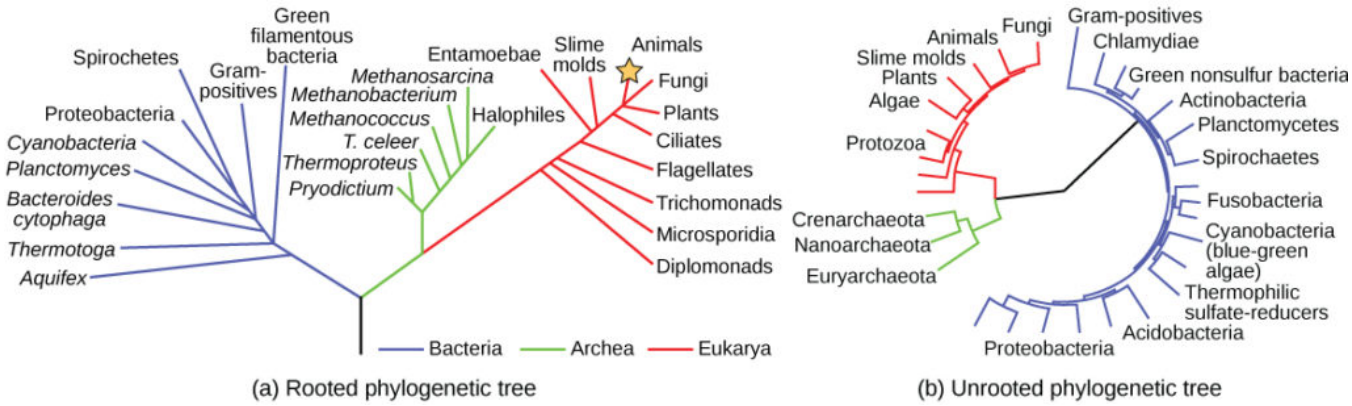

Figure 1. Both of these phylogenetic trees shows the relationship of the three domains of life—Bacteria, Archaea, and Eukarya—but the (a) rooted tree attempts to identify when various species diverged from a common ancestor while the (b) unrooted tree does not. (credit a: modification of work by Eric Gaba)

In a rooted tree, the branching indicates evolutionary relationships (Figure 2). The point where a split occurs, called a **branch point**, represents where a single lineage evolved into a distinct new one. A lineage that evolved early from the root and remains unbranched is called **basal taxon**. When two lineages stem from the same branch point, they are called **sister taxa**. A branch with more than two lineages is called a **polytomy** and serves to illustrate where scientists have not definitively determined all of the relationships. It is important to note that although sister taxa and polytomy do share an ancestor, it does not mean that the groups of organisms split or evolved from each other. Organisms in two taxa may have split apart at a specific branch point, but neither taxa gave rise to the other.

The diagrams above can serve as a pathway to understanding evolutionary history. The pathway can be traced from the origin of life to any individual species by navigating through the evolutionary branches between the two points. Also, by starting with a single species and tracing back towards the “trunk” of the tree, one can discover that species’ ancestors, as well as where lineages share a common ancestry. In addition, the tree can be used to study entire groups of organisms.

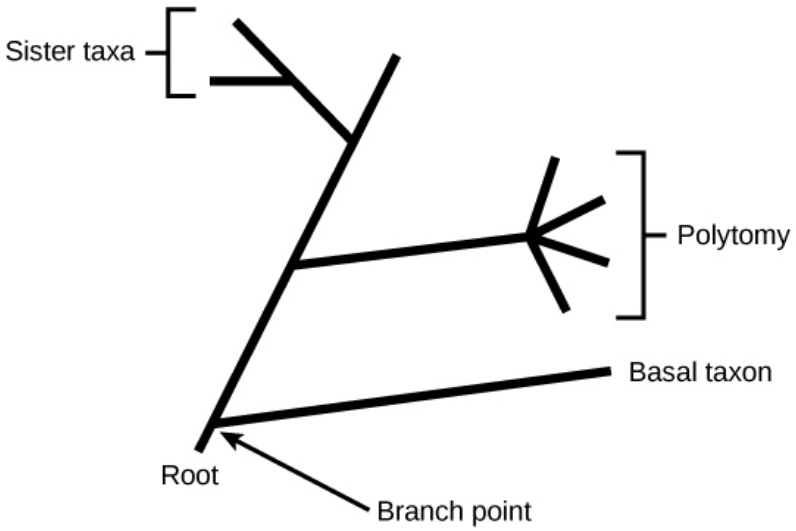

Figure 2. The root of a phylogenetic tree indicates that an ancestral lineage gave rise to all organisms on the tree. A branch point indicates where two lineages diverged. A lineage that evolved early and remains unbranched is a basal taxon. When two lineages stem from the same branch point, they are sister taxa. A branch with more than two lineages is a polytomy.

Many disciplines within the study of biology contribute to understanding how past and present life evolved over time; these disciplines together contribute to building, updating, and maintaining the “tree of life.” Information is used to organize and classify organisms based on evolutionary relationships in a scientific field called systematics. Data may be collected from fossils, from studying the structure of body parts or molecules used by an organism, and by DNA analysis. By combining data from many sources, scientists can put together the phylogeny of an organism; since phylogenetic trees are hypotheses, they will continue to change as new types of life are discovered and new information is learned.

Video Review

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LIMITATIONS OF PHYLOGENETIC TREES

Learning Outcomes

Identify some limitations of phylogenetic trees

It may be easy to assume that more closely related organisms look more alike, and while this is often the case, it is not always true. If two closely related lineages evolved under significantly varied surroundings or after the evolution of a major new adaptation, it is possible for the two groups to appear more different than other groups that are not as closely related. For example, the phylogenetic tree in Figure 1 shows that lizards and rabbits both have amniotic eggs, whereas frogs do not; yet lizards and frogs appear more similar than lizards and rabbits.

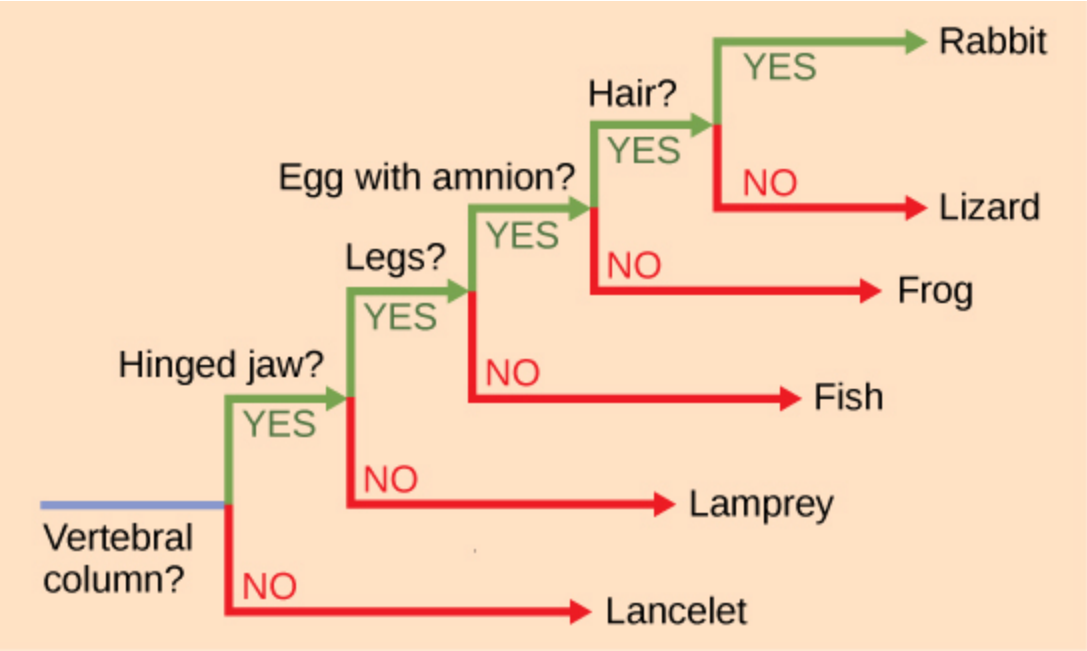

Figure 1. This ladder-like phylogenetic tree of vertebrates is rooted by an organism that lacked a vertebral column. At each branch point, organisms with different characters are placed in different groups based on the characteristics they share.

Another aspect of phylogenetic trees is that, unless otherwise indicated, the branches do not account for length of time, only the evolutionary order. In other words, the length of a branch does not typically mean more time passed, nor does a short branch mean less time passed— unless specified on the diagram. For example, in Figure 1, the tree does not indicate how much time passed between the evolution of amniotic eggs and hair. What the tree does show is the order in which things took place. Again using Figure 1, the tree shows that the oldest trait is the vertebral column, followed by hinged jaws, and so forth. Remember that any phylogenetic tree is a part of the greater whole, and like a real tree, it does not grow in only one direction after a new branch develops.

So, for the organisms in Figure 1, just because a vertebral column evolved does not mean that invertebrate evolution ceased, it only means that a new branch formed. Also, groups that are not closely related, but evolve under similar conditions, may appear more phenotypically similar to each other than to a close relative.

Head to this website to see interactive exercises that allow you to explore the evolutionary relationships among species.

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THE TAXONOMIC CLASSIFICATION SYSTEM

Learning Outcomes

Relate the taxonomic classification system and binomial nomenclature

Taxonomy (which literally means “arrangement law”) is the science of classifying organisms to construct internationally shared classification systems with each organism placed into more and more inclusive groupings. Think about how a grocery store is organized. One large space is divided into departments, such as produce, dairy, and meats. Then each department further divides into aisles, then each aisle into categories and brands, and then finally a single product. This organization from larger to smaller, more specific categories is called a hierarchical system.

The taxonomic classification system (also called the Linnaean system after its inventor, Carl Linnaeus, a Swedish botanist, zoologist, and physician) uses a hierarchical model. Moving from the point of origin, the groups become more specific, until one branch ends as a single species. For example, after the common beginning of all life, scientists divide organisms into three large categories called a domain: Bacteria, Archaea, and Eukarya. Within each domain is a second category called a **kingdom**. After kingdoms, the subsequent categories of increasing specificity are: **phylum**, **class**, **order**, **family**, **genus**, and **species** (Figure 1).

The kingdom Animalia stems from the Eukarya domain. For the common dog, the classification levels would be as shown in Figure 1. Therefore, the full name of an organism technically has eight terms. For the dog, it is: Eukarya, Animalia, Chordata, Mammalia, Carnivora, Canidae, *Canis,* and *lupus*. Notice that each name is capitalized except for species, and the genus and species names are italicized. Scientists generally refer to an organism only by its genus and species, which is its two-word scientific name, in what is called **binomial nomenclature**. Therefore, the scientific name of the dog is *Canis lupus*. The name at each level is also called a **taxon**. In other words, dogs are in order Carnivora. Carnivora is the name of the taxon at the order level; Canidae is the taxon at the family level, and so forth. Organisms also have a common name that people typically use, in this case, dog. Note that the dog is additionally a subspecies: the “*familiaris*” in *Canis lupus familiaris.* Subspecies are members of the same species that are capable of mating and reproducing viable offspring, but they are considered separate subspecies due to geographic or behavioral isolation or other factors.

Figure 2 shows how the levels move toward specificity with other organisms. Notice how the dog shares a domain with the widest diversity of organisms, including plants and butterflies. At each sublevel, the organisms become more similar because they are more closely related. Historically, scientists classified organisms using characteristics, but as DNA technology developed, more precise phylogenies have been determined.

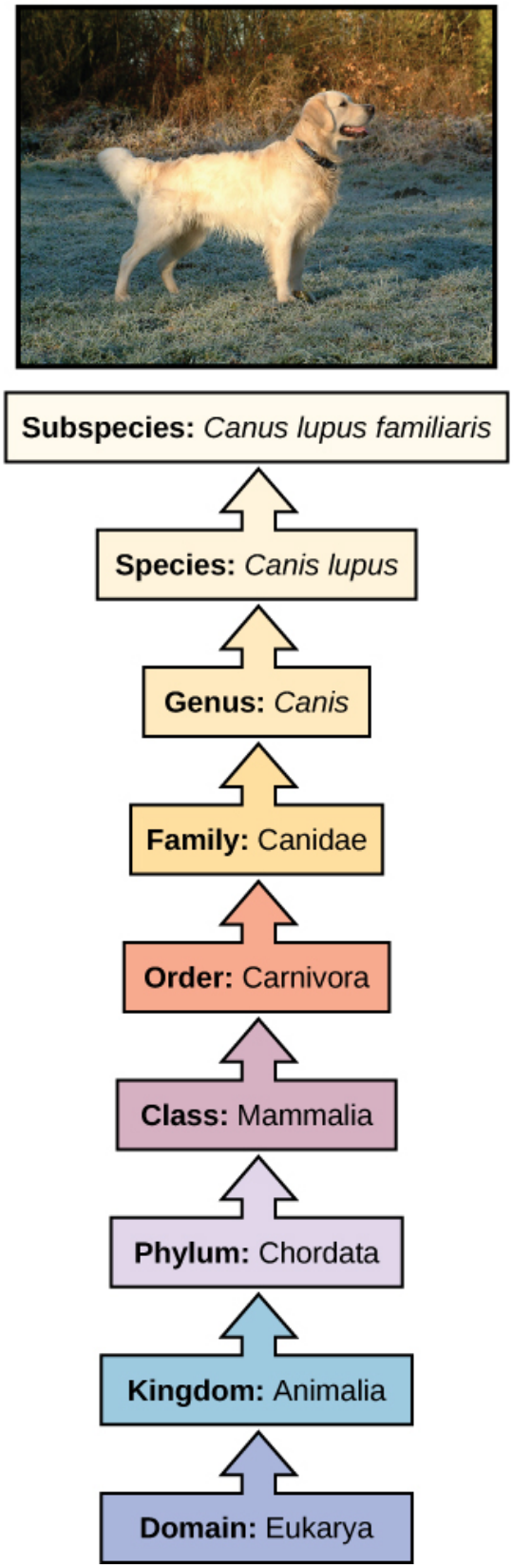

Figure 1. The taxonomic classification system uses a hierarchical model to organize living organisms into increasingly specific categories. The common dog, Canis lupus familiaris, is a subspecies of Canis lupus, which also includes the wolf and dingo. (credit "dog": modification of work by Janneke Vreugdenhil)

Practice Question

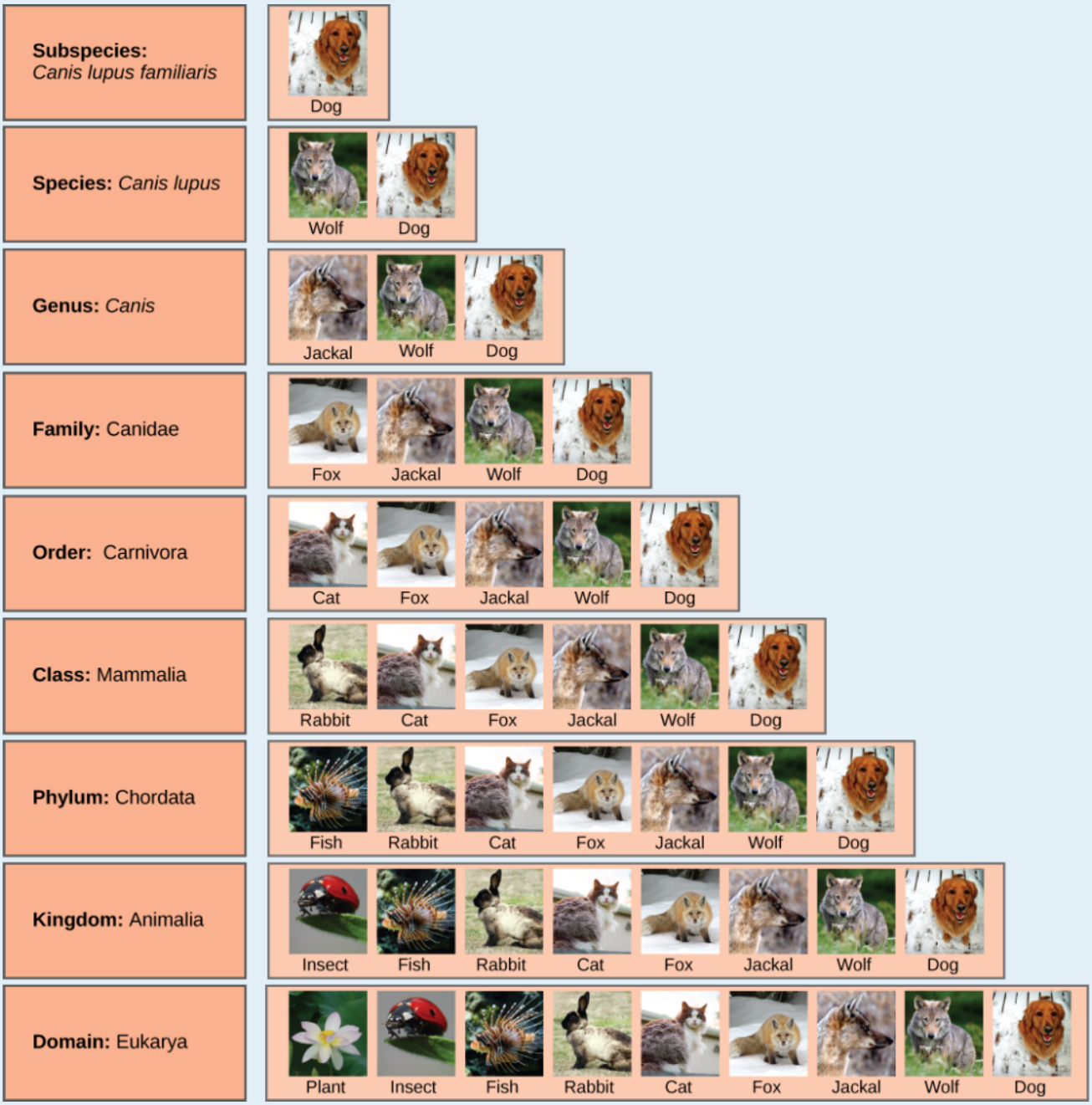

Figure 2. At each sublevel in the taxonomic classification system, organisms become more similar. Dogs and wolves are the same species because they can breed and produce viable offspring, but they are different enough to be classified as different subspecies. (credit "plant": modification of work by "berduchwal"/Flickr; credit "insect": modification of work by Jon Sullivan; credit "fish": modification of work by Christian Mehlführer; credit "rabbit": modification of work by Aidan Wojtas; credit "cat": modification of work by Jonathan Lidbeck; credit "fox": modification of work by Kevin Bacher, NPS; credit "jackal": modification of work by Thomas A. Hermann, NBII, USGS; credit "wolf": modification of work by Robert Dewar; credit "dog": modification of work by "digital_image_fan"/Flickr)

At what levels are cats and dogs considered to be part of the same group?

Answer

Cats and dogs are part of the same group at five levels: both are in the domain Eukarya, the kingdom Animalia, the phylum Chordata, the class Mammalia, and the order Carnivora.

Visit this website to classify three organisms—bear, orchid, and sea cucumber—from kingdom to species. To launch the game, under Classifying Life, click the picture of the bear or the Launch Interactive button.

Recent genetic analysis and other advancements have found that some earlier phylogenetic classifications do not align with the evolutionary past; therefore, changes and updates must be made as new discoveries occur. Recall that phylogenetic trees are hypotheses and are modified as data becomes available. In addition, classification historically has focused on grouping organisms mainly by shared characteristics and does not necessarily illustrate how the various groups relate to each other from an evolutionary perspective. For example, despite the fact that a hippopotamus resembles a pig more than a whale, the hippopotamus may be the closest living relative of the whale.

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PUTTING IT TOGETHER: THEORY OF EVOLUTION

Let's return to our discussion of evolving bacteria from the beginning of the chapter. The World Health Organization (WHO) suggests that based on current evidence, gonorrhea may soon be untreatable; there are no new treatments or vaccinations in development for this bacteria. Within the United States this remain the second most common sexually transmitted disease. Almost a third of the US cases are drug resistant. MRSA remains a growing problem in our healthcare system. Staph bacteria are the most common source of health-care related infections in the United States. Of MRSA infections, nearly 14% result in death.

Think about It

Based on this lesson you now understand that evolution is an inevitable process. Is there any hope of stopping antibiotic resistance from developing?

Our Thoughts

Yes and no. The average person can take a number of steps to minimize selective pressures on parasites. Without these pressures, mutations are less likely to spread in the population.

Think about It

Is there anything you can do?

Our Thoughts

- Take the full course of prescribed antibiotics
- Don’t save or share leftover antibiotics
- Don’t ask for antibiotics if your doctor does not recommend them
- Practice good hygiene and get recommended vaccinations
- Minimize your use of antibacterial products

To learn more about the topic, you can visit the following websites:

- WHO—Antimicrobial Resistance
- CDC—Antibiotic Resistance Threats
- CDC—Mission Critical: Preventing Antibiotic Resistance
- FDA—Antibacterial Soap

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MODULE 13: MODERN BIOLOGY

WHY IT MATTERS: MODERN BIOLOGY

Why learn about techniques used in modern biology?

Biology has impacted nearly every facet of human life. The entire field of medicine is based on biological discovery and exploration; without knowledge of how the human body functions, we would have no idea how to fix our bodies when something goes wrong.

Over the decades, new techniques have been developed to study biology. Scientists recently mapped the complete human genome. New technologies were developed to help speed this process up. Forensic science has also been vastly influenced by biological discovery. New techniques enable law enforcement to gather DNA evidence at crime scenes, which helps ensure they identify the right suspect. DNA based information can be applied to many such situations. Consider the following scenario.

On Memorial Day 1984, the remains of a soldier killed during the Vietnam War were laid to rest in the Tomb of the Unknown at Arlington National Cemetery. A decade later, however, questions began to arise about the identity of this unknown soldier. In particular, people began to wonder if, due to scientific improvement and developments, the soldier could be identified.

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INTRODUCTION TO KEY TECHNOLOGIES

What you'll learn to do: List key technologies enabling modern uses of biology

Biotechnology is the use of biological agents for technological advancement. Biotechnology was used for breeding livestock and crops long before the scientific basis of these techniques was understood. Since the discovery of the structure of DNA in 1953, the field of biotechnology has grown rapidly through both academic research and private companies. The primary applications of this technology are in medicine (production of vaccines and antibiotics) and agriculture (genetic modification of crops, such as to increase nutrient content). Biotechnology also has many industrial applications, such as fermentation, the treatment of oil spills, and the production of biofuels (Figure 1).

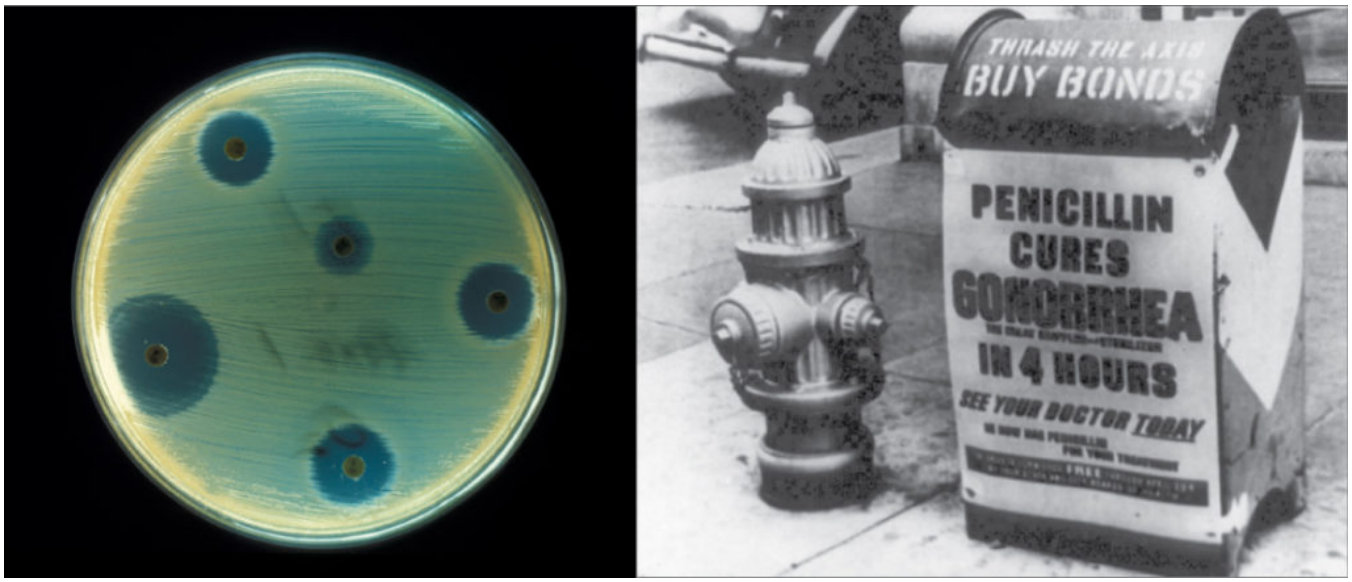

Figure 1. Antibiotics are chemicals produced by fungi, bacteria, and other organisms that have antimicrobial properties. The first antibiotic discovered was penicillin. Antibiotics are now commercially produced and tested for their potential to inhibit bacterial growth. (credit "advertisement": modification of work by NIH; credit "test plate": modification of work by Don Stalons/CDC; scale-bar data from Matt Russell)

In this outcome, we will learn about some modern technologies used in biology today.

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MANIPULATING GENETIC MATERIAL

Learning Outcomes

List basic techniques to manipulation genetic information (DNA and RNA)

To understand the basic techniques used to work with nucleic acids, remember that nucleic acids are macromolecules made of nucleotides (a sugar, a phosphate, and a nitrogenous base) linked by phosphodiester bonds. The phosphate groups on these molecules each have a net negative charge. An entire set of DNA molecules in the nucleus is called the **genome**. DNA has two complementary strands linked by hydrogen bonds between the paired bases. The two strands can be separated by exposure to high temperatures (DNA denaturation) and can be reannealed by cooling. The DNA can be replicated by the DNA polymerase enzyme. Unlike DNA, which is located in the nucleus of eukaryotic cells, RNA molecules leave the nucleus. The most common type of RNA that is analyzed is the messenger RNA (mRNA) because it represents the protein-coding genes that are actively expressed. However, RNA molecules present some other challenges to analysis, as they are often less stable than DNA.

DNA and RNA Extraction

To study or manipulate nucleic acids, the DNA or RNA must first be isolated or extracted from the cells. Various techniques are used to extract different types of DNA (Figure 1). Most nucleic acid extraction techniques involve steps to break open the cell and use enzymatic reactions to destroy all macromolecules that are not desired (such as degradation of unwanted molecules and separation from the DNA sample). Cells are broken using a **lysis buffer** (a solution which is mostly a detergent); lysis means "to split." These enzymes break apart lipid molecules in the cell membranes and nuclear membranes. Macromolecules are inactivated using enzymes such as **proteases** that break down proteins, and **ribonucleases** (RNAses) that break down RNA. The DNA is then precipitated using alcohol. Human genomic DNA is usually visible as a gelatinous, white mass. The DNA samples can be stored frozen at –80°C for several years.

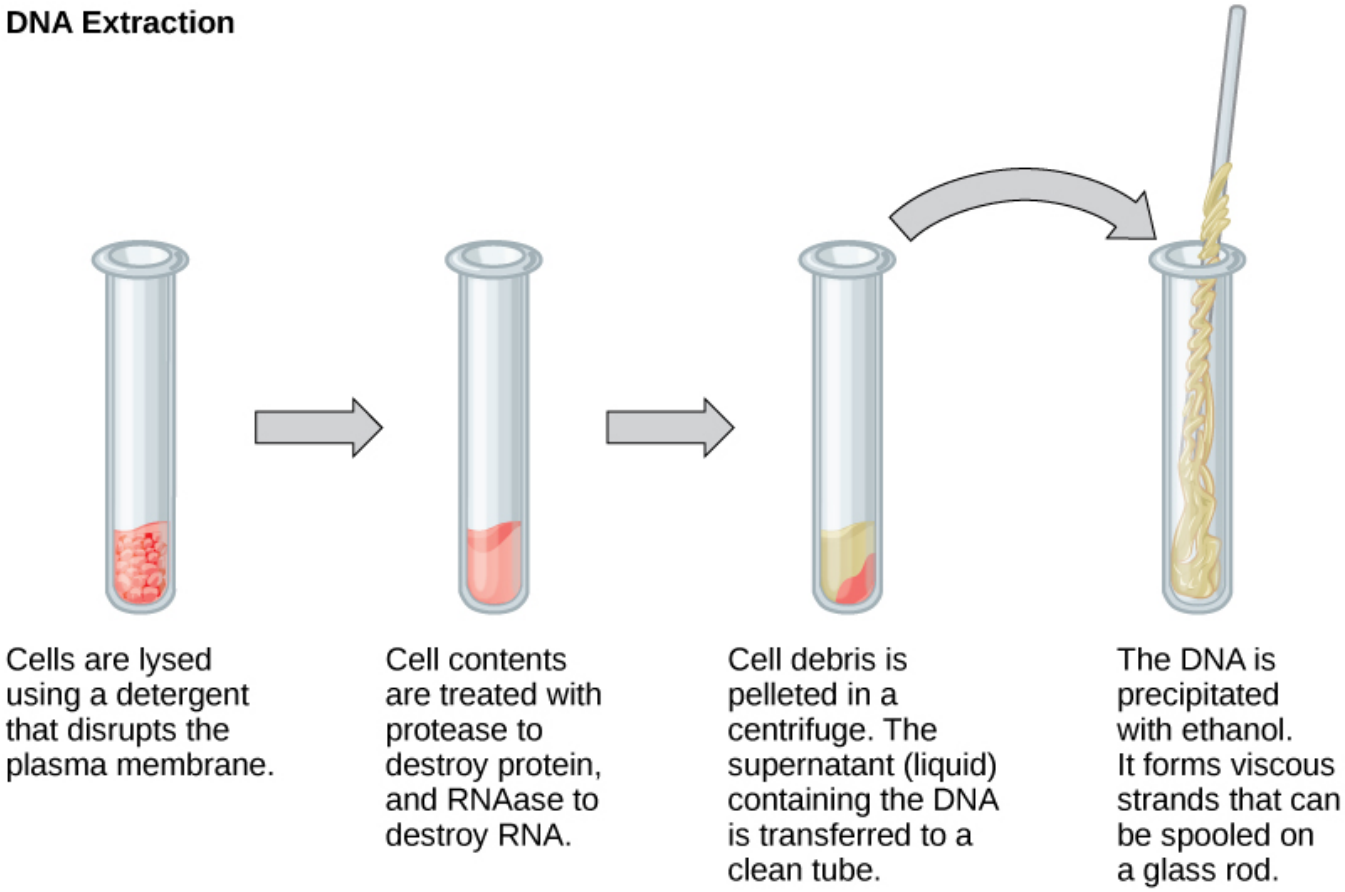

Figure 1. This diagram shows the basic method used for extraction of DNA.

RNA analysis is performed to study gene expression patterns in cells. RNA is naturally very unstable because RNAses are commonly present in nature and very difficult to inactivate. Similar to DNA, RNA extraction involves the use of various buffers and enzymes to inactivate macromolecules and preserve the RNA.

Gel Electrophoresis

Because nucleic acids are negatively charged ions at neutral or basic pH in an aqueous environment, they can be mobilized by an electric field. **Gel electrophoresis** is a technique used to separate molecules on the basis of size, using this charge. The nucleic acids can be separated as whole chromosomes or fragments. The nucleic acids are loaded into a slot near the negative electrode of a semisolid, porous gel matrix and pushed toward the positive electrode at the opposite end of the gel. Smaller molecules move through the pores in the gel faster than larger molecules; this difference in the rate of migration separates the fragments on the basis of size. There are molecular weight standard samples that can be run alongside the molecules to provide a size comparison. Nucleic acids in a gel matrix can be observed using various fluorescent or colored dyes. Distinct nucleic acid fragments appear as bands at specific distances from the top of the gel (the negative electrode end) on the basis of their size (Figure 2). A mixture of genomic DNA fragments of varying sizes appear as a long smear, whereas uncut genomic DNA is usually too large to run through the gel and forms a single large band at the top of the gel.

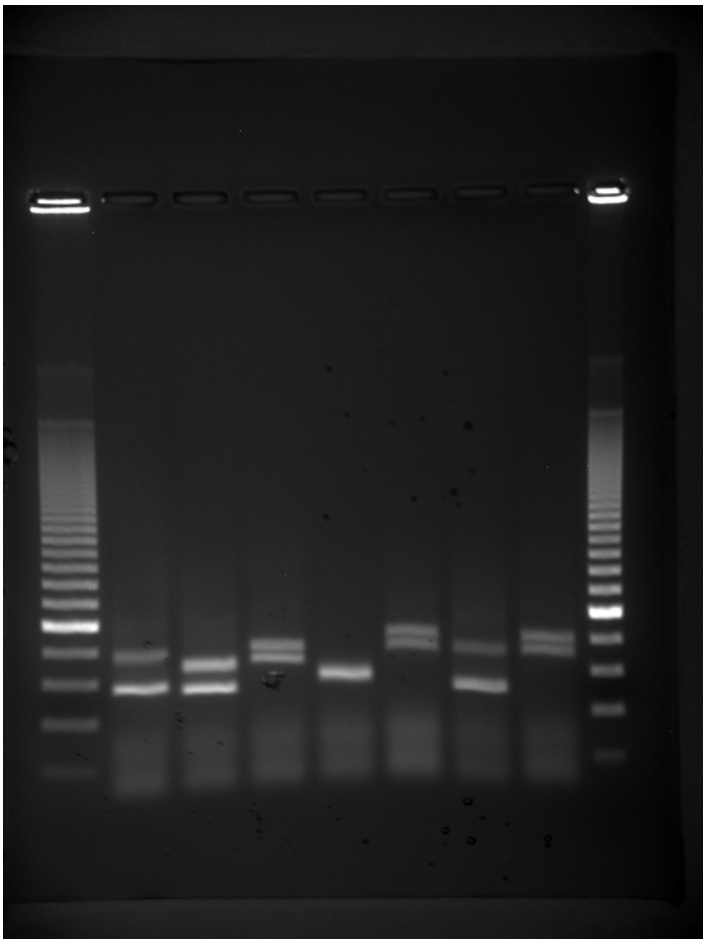

Figure 2. Shown are DNA fragments from seven samples run on a gel, stained with a fluorescent dye, and viewed under UV light. (credit: James Jacob, Tompkins Cortland Community College)

Amplification of Nucleic Acid Fragments by Polymerase Chain Reaction

Although genomic DNA is visible to the naked eye when it is extracted in bulk, DNA analysis often requires focusing on one or more specific regions of the genome. **Polymerase chain reaction (PCR)** is a technique used to amplify specific regions of DNA for further analysis (Figure 3). PCR is used for many purposes in laboratories, such as the cloning of gene fragments to analyze genetic diseases, identification of contaminant foreign DNA in a sample, and the amplification of DNA for sequencing. More practical applications include the determination of paternity and detection of genetic diseases.

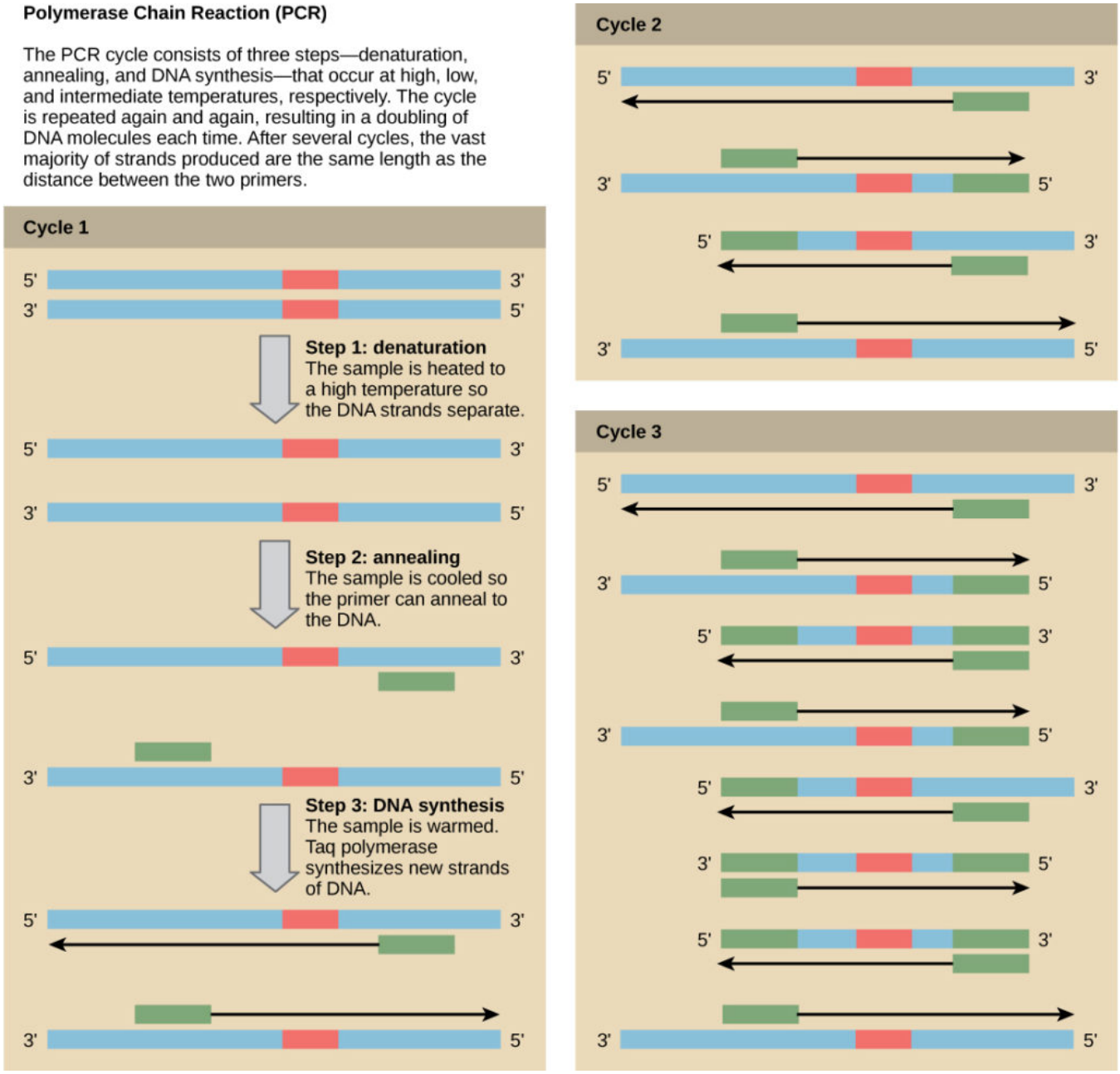

Figure 3. Polymerase chain reaction, or PCR, is used to amplify a specific sequence of DNA. Primers—short pieces of DNA complementary to each end of the target sequence—are combined with genomic DNA, Taq polymerase, and deoxynucleotides. Taq polymerase is a DNA polymerase isolated from the thermostable bacterium Thermus aquaticus that is able to withstand the high temperatures used in PCR. Thermus aquaticus grows naturally in the Lower Geyser Basin of Yellowstone National Park. Reverse transcriptase PCR (RT-PCR) is similar to PCR, but cDNA is made from an RNA template before PCR begins.

DNA fragments can also be amplified from an RNA template in a process called reverse transcriptase PCR (RT-PCR). The first step is to recreate the original DNA template strand (called cDNA) by applying DNA nucleotides to the mRNA. This process is called reverse transcription. This requires the presence of an enzyme called reverse transcriptase. After the cDNA is made, regular PCR can be used to amplify it.

Deepen your understanding of the polymerase chain reaction by clicking through this interactive exercise.

Hybridization, Southern Blotting, and Northern Blotting

Nucleic acid samples, such as fragmented genomic DNA and RNA extracts, can be probed for the presence of certain sequences. Short DNA fragments called **probes** are designed and labeled with radioactive or fluorescent dyes to aid detection. Gel electrophoresis separates the nucleic acid fragments according to their size. The fragments in the gel are then transferred onto a nylon membrane in a procedure called **blotting** (Figure 4). The nucleic acid fragments that are bound to the surface of the membrane can then be probed with specific radioactively or fluorescently labeled probe sequences. When DNA is transferred to a nylon membrane, the technique is called **Southern blotting**, and when RNA is transferred to a nylon membrane, it is called **northern blotting**. Southern blots are used to detect the presence of certain DNA sequences in a given genome, and northern blots are used to detect gene expression. Note that "Southern" blotting is capitalized but no other type of blotting; Southern blotting is named after the scientist who pioneered this technique, Edwin Southern. The other types of blotting were named in reference to the original Southern blot.

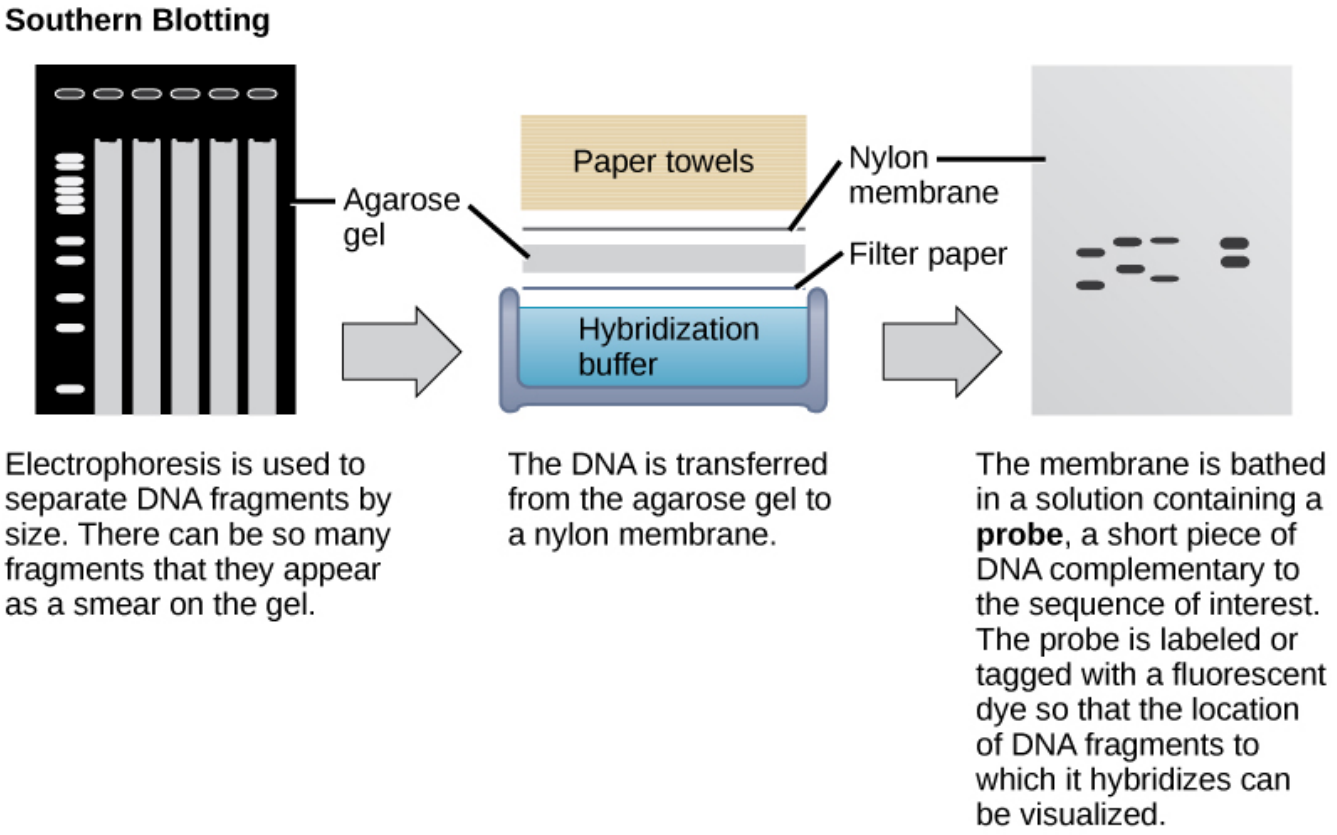

Figure 4. Southern blotting is used to find a particular sequence in a sample of DNA. DNA fragments are separated on a gel, transferred to a nylon membrane, and incubated with a DNA probe complementary to the sequence of interest. Northern blotting is similar to Southern blotting, but RNA is run on the gel instead of DNA. In western blotting, proteins are run on a gel and detected using antibodies.

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DNA SEQUENCING

Learning Outcomes

Describe the most widely used DNA sequencing method

Until the 1990s, the sequencing of DNA (reading the sequence of DNA) was a relatively expensive and long process. Using radiolabeled nucleotides also compounded the problem through safety concerns. With currently available technology and automated machines, the process is cheap, safer, and can be completed in a matter of hours. Fred Sanger developed the sequencing method used for the human genome sequencing project, which is widely used today (Figure 1).

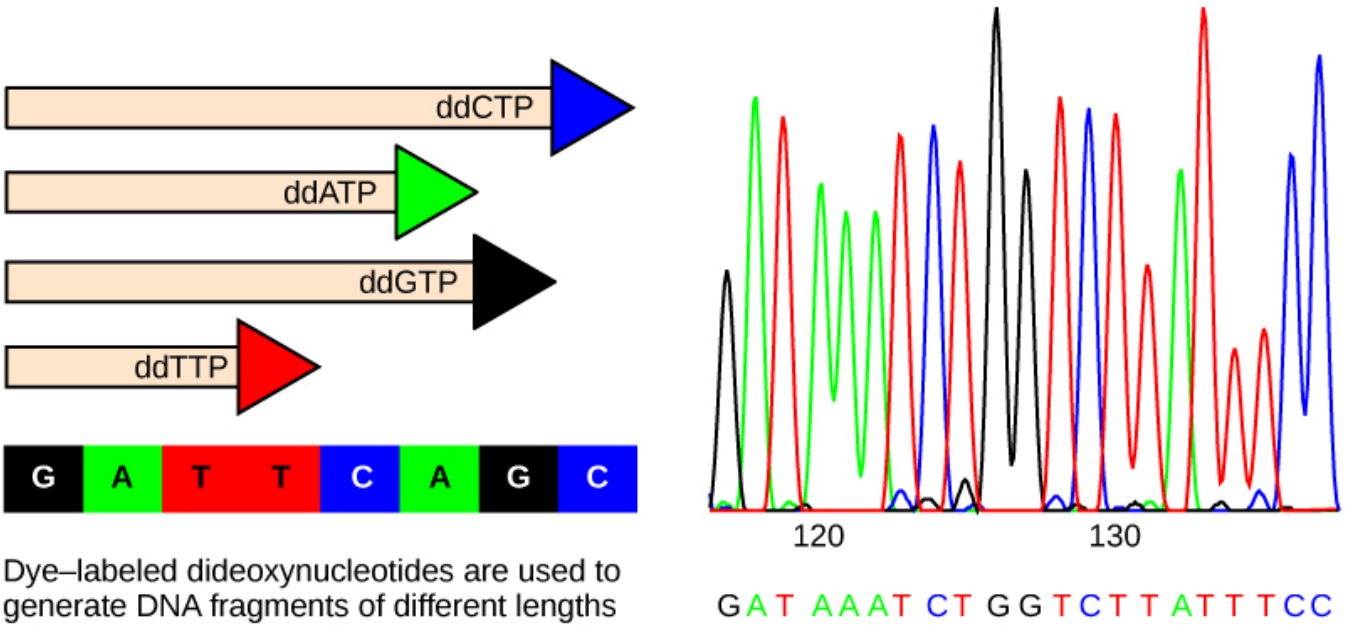

Figure 1. In Frederick Sanger's dideoxy chain termination method, dye-labeled dideoxynucleotides are used to generate DNA fragments that terminate at different points. The DNA is separated by capillary electrophoresis on the basis of size, and from the order of fragments formed, the DNA sequence can be read. The DNA sequence readout is shown on an electropherogram that is generated by a laser scanner.

Visit this site to watch a video explaining the DNA sequence reading technique that resulted from Sanger's work.

The method is known as the dideoxy chain termination method. The sequencing method is based on the use of chain terminators, the dideoxynucleotides (ddNTPs). The dideoxynucleotides, or ddNTPSs, differ from the deoxynucleotides by the lack of a free 3′ OH group on the five-carbon sugar. If a ddNTP is added to a growing a DNA strand, the chain is not extended any further because the free 3′ OH group needed to add another nucleotide is not available. By using a predetermined ratio of deoxyribonucleotides to dideoxynucleotides, it is possible to generate DNA fragments of different sizes.

The DNA sample to be sequenced is denatured or separated into two strands by heating it to high temperatures. The DNA is divided into four tubes in which a primer, DNA polymerase, and all four nucleotides (A, T, G, and C) are added. In addition to each of the four tubes, limited quantities of one of the four dideoxynucleotides are added to each tube respectively. The tubes are labeled as A, T, G, and C according to the ddNTP added. For detection purposes, each of the four dideoxynucleotides carries a different fluorescent label. Chain elongation continues until a fluorescent dideoxy nucleotide is incorporated, after which no further elongation takes place. After the reaction is over, electrophoresis is performed. Even a difference in length of a single base can be detected. The sequence is read from a laser scanner. For his work on DNA sequencing, Sanger received a Nobel Prize in chemistry in 1980.

As already mentioned, gel **electrophoresis** (Figure 2) is a technique used to separate DNA fragments of different sizes. Usually the gel is made of a chemical called agarose. Agarose powder is added to a buffer and heated. After cooling, the gel solution is poured into a casting tray. Once the gel has solidified, the DNA is loaded on the gel and electric current is applied. The DNA has a net negative charge and moves from the negative electrode toward the positive electrode. The electric current is applied for sufficient time to let the DNA separate according to size; the smallest fragments will be farthest from the well (where the DNA was loaded), and the heavier molecular weight fragments will be closest to the well. Once the DNA is separated, the gel is stained with a DNA-specific dye for viewing it.

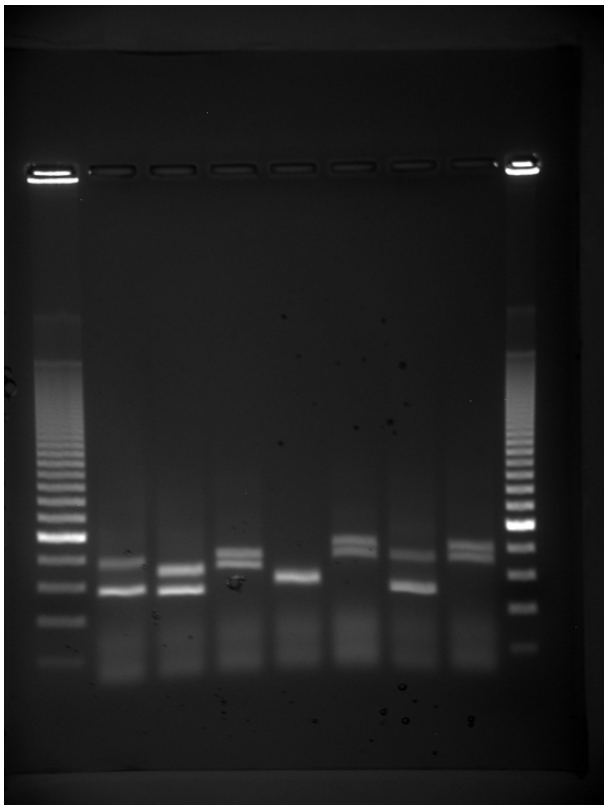

Figure 2. DNA can be separated on the basis of size using gel electrophoresis. (credit: James Jacob, Tompkins Cortland Community College)

Sanger's genome sequencing has led to a race to sequence human genomes at a rapid speed and low cost, often referred to as the $1000 in one day sequence. Learn more by watching the Sequencing at Speed animation here.

Neanderthal Genome: How Are We Related?

The first draft sequence of the Neanderthal genome was published by Richard E. Green et al. in 2010. (Note: Richard E. Green et al., "A Draft Sequence of the Neandertal Genome," *Science* 328 (2010): 710–22.) Neanderthals are the closest ancestors of present-day humans. They were known to have lived in Europe and Western Asia before they disappeared from fossil records approximately 30,000 years ago. Green's team studied almost 40,000-year-old fossil remains that were selected from sites across the world. Extremely sophisticated means of sample preparation and DNA sequencing were employed because of the fragile nature of the bones and heavy microbial contamination. In their study, the scientists were able to sequence some four billion base pairs. The Neanderthal sequence was compared with that of present-day humans from across the world. After comparing the sequences, the researchers found that the Neanderthal genome had 2 to 3 percent greater similarity to people living outside Africa than to people in Africa. While current theories have suggested that all present-day humans can be traced to a small ancestral population in Africa, the data from the Neanderthal genome may contradict this view. Green and his colleagues also discovered DNA segments among people in Europe and Asia that are more similar to Neanderthal sequences than to other contemporary human sequences. Another interesting observation was that Neanderthals are as closely related to people from Papua New Guinea as to those from China or France. This is surprising because Neanderthal fossil remains have been located only in Europe and West Asia. Most likely, genetic

exchange took place between Neanderthals and modern humans as modern humans emerged out of Africa, before the divergence of Europeans, East Asians, and Papua New Guineans.

Several genes seem to have undergone changes from Neanderthals during the evolution of present-day humans. These genes are involved in cranial structure, metabolism, skin morphology, and cognitive development. One of the genes that is of particular interest is *RUNX2*, which is different in modern day humans and Neanderthals. This gene is responsible for the prominent frontal bone, bell-shaped rib cage, and dental differences seen in Neanderthals. It is speculated that an evolutionary change in *RUNX2* was important in the origin of modern-day humans, and this affected the cranium and the upper body.

Watch Svante Pääbo's talk explaining the Neanderthal genome research at the 2011 annual TED (Technology, Entertainment, Design) conference.

Watch this video online: https://youtu.be/kU0ei9ApmsY

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CLONING

Learning Outcomes

Recognize technologies used for molecular, cellular, and reproductive cloning

Molecular Cloning

In general, the word "cloning" means the creation of a perfect replica; however, in biology, the re-creation of a whole organism is referred to as "reproductive cloning." Long before attempts were made to clone an entire organism, researchers learned how to reproduce desired regions or fragments of the genome, a process that is referred to as **molecular cloning**.

Cloning small fragments of the genome allows for the manipulation and study of specific genes (and their protein products), or noncoding regions in isolation. A **plasmid** (also called a **vector**) is a small circular DNA molecule that replicates independently of the chromosomal DNA of microorganisms such as *E. coli*. In cloning, the plasmid molecules can be used to provide a "folder" in which to insert a desired DNA fragment. Plasmids are usually introduced into a bacterial host for proliferation. In the bacterial context, the fragment of DNA from the human genome (or the genome of another organism that is being studied) is referred to as **foreign DNA**, or a transgene, to differentiate it from the DNA of the bacterium, which is called the **host DNA**.

Plasmids occur naturally in bacterial populations (such as *Escherichia coli*) and have genes that can contribute favorable traits to the organism, such as **antibiotic resistance** (the ability to be unaffected by antibiotics). Plasmids have been repurposed and engineered as vectors for molecular cloning and the large-scale production of important reagents, such as **insulin** and **human growth hormone**. An important feature of plasmid vectors is the ease with which a foreign DNA fragment can be introduced via the **multiple cloning site (MCS)**. The MCS is a short DNA sequence containing multiple sites that can be cut with different commonly available restriction endonucleases. **Restriction endonucleases** recognize specific DNA sequences and cut them in a predictable manner; they are naturally produced by bacteria as a defense mechanism against foreign DNA. Many restriction

endonucleases make staggered cuts in the two strands of DNA, such that the cut ends have a 2- or 4-base single-stranded overhang. Because these overhangs are capable of annealing with complementary overhangs, these are called “sticky ends.” Addition of an enzyme called DNA ligase permanently joins the DNA fragments via phosphodiester bonds. In this way, any DNA fragment generated by restriction endonuclease cleavage can be spliced between the two ends of a plasmid DNA that has been cut with the same restriction endonuclease (Figure 1).

Recombinant DNA Molecules

Plasmids with foreign DNA inserted into them are called **recombinant DNA** molecules because they are created artificially and do not occur in nature. They are also called chimeric molecules because the origin of different parts of the molecules can be traced back to different species of biological organisms or even to chemical synthesis. Proteins that are expressed from recombinant DNA molecules are called **recombinant proteins**. Not all recombinant plasmids are capable of expressing genes. The recombinant DNA may need to be moved into a different vector (or host) that is better designed for gene expression. Plasmids may also be engineered to express proteins only when stimulated by certain environmental factors, so that scientists can control the expression of the recombinant proteins.

View an animation of recombination in cloning from the DNA Learning Center.

Practice Question

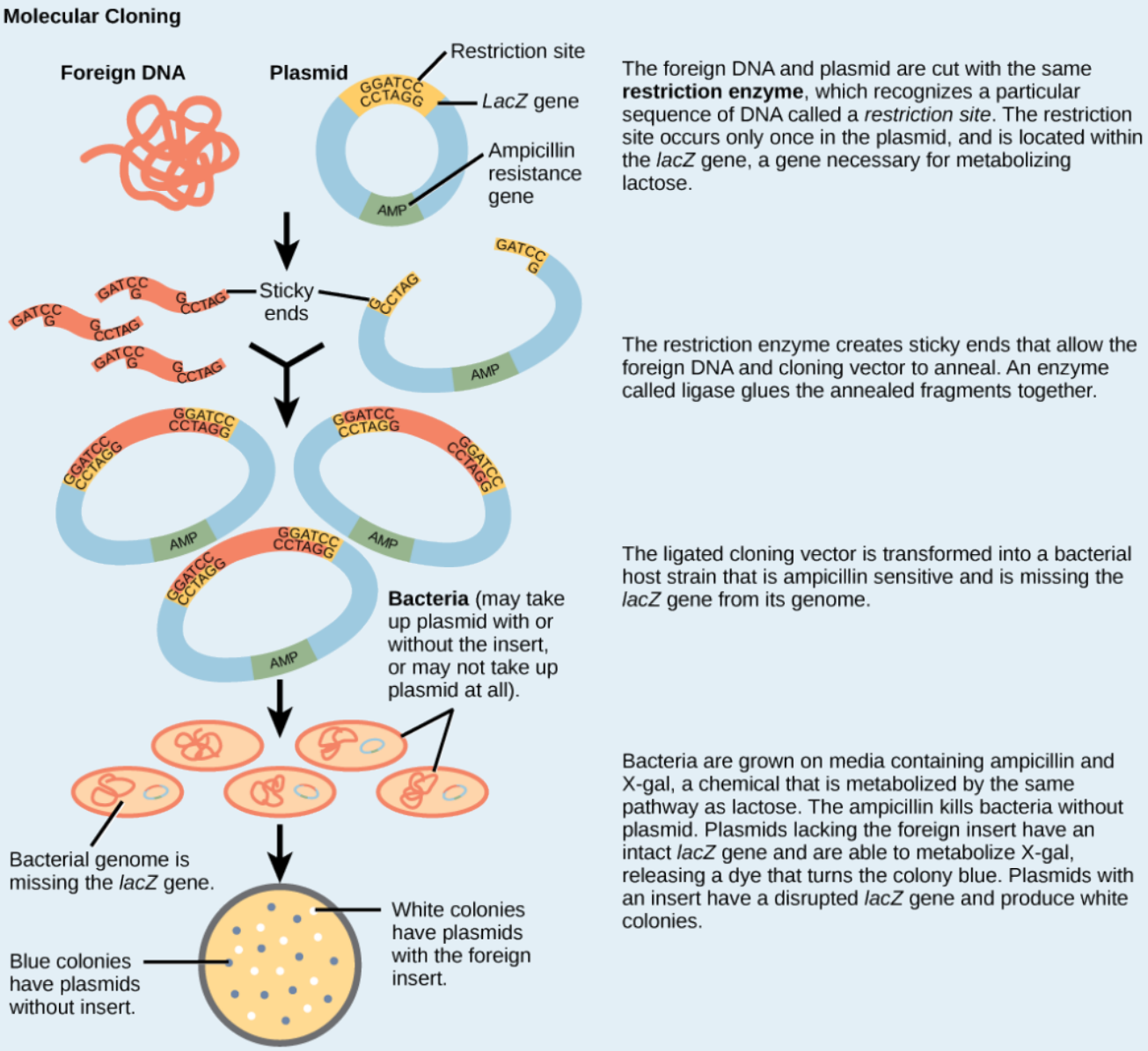

Figure 1. This diagram shows the steps involved in molecular cloning.

You are working in a molecular biology lab and, unbeknownst to you, your lab partner left the foreign genomic DNA that you are planning to clone on the lab bench overnight instead of storing it in the freezer. As a result, it was degraded by nucleases, but still used in the experiment. The plasmid, on the other hand, is fine. What results would you expect from your molecular cloning experiment?

a. There will be no colonies on the bacterial plate.
b. There will be blue colonies only.
c. There will be blue and white colonies.
d. The will be white colonies only.

Answer

Answer b. The experiment would result in blue colonies only.

Cellular Cloning

Unicellular organisms, such as bacteria and yeast, naturally produce clones of themselves when they replicate asexually by binary fission; this is known as **cellular cloning**. The nuclear DNA duplicates by the process of mitosis, which creates an exact replica of the genetic material. Cellular cloning is often used as a tool in molecular biology studies, when an asexually reproducing organism is “cloned” in order to increase a portion of DNA added to the cell.

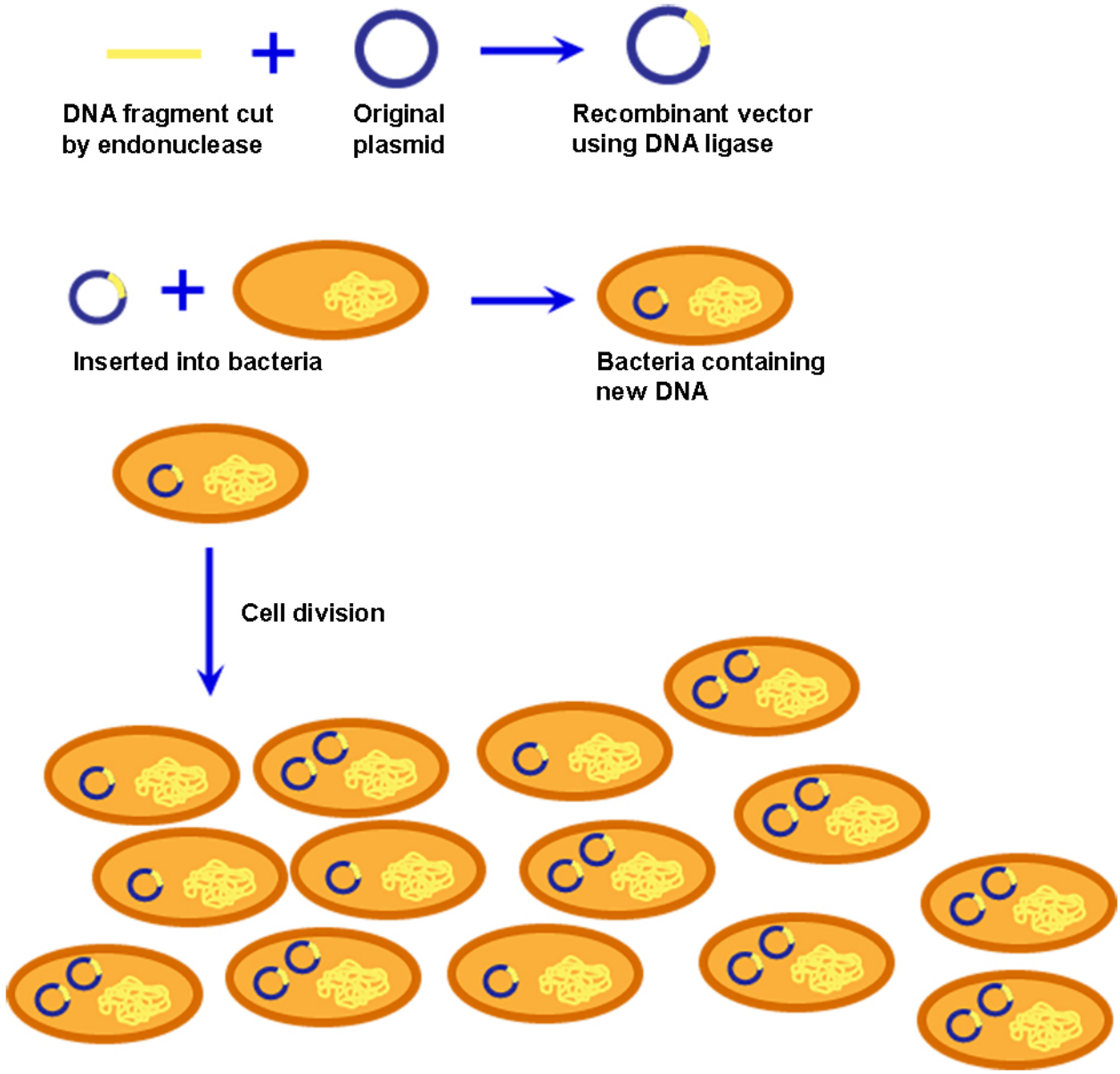

Figure 2. Diagram of the steps of cellular cloning

Reproductive Cloning

Reproductive cloning is a method used to make a clone or an identical copy of an entire multicellular organism. Most multicellular organisms undergo reproduction by sexual means, which involves genetic hybridization of two individuals (parents), making it impossible for generation of an identical copy or a clone of either parent. Recent

advances in biotechnology have made it possible to artificially induce asexual reproduction of mammals in the laboratory.

Parthenogenesis, or “virgin birth,” occurs when an embryo grows and develops without the fertilization of the egg occurring; this is a form of asexual reproduction. An example of parthenogenesis occurs in species in which the female lays an egg and if the egg is fertilized, it is a diploid egg and the individual develops into a female; if the egg is not fertilized, it remains a haploid egg and develops into a male. The unfertilized egg is called a parthenogenic, or virgin, egg. Some insects and reptiles lay parthenogenic eggs that can develop into adults.

Sexual reproduction requires two cells; when the haploid egg and sperm cells fuse, a diploid zygote results. The zygote nucleus contains the genetic information to produce a new individual. However, early embryonic development requires the cytoplasmic material contained in the egg cell. This idea forms the basis for reproductive cloning. Therefore, if the haploid nucleus of an egg cell is replaced with a diploid nucleus from the cell of any individual of the same species (called a donor), it will become a zygote that is genetically identical to the donor. Somatic cell nuclear transfer is the technique of transferring a diploid nucleus into an enucleated egg. It can be used for either therapeutic cloning or reproductive cloning.

The first cloned animal was Dolly, a sheep who was born in 1996. The success rate of reproductive cloning at the time was very low. Dolly lived for seven years and died of respiratory complications (Figure 3). There is speculation that because the cell DNA belongs to an older individual, the age of the DNA may affect the life expectancy of a cloned individual. Since Dolly, several animals such as horses, bulls, and goats have been successfully cloned, although these individuals often exhibit facial, limb, and cardiac abnormalities. There have been attempts at producing cloned human embryos as sources of embryonic stem cells, sometimes referred to as cloning for therapeutic purposes. Therapeutic cloning produces stem cells to attempt to remedy detrimental diseases or defects (unlike reproductive cloning, which aims to reproduce an organism). Still, therapeutic cloning efforts have met with resistance because of bioethical considerations.

Practice Question

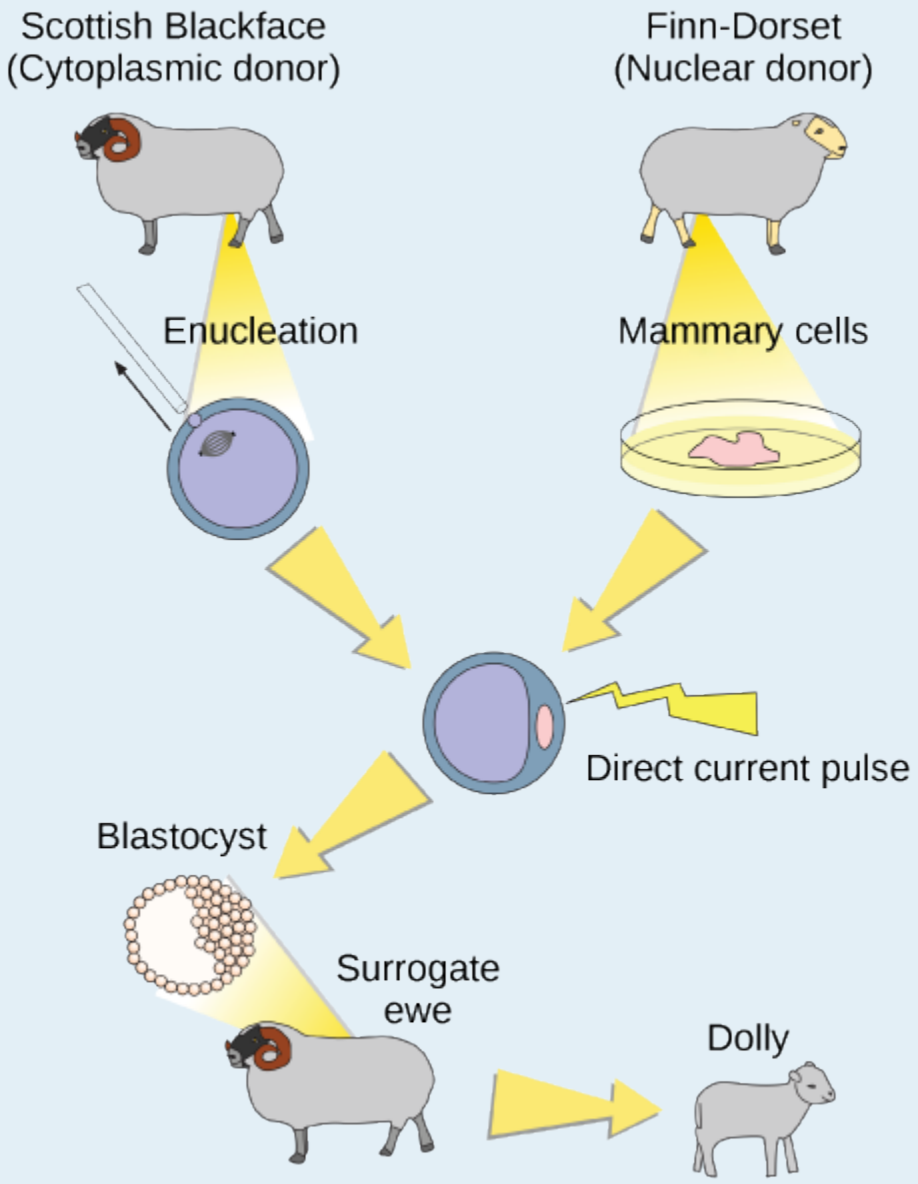

Figure 3. Dolly the sheep was the first mammal to be cloned. To create Dolly, the nucleus was removed from a donor egg cell. The nucleus from a second sheep was then introduced into the cell, which was allowed to divide to the blastocyst stage before being implanted in a surrogate mother. (credit: modification of work by "Squidonius"/Wikimedia Commons)

Do you think Dolly was a Finn-Dorset or a Scottish Blackface sheep?

Answer

Dolly was a Finn-Dorset sheep because even though the original cell came from a Scottish blackface sheep and the surrogate mother was a Scottish blackface, the DNA came from a Finn-Dorset.

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GENETIC ENGINEERING

Learning Outcomes

Understand the basics of genetic engineering

Genetic engineering is the alteration of an organism's genotype using recombinant DNA technology to modify an organism's DNA to achieve desirable traits. The addition of foreign DNA in the form of recombinant DNA vectors generated by molecular cloning is the most common method of genetic engineering. The organism that receives the recombinant DNA is called a **genetically modified organism (GMO)**. If the foreign DNA that is introduced comes from a different species, the host organism is called **transgenic**. Bacteria, plants, and animals have been genetically modified since the early 1970s for academic, medical, agricultural, and industrial purposes. In the US, GMOs such as Roundup-ready soybeans and borer-resistant corn are part of many common processed foods.

Gene Targeting

Although classical methods of studying the function of genes began with a given phenotype and determined the genetic basis of that phenotype, modern techniques allow researchers to start at the DNA sequence level and ask: "What does this gene or DNA element do?" This technique, called reverse genetics, has resulted in reversing the classic genetic methodology. This method would be similar to damaging a body part to determine its function. An insect that loses a wing cannot fly, which means that the function of the wing is flight. The classical genetic method would compare insects that cannot fly with insects that can fly, and observe that the non-flying insects have lost wings. Similarly, mutating or deleting genes provides researchers with clues about gene function. The methods used to disable gene function are collectively called gene targeting. **Gene targeting** is the use of recombinant DNA vectors to alter the expression of a particular gene, either by introducing mutations in a gene, or by eliminating the expression of a certain gene by deleting a part or all of the gene sequence from the genome of an organism.

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INTRODUCTION TO BIOTECHNOLOGY APPLICATIONS

What you'll learn to do: Identify societal uses of biotechnology

This video provides an introduction to some of the newest breakthroughs in biotechnology: biofuel, health, and agriculture.

Watch this video online: https://youtu.be/ebSWG1QmxnY

In this outcome we will discuss the ways biotechnology is used in our society.

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MEDICINAL BIOTECHNOLOGY

Learning Outcomes

List at least 3 medicinal uses of biotechnology advances

It is easy to see how biotechnology can be used for medicinal purposes. Knowledge of the genetic makeup of our species, the genetic basis of heritable diseases, and the invention of technology to manipulate and fix mutant genes provides methods to treat the disease.

Genetic Diagnosis and Gene Therapy

The process of testing for suspected genetic defects before administering treatment is called **genetic diagnosis** by **genetic testing**. Depending on the inheritance patterns of a disease-causing gene, family members are advised to undergo genetic testing. For example, women diagnosed with breast cancer are usually advised to have a biopsy so that the medical team can determine the genetic basis of cancer development. Treatment plans are based on the findings of genetic tests that determine the type of cancer. If the cancer is caused by inherited gene mutations, other female relatives are also advised to undergo genetic testing and periodic screening for breast cancer. Genetic testing is also offered for fetuses (or embryos with *in vitro* fertilization) to determine the presence or absence of disease-causing genes in families with specific debilitating diseases.

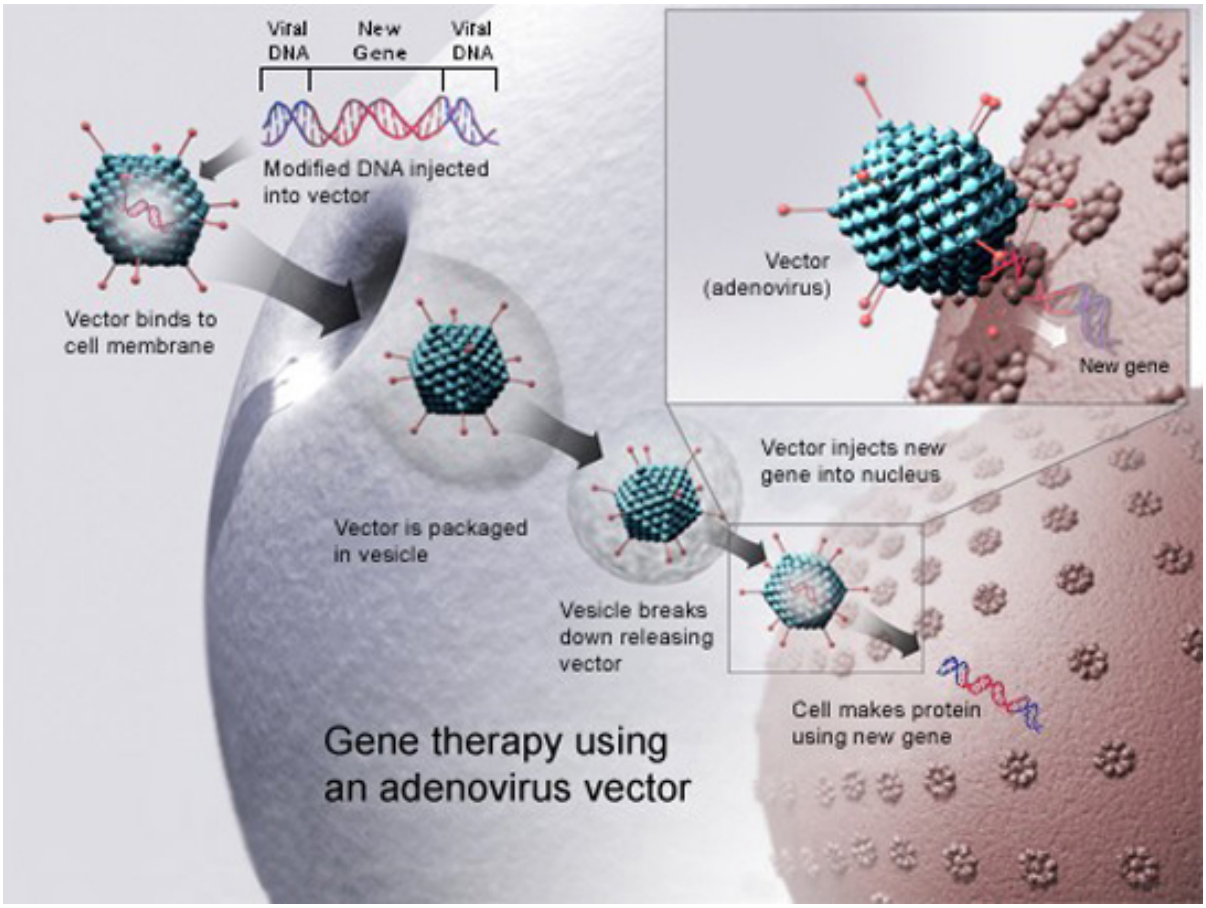

Figure 1. Gene therapy using an adenovirus vector can be used to cure certain genetic diseases in which a person has a defective gene. (credit: NIH)

Gene therapy is a genetic engineering technique used to cure disease. In its simplest form, it involves the introduction of a good gene at a random location in the genome to aid the cure of a disease that is caused by a mutated gene. The good gene is usually introduced into diseased cells as part of a vector transmitted by a virus that can infect the host cell and deliver the foreign DNA (Figure 1). More advanced forms of gene therapy try to correct the mutation at the original site in the genome, such as is the case with treatment of severe combined immunodeficiency (SCID).

Here's a website to help you learn more information on SCID and its gene therapy trials.

Production of Vaccines, Antibiotics, and Hormones

Traditional vaccination strategies use weakened or inactive forms of microorganisms to mount the initial immune response. Modern techniques use the genes of microorganisms cloned into vectors to mass produce the desired antigen. The antigen is then introduced into the body to stimulate the primary immune response and trigger immune memory. Genes cloned from the influenza virus have been used to combat the constantly changing strains of this virus.

Antibiotics are a biotechnological product. They are naturally produced by microorganisms, such as fungi, to attain an advantage over bacterial populations. Antibiotics are produced on a large scale by cultivating and manipulating fungal cells.

Recombinant DNA technology was used to produce large-scale quantities of human insulin in *E. coli* as early as 1978. Previously, it was only possible to treat diabetes with pig insulin, which caused allergic reactions in humans because of differences in the gene product. In addition, human growth hormone (HGH) is used to treat growth

disorders in children. The HGH gene was cloned from a cDNA library and inserted into *E. coli* cells by cloning it into a bacterial vector.

In Summary: Medicinal Biotechnology

Transgenic organisms possess DNA from a different species, usually generated by molecular cloning techniques. Vaccines, antibiotics, and hormones are examples of products obtained by recombinant DNA technology. Transgenic plants are usually created to improve characteristics of crop plants.

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AGRICULTURAL BIOTECHNOLOGY

Learning Outcomes

List at least 3 agricultural uses of biotechnology advances

Biotechnology in agriculture can enhance resistance to disease, pest, and environmental stress, and improve both crop yield and quality.

Transgenic Animals

Although several recombinant proteins used in medicine are successfully produced in bacteria, some proteins require a eukaryotic animal host for proper processing. For this reason, the desired genes are cloned and expressed in animals, such as sheep, goats, chickens, and mice. Animals that have been modified to express recombinant DNA are called transgenic animals. Several human proteins are expressed in the milk of transgenic sheep and goats, and some are expressed in the eggs of chickens. Mice have been used extensively for expressing and studying the effects of recombinant genes and mutations.

Transgenic Plants

Manipulating the DNA of plants (i.e., creating GMOs) has helped to create desirable traits, such as disease resistance, herbicide and pesticide resistance, better nutritional value, and better shelf-life (Figure 1). Plants are the most important source of food for the human population. Farmers developed ways to select for plant varieties with desirable traits long before modern-day biotechnology practices were established.

Figure 1. Corn, a major agricultural crop used to create products for a variety of industries, is often modified through plant biotechnology. (credit: Keith Weller, USDA)

Plants that have received recombinant DNA from other species are called transgenic plants. Because they are not natural, transgenic plants and other GMOs are closely monitored by government agencies to ensure that they are fit for human consumption and do not endanger other plant and animal life. Because foreign genes can spread to other species in the environment, extensive testing is required to ensure ecological stability. Staples like corn, potatoes, and tomatoes were the first crop plants to be genetically engineered.

Transformation of Plants Using *Agrobacterium tumefaciens*

Gene transfer occurs naturally between species in microbial populations. Many viruses that cause human diseases, such as cancer, act by incorporating their DNA into the human genome. In plants, tumors caused by the bacterium *Agrobacterium tumefaciens* occur by transfer of DNA from the bacterium to the plant. Although the tumors do not kill the plants, they make the plants stunted and more susceptible to harsh environmental conditions. Many plants, such as walnuts, grapes, nut trees, and beets, are affected by *A. tumefaciens*. The artificial introduction of DNA into plant cells is more challenging than in animal cells because of the thick plant cell wall.

Researchers used the natural transfer of DNA from *Agrobacterium* to a plant host to introduce DNA fragments of their choice into plant hosts. In nature, the disease-causing *A. tumefaciens* have a set of plasmids, called the Ti plasmids (tumor-inducing plasmids), that contain genes for the production of tumors in plants. DNA from the Ti plasmid integrates into the infected plant cell's genome. Researchers manipulate the Ti plasmids to remove the tumor-causing genes and insert the desired DNA fragment for transfer into the plant genome. The Ti plasmids carry antibiotic resistance genes to aid selection and can be propagated in *E. coli* cells as well.

Flavr Savr Tomato

The first GM crop to be introduced into the market was the Flavr Savr Tomato produced in 1994. Antisense RNA technology was used to slow down the process of softening and rotting caused by fungal infections, which led to increased shelf life of the GM tomatoes. Additional genetic modification improved the flavor of this tomato. The Flavr Savr tomato did not successfully stay in the market because of problems maintaining and shipping the crop. However, since that time numerous crop plants have been developed and approved for sale and consumption. Corn, soybeans, and cotton in particular have been widely adopted by U.S. farmers.

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INTRODUCTION TO RISKS AND BENEFITS OF GENOMIC SCIENCE

What you'll learn to do: Discuss the risks and benefits involved in applications of genetic and genomic science

Scientific developments can be used in a lot of different ways that impact our lives. Genetics and genomics both play roles in health and disease.

Genetics helps individuals and families learn about how conditions such as sickle cell anemia and cystic fibrosis are inherited in families, what screening and testing options are available, and, for some genetic conditions, what treatments are available.

Genomics is helping researchers discover why some people get sick from certain infections, environmental factors, and behaviors, while others do not. For example, there are some people who exercise their whole lives, eat a healthy diet, have regular medical checkups, and die of a heart attack at age 40. There are also people who smoke, never exercise, eat unhealthy foods and live to be 100. Genomics may hold the key to understanding these differences.

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GENETIC INFORMATION USED FOR IDENTIFICATION

Learning Outcomes

Outline how genetic information is used in personal identification

DNA as a Forensic Tool

Information and clues obtained from DNA samples found at crime scenes have been used as evidence in court cases, and genetic markers have been used in forensic analysis. Genomic analysis has also become useful in this field. In 2001, the first use of genomics in forensics was published. It was a collaborative attempt between academic research institutions and the FBI to solve the mysterious cases of anthrax communicated via the US Postal Service. Using microbial genomics, researchers determined that a specific strain of anthrax was used in all the mailings.

Mitochondrial Genomics

Mitochondria are intracellular organelles that contain their own DNA. Mitochondrial DNA mutates at a rapid rate and is often used to study evolutionary relationships. Another feature that makes studying the mitochondrial genome interesting is that the mitochondrial DNA in most multicellular organisms is passed on only from the mother during the process of fertilization. For this reason, mitochondrial genomics is often used to trace genealogy.

DNA Fingerprinting

DNA fingerprinting (also called DNA profiling, DNA testing, or DNA typing) is a forensic technique used to identify individuals by characteristics of their DNA. A DNA profile is a small set of DNA variations that is very likely to be different in all unrelated individuals, thereby being as unique to individuals as are fingerprints (hence the name for the technique).

Although 99.9% of human DNA sequences are the same in every person, enough of the DNA is different that it is possible to distinguish one individual from another, unless they are monozygotic ("identical") twins. DNA fingerprinting uses repetitive sequences that are highly variable, called variable number tandem repeats (VNTRs). Modern law enforcement in particular uses short tandem repeats (STRs). STR loci are very similar between closely related individuals, but are so variable that unrelated individuals are extremely unlikely to have the same STRs. The combination of STRs used by law enforcement enable identification though because even closely related individuals will not share all the same STR loci.

The modern process of DNA fingerprinting was developed in 1984 by Sir Alec Jeffreys, while he was working in the Department of Genetics at the University of Leicester. DNA fingerprinting can be used to identify a person or to place a person at a crime scene and to help clarify paternity. DNA fingerprinting has also been widely used in the study of animal and floral populations and has revolutionized the fields of zoology, botany, and agriculture.

Video Review

Watch this video on the process of DNA fingerprinting and DNA profiling

Watch this video online: https://youtu.be/DbR9xMXuK7c

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MEDICAL USES OF GENETIC INFORMATION

Learning Outcomes

Discuss medical uses of genetic information and the potential benefits and risks of this

Personalized Medicine

Watch this video and consider whether you would be interested in knowing details about your own personal disease risk or susceptibility.

Watch this video online: https://youtu.be/OON70d8gTnc

Predicting Disease Risk at the Individual Level

Predicting the risk of disease involves screening currently healthy individuals by genome analysis at the individual level. Intervention with lifestyle changes and drugs can be recommended before disease onset. However, this approach is most applicable when the problem resides within a single gene defect. Such defects only account for approximately 5 percent of diseases in developed countries. Most of the common diseases, such as heart disease, are multi-factored or polygenic, which is a phenotypic characteristic that involves two or more genes, and also involve environmental factors such as diet. In April 2010, scientists at Stanford University published the genome analysis of a healthy individual (Stephen Quake, a scientist at Stanford University, who had his genome sequenced); the analysis predicted his propensity to acquire various diseases. A risk assessment was performed to analyze Quake's percentage of risk for 55 different medical conditions. A rare genetic mutation was found, which showed him to be at risk for sudden heart attack. He was also predicted to have a 23 percent risk of developing prostate cancer and a 1.4 percent risk of developing Alzheimer's. The scientists used databases and several publications to analyze the genomic data. Even though genomic sequencing is becoming more affordable and analytical tools are becoming more reliable, ethical issues surrounding genomic analysis at a population level remain to be addressed.

Debate remains over what to do with individual level data as well, such as the data from the genomic analysis of Quake's DNA. As a result of the study it was recommended that Quake start a regiment of preventative statins; the long-term effects of this study or treatment remain unknown at this stage.

For example, in 2011, the United States Preventative Services Task Force recommended against using the PSA test to screen healthy men for prostate cancer. Their recommendation is based on evidence that screening does not reduce the risk of death from prostate cancer. Prostate cancer often develops very slowly and does not cause problems, while the cancer treatment can have severe side effects. The *PCA3* (Figure 1) test is considered to be more accurate, but screening may still result in men who would not have been harmed by the cancer itself suffering side effects from treatment.

What do you think? Should all healthy men be screened for prostate cancer using the *PCA3* or PSA test? Should people in general be screened to find out if they have a genetic risk for cancer or other diseases?

There are no right or wrong answers to these questions. While it is true that prostate cancer treatment itself can be harmful, many men would rather be aware that they have cancer so they can monitor the disease and begin treatment if it progresses. And while genetic screening may be useful, it is expensive and may cause needless worry. People with certain risk factors may never develop the disease, and preventative treatments may do more harm than good.

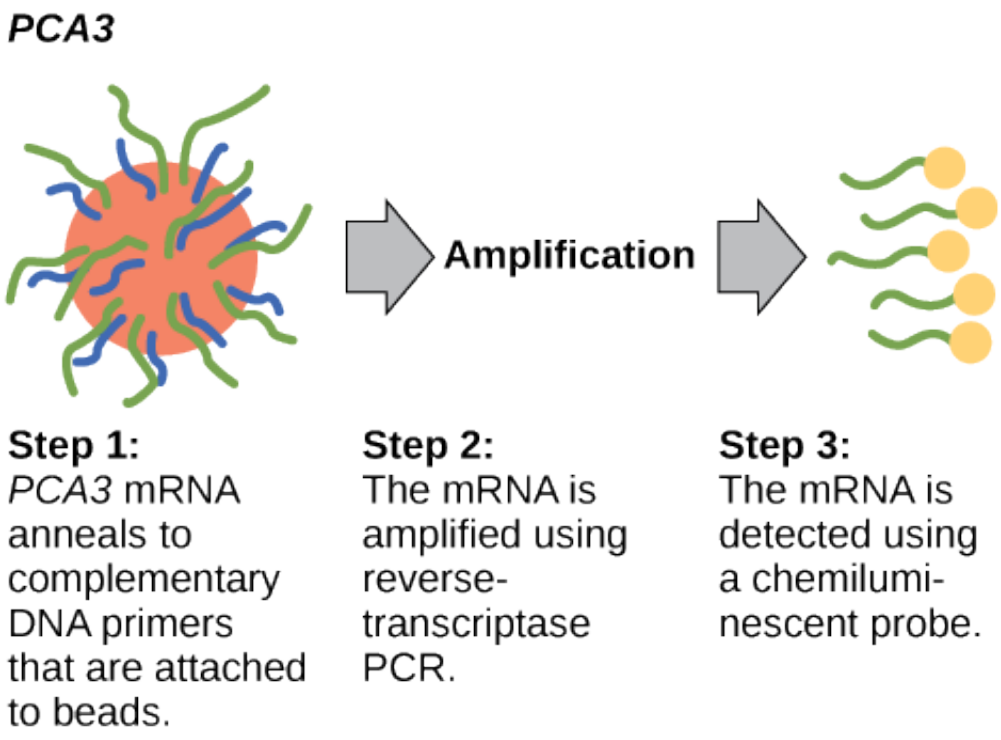

Figure 1. PCA3 is a gene that is expressed in prostate epithelial cells and overexpressed in cancerous cells. A high concentration of PCA3 in urine is indicative of prostate cancer. The PCA3 test is considered to be a better indicator of cancer than the more well know PSA test, which measures the level of PSA (prostate-specific antigen) in the blood.

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GENOMICS IN AGRICULTURE

Learning Outcomes

Outline the potential benefits and risks associated with agricultural uses of biotechnology

Genomics can reduce the trials and failures involved in scientific research to a certain extent, which could improve the quality and quantity of crop yields in agriculture. Linking traits to genes or gene signatures helps to improve crop breeding to generate hybrids with the most desirable qualities. Scientists use genomic data to identify desirable traits, and then transfer those traits to a different organism. Scientists are discovering how genomics can improve the quality and quantity of agricultural production. For example, scientists could use desirable traits to create a useful product or enhance an existing product, such as making a drought-sensitive crop more tolerant of the dry season.

GMO Controversies: Science versus Public Fear

Watch Borut Bohanec, the Chair of the Department of Agronomy at the University of Ljubljana (in Slovenia), as he discusses the fears and potential benefits surrounding GMOs.

Watch this video online: https://youtu.be/mz4_TwdaYeI

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- GMO controversies - science vs. public fear: Borut Bohanec at TEDxLjubljana. **Authored by**: TEDx Talks. **Located at**: https://youtu.be/mz4_TwdaYeI. **License**: *All Rights Reserved*. **License Terms**: Standard YouTube License

PUTTING IT TOGETHER: MODERN BIOLOGY

Let's return to our discussion of the unknown solider we discussed at the beginning of the module. First Lieutenant Michael Joseph Blassie, 24, was shot down over South Vietnam in 1972 and presumed dead.

In the early 1990s, Blassie's family received word that his remains might be buried in the Tomb of the Unknown. They petitioned the Department of Defense to open the site and conduct DNA testing, a technology that had been unavailable when the remains were first brought to and buried in the Tomb of the Unknown. In 1998, the Tomb of the Unknown was opened and the remains of the Vietnam Unknown—previously identified as X-26—were removed.

Forensic anthropologists took the aged and damaged samples of bone for mitochondrial DNA (mtDNA) testing. Because mtDNA is passed along the maternal line, scientists compared the Unknown Soldier's DNA against two samples submitted by First Lieutenant Blassie's mother and sister and found a match.

On July 11, 1998, 1st Lt. Michael Blassie was buried with full military honors in Jefferson National Cemetery, Missouri, near his hometown, in the same cemetery as his father.

Figure 1. A photo of Michael Blassie in his Air Force Academy Cadet Uniform.

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