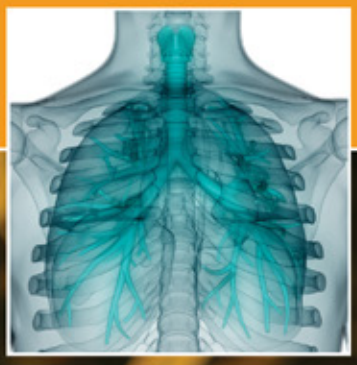
THIRD EDITION

ANATOMY AND PHYSIOLOGY

FOR HEALTH PROFESSIONALS





JAHANGIR MOINI

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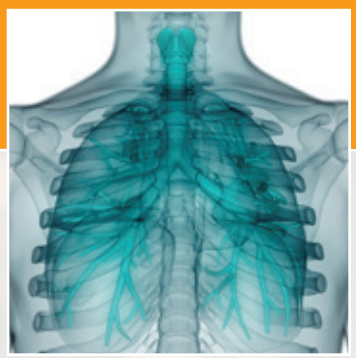

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Dedication

This book is dedicated to my wonderful and amazing wife, Hengameh, and two beautiful daughters, Mahkameh and Morvarid.

It is also dedicated to my granddaughters, Laila Jade and Annabelle Jasmine Mabry.

Brief Contents

Contents

Preface

In 35 years of teaching anatomy and physiology, I have utilized numerous books related to the subject. Some were very high level while others were very low level, and I could not find a "middle ground" book that really taught the subject to allied health students; surprising, given that this is a time when the field is growing exponentially. Therefore, I undertook the writing of this book for all allied health professionals. Anatomy and physiology are two of the major core subjects for almost all allied health professionals—they must understand the structures and normal functions of the body in the simplest possible terms. This book strives to make that possible.

Organization of This Text

This text is based on levels of organization within the body and becomes more multifaceted as the student incorporates the understanding of basic, then intermediate, and finally more complex subjects.

In total, the text consists of six units:

- *Unit I, Levels of Organization*: This unit begins by providing a general introduction to human anatomy and physiology along with the organization levels through which the body is understood. It then delves into the atomic, molecular, and chemical interactions on which life is based before moving on to discussions of the cells and tissues that comprise the body.
- *Unit II, Support and Movement:* This unit focuses on the body systems that support the body and allow for a range of motion. It first considers the integumentary system, composed of the skin and its accessory structures; these are the body's first line of defense against the environment. The text then approaches the bones and joints that comprise the skeletal system before discussing the muscular system.
- *Unit III, Control and Coordination*: This unit tackles the critical components of the body that control all body functions. The text considers the all-important nervous system across four chapters on neural tissue, the central nervous system, the peripheral nervous system, and the senses. The unit then ends with a chapter on the endocrine system, which works along with the nervous system to regulate the functions of the human body to maintain homeostasis.
- *Unit IV, Transport*: This unit focuses on the cardiovascular and lymphatic systems, which keep the body running. The first three chapters discuss the major components of the cardiovascular system: the heart, blood, and blood vessels. The last chapter in this unit focuses on the lymphatic system. Like the cardiovascular system, it transports fluids through a network of vessels; without the lymphatic system, fluid would accumulate in tissue spaces.
- *Unit V, Environmental Exchange*: This unit considers the systems and processes that balance what the body intakes with what it expels. The unit first examines the respiratory system, which intakes oxygen and removes carbon dioxide from the body, before shifting focus to the urinary system, which eliminates wastes and maintains homeostatic regulation of the volume and solute concentration of blood plasma. The text then surveys fluid, electrolyte, and acid-base balance before moving on to the digestive system, which in simplest terms supplies nutrients for body cells.
- *Unit VI, Continuity of Life*: In this unit, the focus shifts to the male and female reproductive systems, which, while not essential to the survival of an individual, are needed to ensure the continued existence of the human species. The final chapter, then, discusses pregnancy before delving into a brief discussion of genetics.

In addition to the recurring features that guide the student through each chapter (of which an overview is given in the "How to Use This Book" section), the body systems chapters contained in this text address the effects of aging on each specific system, information

that is especially critical at a time when the number of older adults is on the rise.

New to This Edition

This *Third Edition* has been updated to take into account both advancements in medical knowledge in the last several years as well as feedback from valued users of the *First* and *Second Editions*.

For the *Third Edition*, the text was expanded from 24 to 27 chapters. Changes include:

- **NEW! Case Studies** are included for instructors and students as part of the Teaching and Learning Package as interactive or printable PDFs to help students apply concepts to real-world scenarios.
- **NEW!** *Chapter 7, Muscle Tissue*: This chapter focuses on the more basic concepts of the various types of muscle tissues, including structures and functions.
- **REVISED!** *Chapter 8, Muscular System*: This chapter focuses on the actual muscles of the body and their functions. Lever systems is a topic that has been added, and this chapter includes discussion of skeletal, smooth, and cardiac muscles.
- **NEW and REVISED!** *Chapter 14, Autonomic Nervous System*: This chapter has been separated from the second edition's "Peripheral Nervous System" chapter, and both these new chapters have been greatly expanded.
- **NEW!** *Chapter 27, Heredity*: This is a new chapter that explains how our genes and chromosomes influence every facet of life. It focuses on various types of inheritance and genetic screening.
- **NEW!** There are four new Appendices in this edition. They include:
  - Common Abbreviations
  - Medical Terminology
  - The Metric System
  - Reference Values for Blood and Urine

New tables and figures have been added as appropriate.

The Teaching and Learning Package

The Teaching and Learning packages for instructors and students encourage going beyond the content of the text as well as driving home key concepts within the text.

Instructor Teaching Package

Qualified instructors can receive a full suite of extensive Instructor Resources, including:

- *Slides in PowerPoint format*: Featuring more than 2,000 slides. At the bottom of each slide, **Lecture Notes** have been added to assist instructors in better explanations of the concepts summarized on the slides as well as to provide students in the form of study notes.
- *Test Bank*: Containing more than 1,000 questions
- *Instructor's Manual*: Including teaching strategies, lecture outlines, discussion topics, and answers to end of chapter questions as well as answers to Case Studies
- *Image Bank*: Supplying key figures from the text
- *Sample Syllabus*: Showing how a course can be structured around this text
- *Transition Guide*: Providing guidance in switching from the previous edition

Student Learning Package

The Student Learning Package includes:

- *Interactive eBook:* Featuring more than 20 **animations** to help students conceptualize key points of interest
- *Lab Exercises*: Interactive or printable PDFs, which students can use to visually apply the concepts learned
- *Practice Activities*: To test student understanding for each chapter
- *Case Studies*: Available as interactive or printable PDFs to help students apply concepts to real-world scenarios
- *Flash Cards*: For self-study

About the Author

Dr. Jahangir Moini is currently a professor at Eastern Florida State College, where he teaches anatomy and physiology as well as other science courses. He was previously assistant professor at Tehran University School of Medicine for nine years, where he taught medical and allied health students. Dr. Moini is a former professor and director (for 15 years) of allied health programs at Everest University. In total, he served at Everest University for 24 years.

As a physician and instructor for the past 45 years, he advocates that all health professionals must understand the structures and functions of the human body. Other sciences such as pathology, pharmacology, and chemistry are correlated with the knowledge of anatomy and physiology.

Dr. Moini is actively involved in teaching and helping students prepare for service in various health professions. He has been an internationally published author of 25 allied health books since 1999.

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I would like to acknowledge the following individuals for their time and efforts in aiding with this book:

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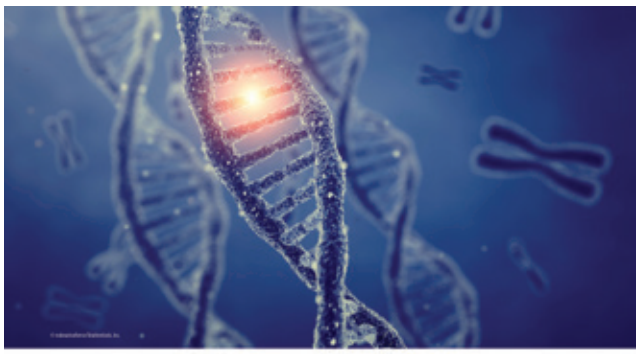
A Visual Walkthrough

Anatomy and Physiology for Health Professionals, Third Edition incorporates a number of engaging pedagogical features to aid in the students' understanding and retention of the material. A colorful and engaging layout enables easy reading and supports the retention of important concepts. Hundreds of full-color photographs and medically accurate illustrations provide valuable insight into human anatomy and physiology.

Objectives and Outline

Each chapter begins with a framework for learning the most important topics by presenting *Objectives* that list the chapter's desired outcomes and an *Outline* indicating the material to be discussed.



CHAPTER 27

Heredity

OUTLINE

OVERVIEW
GENES
- Alleles
- Genotype and Phenotype

GENETIC VARIATION
- Chromosome Segregation and Independent Assortment
- Crossover of Homologues and Gene Recombination
- Random Fertilization

PATTERNS OF INHERITANCE
- Dominant-Recessive Inheritance
- Polygene Inheritance

ENVIRONMENTAL FACTORS
- Factors that Affect Expression of Single Genes
- Regulation of Gene Expression
- Mitochondrial Inheritance

GENDER-RELATED FACTORS
- Sex Determination
- Chromosome Disorders

GENETIC SCREENING
- Carrier Recognition
- Fetal Testing
- Human Gene Therapy

SUMMARY
KEY TERMS
LEARNING GOALS
CRITICAL THINKING QUESTIONS
REVIEW QUESTIONS
ESSAY QUESTIONS

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Genes 715

OBJECTIVES

After studying this chapter, the reader should be able to:

1. Identify what was achieved by the Human Genome Project.
2. Define the terms *genes, chromosomes, autosomes, alleles,* and *homozygous.*
3. Distinguish between dominant and recessive alleles.
4. List the three events that occur as part of genetic variation.
5. Explain the concept of multiple allele inheritance using the ABO blood group.
6. Identify factors that may influence or override gene expression.
7. Contrast Prader-Willi syndrome and Angelman's syndrome.
8. Explain why mitochondrial (extranuclear) genes are usually transmitted from the mother to her offspring.
9. Identify the newer procedures that will likely replace amniocentesis and chorionic villus sampling.
10. Give examples of how gene therapy may be used.

Overview

The science of *genetics* is the study of *heredity.* In the mid-1800s, Gregor Mendel was the first scientist to propose basic principles of heredity. Today's *geneticists* are able to engineer and manipulate human genes in order to examine the ways in which they are expressed. The *Human Genome Project* was undertaken to determine the actual human DNA sequence. As the field of genetic screening develops, many future medical outcomes as yet not even imagined will be possible. Already, the use of the human genome is allowing for drugs to be developed that are more effective in treating diseases and even curing them.

Genes

Children inherit traits from their parents and relatives as determined by DNA. Individual segments of DNA are known as *genes.* When a gene's DNA sequence changes or *mutates,* an illness or other condition may result. *Spontaneous mutations* occur because of random DNA replication errors. The field of **genetics** investigates how genes result in certain characteristics affecting health or contributing to natural variation. It also focuses on how genes are passed from generation to generation. Today, fetal **chromosome** checks provide clues to a child's future health.

All human cells, except **gametes,** have a *diploid* number of chromosomes (46). This number consists of 23 *pairs* of **homologous chromosomes.** One pair is from the father (via the sperm) and the other pair is from the mother (via the egg). Homologous chromosomes appear similar, carrying genes for the same traits. However, they do not always bring about the same expressions of the traits.

Autosomes are pairs 1 through 22 and do not carry genes that determine sex, but function to guide the expression of most other traits. Only pair 23, the X and Y chromosomes, is **sex chromosomes.** Females have two X chromosomes and males have one X and one Y chromosome. A sex chromosome pair may be very different in size. **Karyotypes** are charts that display the 23 chromosome pairs of human somatic cells (FIGURE 27-1). The diploid **genome** is actually an individual's genetic (DNA) makeup. It represents the two sets of genetic instructions, from the egg and from the sperm.

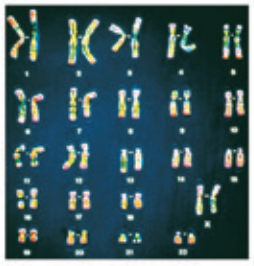

FIGURE 27-1 The 23 pairs of the human karyotype.

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Alleles

Somatic cells have two copies of each autosome, and therefore two copies of each gene. Gene copies can be identical or slightly different in a DNA sequence. Such varying forms of a gene are called **alleles**. A gene's position on a chromosome is called a **locus**. An allele may code for identical or different forms of a given trait. An individual with two identical alleles of a gene is **homozygous** for that gene. A person with two different alleles is **heterozygous** for that gene. A heterozygote is also called a *carrier*. A person with only one copy (allele) of a gene is referred to as **hemizygous**.

Patterns of inheritance through families are known as *modes of inheritance*. A **dominant** allele masks expression of a **recessive** allele. Genes cannot be expressed unless they are present in a homozygous condition. Capital letters are used to designate dominant alleles. The extent to which a certain allele is expressed when it is present is called its *expressivity*. The following generalizations describe modes of inheritance:

- *Autosomal conditions that affect both sexes:* X-linked characteristics affect males much more often than females. Y-linked characteristics are only passed from father to son.
- *Autosomal recessive conditions from two healthy carrier parents:* Recessive conditions can "skip" generations.
- *Dominant condition with at least one affected parent:* Generations are not skipped in these cases.

Genotype and Phenotype

The alleles situated at one or more sites on homologous chromosomes make up an individual's **genotype** (actual genetic makeup). A pair of alleles is usually designated by letters or symbols, such as "AA" when the alleles are identical and "Aa" when they are different. *Penetrance* is the regularity with which an allele is expressed in an individual who carries it. If the allele always produces its effect on the phenotype, it is called *fully penetrant*. The condition called *achondroplasia* is caused by a fully penetrant allele. When the allele is present, achondroplasia results.

A **phenotype** consists of the complete observable characteristics of an organism or group. It includes anatomic, physiologic, biochemical, and behavior traits that are determined by the interaction of genetic makeup and environmental factors. A certain inherited trait may be anticipated to occur in the offspring of two individuals when their genes and chromosomes are considered.

TEST YOUR UNDERSTANDING

1. Explain the modes of inheritance.
2. How do the sex chromosomes differ between males and females?
3. Explain dominant and recessive alleles.
4. How does X-linked recessive inheritance affect males and females?
5. What are the differences between a genotype and a phenotype?

FOCUS ON PATHOLOGY

The most common conditions related to chromosomal abnormalities include Down syndrome, Klinefelter syndrome, and Turner syndrome. **Down syndrome**, also called trisomy 21, affects approximately one of every 700 infants in the United States. They usually are short in stature with rounded faces, a protruding tongue, slanted eyes, and mental retardation. Life span may be shorter than normal, and many Down syndrome babies die in infancy from heart abnormalities or respiratory infections. Incidence increases dramatically with a maternal age of over 35 years.

Klinefelter syndrome is a condition of gonadal defects that appear in males after puberty. It is caused by an extra X chromosome in one or more cell lines. Its characteristics include **hypogonadism**, long legs, mild **gynecomastia**, infertility, poor social adaptation, subnormal intelligence, chronic pulmonary disease, and varicose veins. Severity of its abnormalities is higher with a greater number of X chromosomes. The most common abnormality is a 47 XXY karyotype. Men with the XXXXY karyotype have significant congenital malformations and mental retardation.

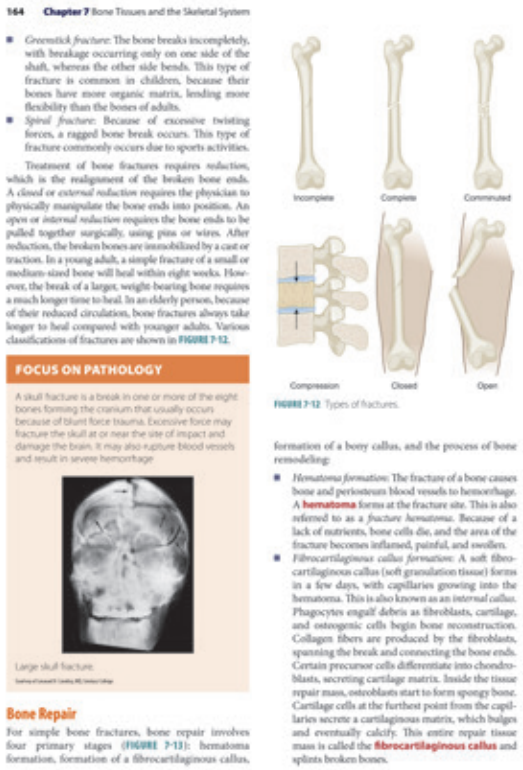
Turner syndrome is a chromosomal anomaly seen in about one in 3,000 live female births. It is characterized by the absence of one X chromosome. It results in congenital ovarian failure, genital hypoplasia, dwarfism, and underdevelopment of the breasts, uterus, and vagina. The karyotype for Turner syndrome is 45 XO.

Genetic Variation

Genetic variation results from the crossing over and exchange of chromosomal parts, and occurs during meiosis II. Every individual has a unique genotype and phenotype. This is based on three events: chromosome segregation and independent assortment, cross of homologues and gene recombination, and random fertilization.

Test Your Understanding

Each chapter contains *Test Your Understanding* boxes scattered throughout, which present open-ended questions that reinforce key content covered in the preceding sections.



164 Chapter 7 Bone Tissues and the Skeletal System

- *Greenstick fracture:* The bone breaks incompletely, with breakage occurring only on one side of the shaft, whereas the other side bends. This type of fracture is common in children, because their bones have more organic matrix, lending more flexibility than the bones of adults.
- *Spiral fracture:* Because of excessive twisting forces, a ragged bone break occurs. This type of fracture commonly occurs due to sports activities.

Treatment of bone fractures requires *reduction*, which is the realignment of the broken bone ends. A *closed* or *external reduction* requires the physician to physically manipulate the bone ends into position. An *open* or *internal reduction* requires the bone ends to be pulled together surgically, using pins or wires. After reduction, the broken bones are immobilized by a cast or traction. In a young adult, a simple fracture of a small or medium-sized bone will heal within eight weeks. However, the break of a larger, weight-bearing bone requires a much longer time to heal. In an elderly person, because of their reduced circulation, bone fractures always take longer to heal compared with younger adults. Various classifications of fractures are shown in FIGURE 7-12.

FOCUS ON PATHOLOGY

A skull fracture is a break in one or more of the eight bones forming the cranium that usually occurs because of blunt force trauma. Excessive force may fracture the skull at or near the site of impact and damage the brain. It may also rupture blood vessels and result in severe hemorrhage.

Large skull fracture.

Bone Repair

For simple bone fractures, bone repair involves four primary stages (FIGURE 7-13): hematoma formation, formation of a fibrocartilaginous callus, formation of a bony callus, and the process of bone remodeling:

- *Hematoma formation:* The fracture of a bone causes bone and periosteum blood vessels to hemorrhage. A **hematoma** forms at the fracture site. This is also referred to as a *fracture hematoma*. Because of a lack of nutrients, bone cells die, and the area of the fracture becomes inflamed, painful, and swollen.
- *Fibrocartilaginous callus formation:* A soft fibrocartilaginous callus (soft granulation tissue) forms in a few days, with capillaries growing into the hematoma. This is also known as an *internal callus*. Phagocytes engulf debris as fibroblasts, cartilage, and osteogenic cells begin bone reconstruction. Collagen fibers are produced by the fibroblasts, spanning the break and connecting the bone ends. Certain precursor cells differentiate into chondroblasts, secreting cartilage matrix. Inside the tissue repair mass, osteoblasts start to form spongy bone. Cartilage cells at the furthest point from the capillaries secrete a cartilaginous matrix, which bulges and eventually calcify. This entire repair tissue mass is called the **fibrocartilaginous callus** and splints broken bones.

Incomplete Complete Comminuted

Compression Closed Open

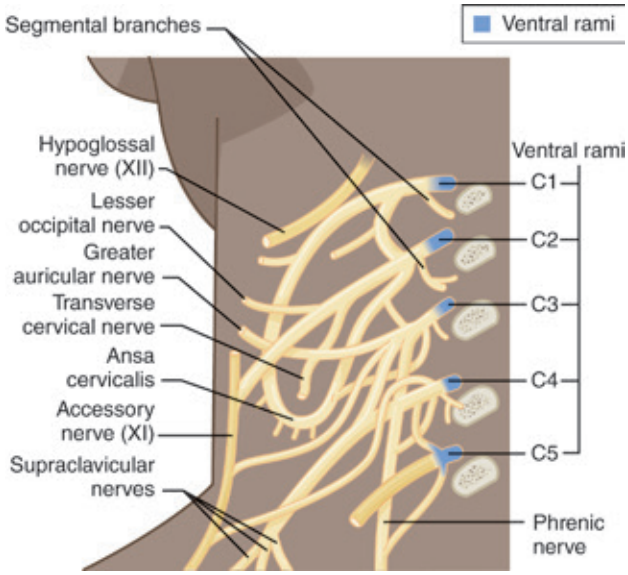
FIGURE 7-12 Types of fractures.

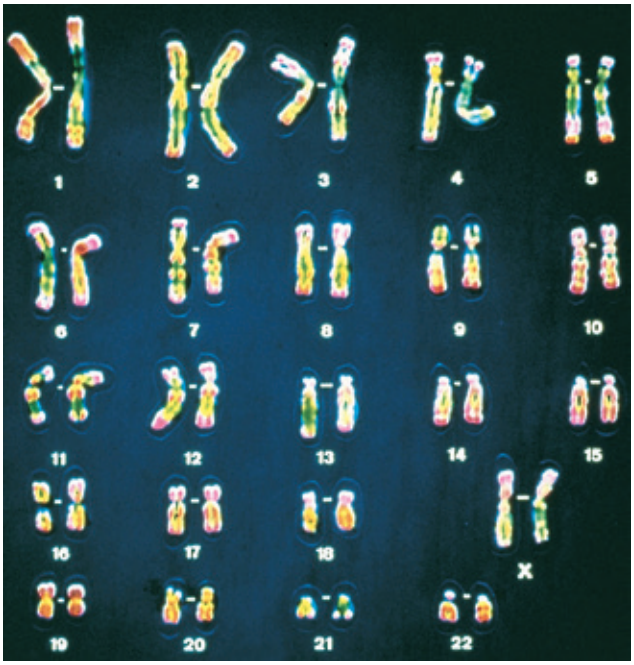
Focus on Pathology

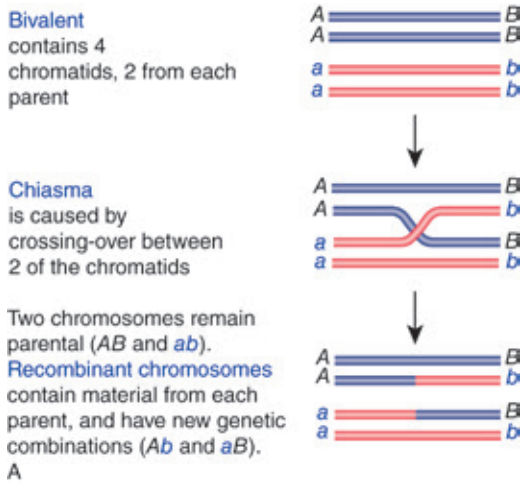
Focus on Pathology boxes connect the book's coverage of anatomy and physiology to important topics in pathology or the study of disease.

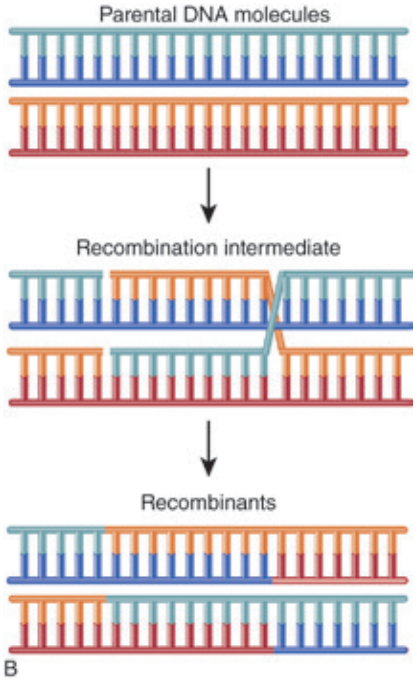
Extensive Art Program

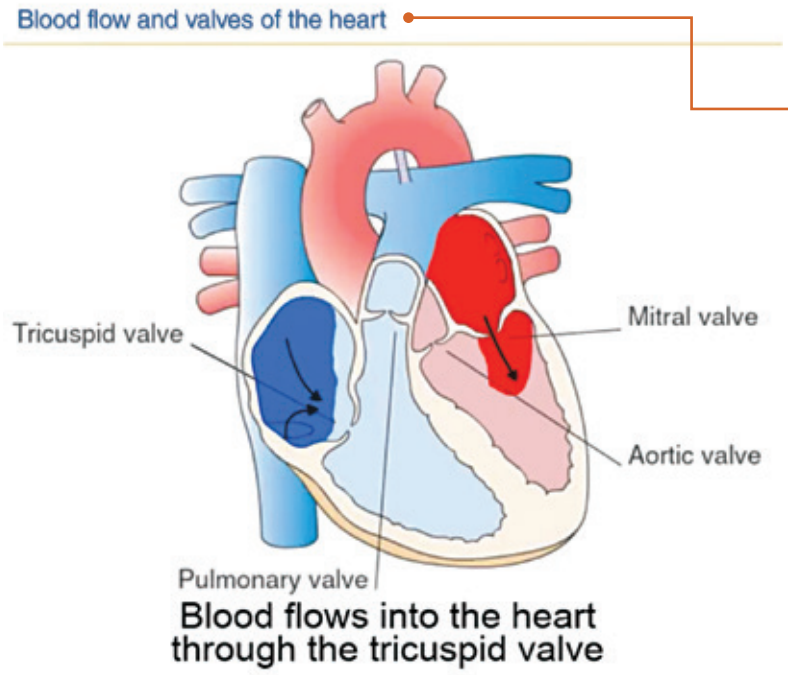
The extensive art program has been revised to include a number of new photos and illustrations to provide additional visual support for student comprehension.











Animations

Animations come with new, unused purchases of this book. Animations add visual clarity to key concepts and competencies.

severe fetal disorder is higher than the probability of any harm caused by the procedures.

Both amniocentesis and CVS will become obsolete because of advances in the sequencing of DNA. Today, maternal blood samples can be obtained easily, to examine free-floating maternal DNA as well as the DNA of the fetus that is released from the placenta. Maternal blood samples are tested for Down syndrome and other fetal chromosomal abnormalities. Similar blood testing can give information about the fetal genome, identifying mutations that may make it more likelihood for certain diseases to develop.

Human Gene Therapy

Human gene therapy is helping to treat or cure various disorders, especially those caused by a single gene or protein that is defective. It is possible to insert a virus that contains a functional gene into defective cells. Another method is to inject DNA that has been "corrected" into a patient's cells. These therapies have shown varying results in treating muscular dystrophy and cystic fibrosis. Genetic engineering is still very expensive, however, and there are opposing opinions about its ethics, infringement on religious beliefs, and how it can or should be used.

TEST YOUR UNDERSTANDING

1. What common disorders are routinely checked for by using genetic screening?
2. Describe a "pedigree."
3. Contrast how amniocentesis and CVS are performed.

Summary

The field of genetics studies how children inherit traits from their parents and relatives as determined by DNA. While a karyotype is a representation of a person's chromosomes, his or her complete genetic complement is the genome. The combination of varying forms of genes, called alleles, makes up a person's genotype. When an allele pair is identical, the individual is homozygous for that trait. When alleles are different, the individual is heterozygous. A genotype is the way in which these genes are expressed. Genetic variation occurs because of independent assortment, homologue crossover, and random fertilization.

Patterns of inheritance through families are called modes of inheritance, which include autosomal recessive, autosomal dominant, and X-linked recessive. Genetic disorders usually reflect homozygous recessive conditions. Carriers are heterozygotes that do not express a trait but can pass it to their offspring. Traits determined by genes on the X and Y chromosomes are called "sex-linked traits." Gene expression can be influenced or overridden by environmental factors. Inheritance may be determined by factors other than nuclear DNA sequence, which include protein-coding genes, RNA-only genes, and epigenetic marks. Advances in genetics, including the outstanding results of the Human Genome Project, are improving treatments and even cures for a large variety of diseases and disorders linked to gene and chromosomal defects. Procedures used to check for genetic conditions include pedigrees, amniocentesis, CVS, and blood tests.

Summary

At the end of each chapter, the *Summary* recaps the most important points in the chapter and connects it to the student's overall journey.

Key Terms

Key Terms list the most important new terms covered in the chapter; correlating definitions can be found in the end of text glossary.

KEY TERMS

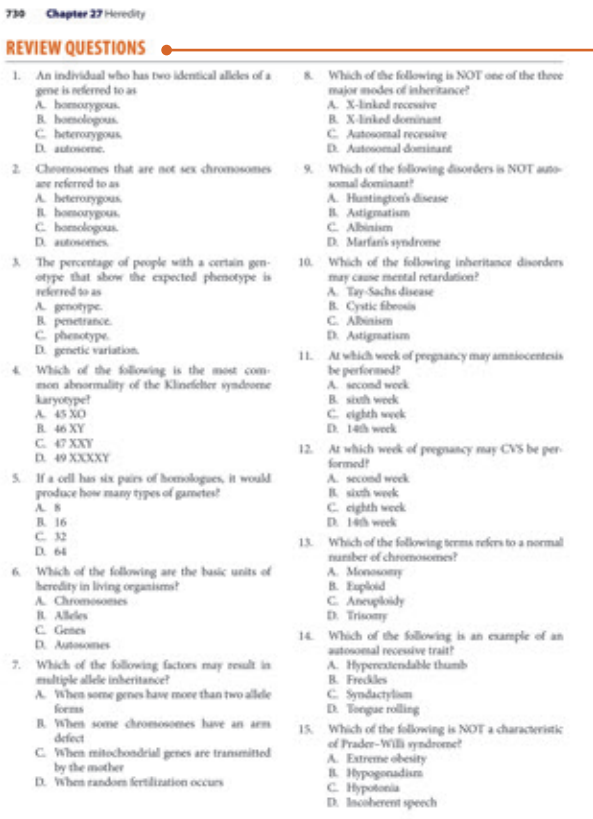
Alleles
Amniocentesis
Autosomes
Chiasma
Chorionic villus sampling (CVS)
Chromosome
Codominant
Color blindness
Congenital hip dysplasia
Cystic fibrosis
Delayed action gene
Deletion
Dominant
Dominant-recessive inheritance
Down syndrome
Dwarfism
Epigenetic marks
Gametes
Gene expression
Genetics
Genome
Genomic imprinting
Genotype
Gynecomastia
Hemizygous
Hemophilia
Heterozygous
Homologous chromosomes
Homozygous
Huntington's disease
Hypogonadism
Hyperphagia
Imperforate anus
Incomplete dominance
Independent assortment
Karyotypes
Klinefelter syndrome
Lethal dominant genes
Linked

Learning Goals

Learning Goals encapsulate how each *Objective* has been addressed over the course of the chapter.

Critical Thinking Questions

A range of questions are also included at the end of each chapter; the student can use these for self-study or submit their answers to the instructor. A case is presented at the end of each chapter, with *Critical Thinking Questions* that cause the student to reflect on the situation described.



730 Chapter 27 Heredity

REVIEW QUESTIONS

1. An individual who has two identical alleles of a gene is referred to as
   A. homozygous.
   B. homologous.
   C. heterozygous.
   D. autosome.
2. Chromosomes that are not sex chromosomes are referred to as
   A. heterozygous.
   B. homozygous.
   C. homologous.
   D. autosomes.
3. The percentage of people with a certain genotype that show the expected phenotype is referred to as
   A. genotype.
   B. penetrance.
   C. phenotype.
   D. genetic variation.
4. Which of the following is the most common abnormality of the Klinefelter syndrome karyotype?
   A. 45 XO
   B. 46 XY
   C. 47 XXY
   D. 49 XXXXY
5. If a cell has six pairs of homologues, it would produce how many types of gametes?
   A. 8
   B. 16
   C. 32
   D. 64
6. Which of the following are the basic units of heredity in living organisms?
   A. Chromosomes
   B. Alleles
   C. Genes
   D. Autosomes
7. Which of the following factors may result in multiple allele inheritance?
   A. When some genes have more than two allele forms
   B. When some chromosomes have an arm defect
   C. When mitochondrial genes are transmitted by the mother
   D. When random fertilization occurs
8. Which of the following is NOT one of the three major modes of inheritance?
   A. X-linked recessive
   B. X-linked dominant
   C. Autosomal recessive
   D. Autosomal dominant
9. Which of the following disorders is NOT autosomal dominant?
   A. Huntington's disease
   B. Astigmatism
   C. Albinism
   D. Marfan's syndrome
10. Which of the following inheritance disorders may cause mental retardation?
    A. Tay-Sachs disease
    B. Cystic fibrosis
    C. Albinism
    D. Astigmatism
11. At which week of pregnancy may amniocentesis be performed?
    A. second week
    B. sixth week
    C. eighth week
    D. 14th week
12. At which week of pregnancy may CVS be performed?
    A. second week
    B. sixth week
    C. eighth week
    D. 14th week
13. Which of the following terms refers to a normal number of chromosomes?
    A. Monosomy
    B. Euploid
    C. Aneuploidy
    D. Trisomy
14. Which of the following is an example of an autosomal recessive trait?
    A. Hyperextendable thumb
    B. Freckles
    C. Syndactylism
    D. Tongue rolling
15. Which of the following is NOT a characteristic of Prader–Willi syndrome?
    A. Extreme obesity
    B. Hypogonadism
    C. Hypotonia
    D. Incoherent speech

Essay Questions 731

ESSAY QUESTIONS

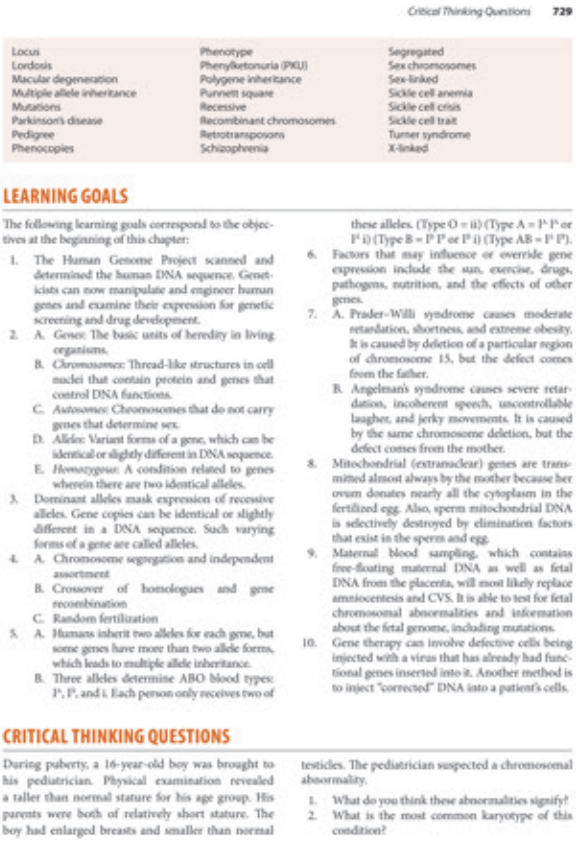
1. Explain what the nuclei of all cells, except gametes, contain.
2. Differentiate between the terms "homozygous" and "heterozygous."
3. Explain why maternal and paternal chromosomes are randomly distributed to daughter nuclei.
4. Identify the differences in how dominant and recessive traits are abbreviated.
5. Explain why recessive genetic disorders are more frequent than those caused by dominant alleles.
6. Identify the amounts of genes carried by the X and Y chromosomes and give a brief overview of their functions.
7. Discuss the genes and alleles that determine skin color.
8. Explain how altered nutrition can affect genetic expression after birth.
9. Identify which level of control is believed to influence 80% of the genome.
10. Discuss tests used to screen for the sickling gene, Tay-Sachs disease, and cystic fibrosis.

Review Questions

Review Questions provide students with a chance to answer multiple choice questions.

Essay Questions

Finally, *Essay Questions* ask students to delve deeply into the content.



Critical Thinking Questions 729

Locus	Phenotype	Segregated
Lordosis	Phenylketonuria (PKU)	Sex chromosomes
Macular degeneration	Polygene inheritance	Sex-linked
Multiple allele inheritance	Punnett square	Sickle cell anemia
Mutations	Recessive	Sickle cell crisis
Parkinson's disease	Recombinant chromosomes	Sickle cell trait
Pedigree	Retrotransposons	Turner syndrome
Phenocopies	Schizophrenia	X-linked

LEARNING GOALS

The following learning goals correspond to the objectives at the beginning of this chapter:

1. The Human Genome Project scanned and determined the human DNA sequence. Geneticists can now manipulate and engineer human genes and examine their expression for genetic screening and drug development.
2. A. *Genes*: The basic units of heredity in living organisms.
   B. *Chromosomes*: Thread-like structures in cell nuclei that contain protein and genes that control DNA functions.
   C. *Autosomes*: Chromosomes that do not carry genes that determine sex.
   D. *Alleles*: Variant forms of a gene, which can be identical or slightly different in DNA sequence.
   E. *Homozygous*: A condition related to genes wherein there are two identical alleles.
3. Dominant alleles mask expression of recessive alleles. Gene copies can be identical or slightly different in a DNA sequence. Such varying forms of a gene are called alleles.
4. A. Chromosome segregation and independent assortment
   B. Crossover of homologues and gene recombination
   C. Random fertilization
5. A. Humans inherit two alleles for each gene, but some genes have more than two allele forms, which leads to multiple allele inheritance.
   B. Three alleles determine ABO blood types: I^A, I^B, and i. Each person only receives two of these alleles. (Type O = ii) (Type A = $I^A I^A$ or I^A i) (Type B = $I^B I^B$ or I^B i) (Type AB = $I^A I^B$).
6. Factors that may influence or override gene expression include the sun, exercise, drugs, pathogens, nutrition, and the effects of other genes.
7. A. Prader–Willi syndrome causes moderate retardation, shortness, and extreme obesity. It is caused by deletion of a particular region of chromosome 15, but the defect comes from the father.
   B. Angelman's syndrome causes severe retardation, incoherent speech, uncontrollable laugher, and jerky movements. It is caused by the same chromosome deletion, but the defect comes from the mother.
8. Mitochondrial (extranuclear) genes are transmitted almost always by the mother because her ovum donates nearly all the cytoplasm in the fertilized egg. Also, sperm mitochondrial DNA is selectively destroyed by elimination factors that exist in the sperm and egg.
9. Maternal blood sampling, which contains free-floating maternal DNA as well as fetal DNA from the placenta, will most likely replace amniocentesis and CVS. It is able to test for fetal chromosomal abnormalities and information about the fetal genome, including mutations.
10. Gene therapy can involve defective cells being injected with a virus that has already had functional genes inserted into it. Another method is to inject "corrected" DNA into a patient's cells.

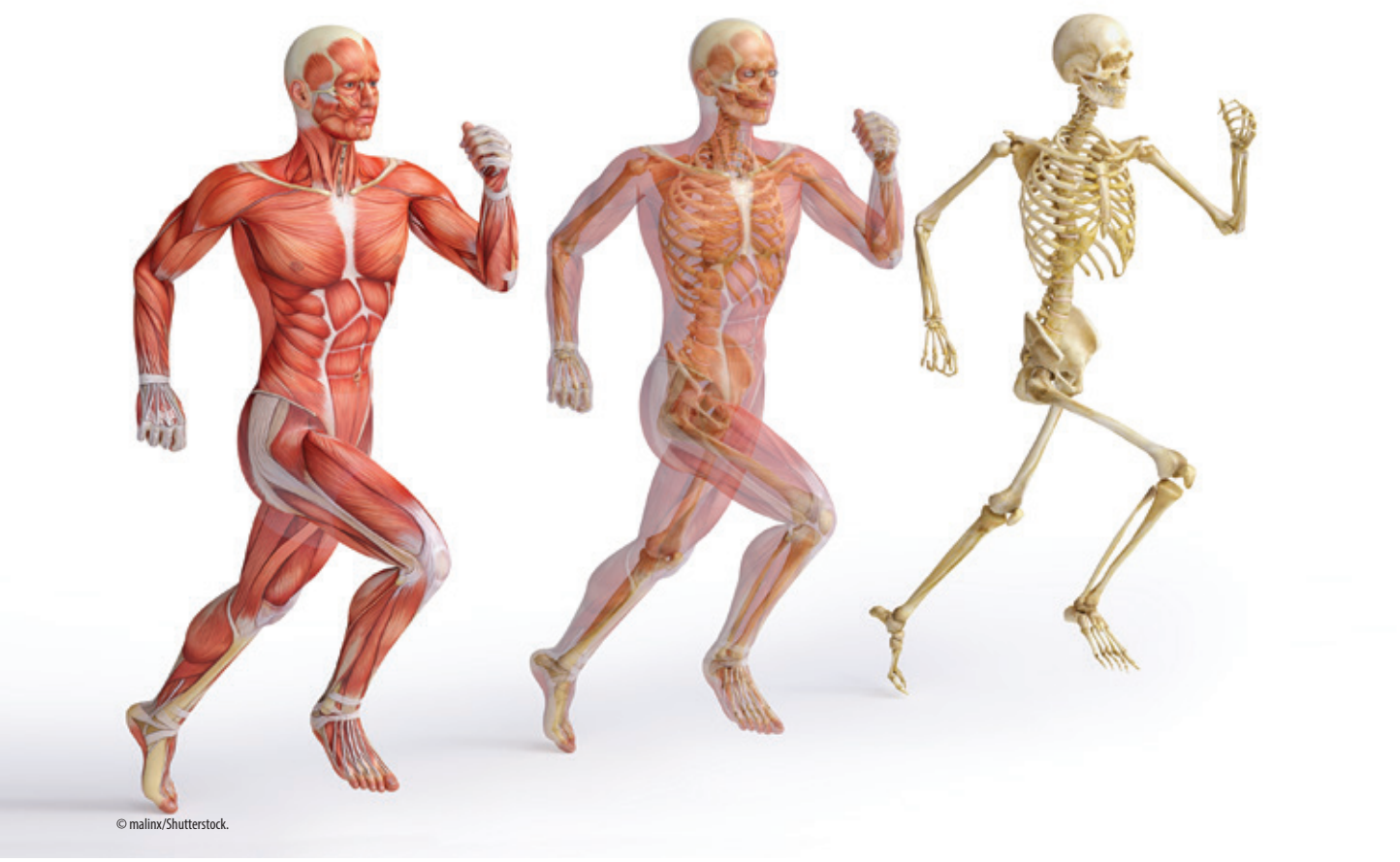
CRITICAL THINKING QUESTIONS

During puberty, a 16-year-old boy was brought to his pediatrician. Physical examination revealed a taller than normal stature for his age group. His parents were both of relatively short stature. The boy had enlarged breasts and smaller than normal testicles. The pediatrician suspected a chromosomal abnormality.

1. What do you think these abnormalities signify?
2. What is the most common karyotype of this condition?

SECTION I

Levels of Organization



CHAPTER 1

Introduction to Human Anatomy and Physiology

OUTLINE

OBJECTIVES

After studying this chapter, readers should be able to

1. Define anatomy and physiology.
2. Name the components that make up the organization levels of the body.
3. Describe the major essentials of life.
4. Define homeostasis and describe its importance to survival.
5. Describe the major body cavities.
6. List the systems of the body and give the organs in each system.
7. Describe directions and planes of the body.
8. Discuss the membranes near the heart, lungs, and abdominal cavity.
9. List the nine abdominal regions.
10. Compare positive and negative feedback mechanisms.

Overview

The study of anatomy and physiology is vital for all health professionals and it involves many different areas of science to understand how the human body works and how it is structured. The study of anatomy and physiology provides answers to many questions about the functions of the body in both health and disease. As a result of this understanding, it is possible to see what happens to the body when it is injured, stressed, or contracts a disease or infection. It is important for all allied health students to be familiar with the terminology used in anatomy and physiology. In this chapter, the focus is on a complete introduction to anatomy and physiology.

The structures and functions of the human body are closely related. **Anatomy** is the study of the structure of body parts and how they are organized. This term is derived from the Greek words meaning *to cut apart*. **Physiology** is the study of how body parts work. Every body part functions to assist the human body in different ways. It is not easy to separate the topics of anatomy and physiology because the structures of body parts are so closely associated with their functions. Each part has its own unique substructures that allow it to perform its needed functions. **Pathophysiology** is the study of changes associated with, or resulting from, disease or injury. It is also concerned with biological and physical manifestations of disease as they relate to underlying abnormalities and physiological disturbances. Pathophysiology explains the processes within the body that result in disease signs and symptoms but does not focus directly on the treatment of disease.

The human body has been studied for hundreds of years. Even though its inner workings are well understood, new discoveries are being made even today. In 2003, the human genome (instructions that allow the body to operate) was deciphered for the first time. There are more than 20,000 genes in the human body, and this substantial discovery took many years to complete. Researchers frequently discover new information about physiology, particularly at the molecular level, but basic human anatomy changes very slowly.

Classifications of Anatomy

The many subdivisions of anatomy include gross (macroscopic) anatomy, microscopic anatomy, and developmental anatomy. These can be further broken down as follows:

- **Gross (macroscopic) anatomy:** The study of large body structures that can be seen without a microscope. These include the brain, heart, kidneys, lungs, and skin. Studies conducted to understand gross anatomy made use of dissected animals and their organs.
  - **Regional anatomy:** All structures in a certain body region are examined at the same time. For example, for an arm, the structures being examined would include skin, muscles, bones, nerves, blood vessels, and others.
  - **Systemic anatomy:** Each body system is examined. For example, the heart would be examined when studying the cardiovascular system, but so would all the blood vessels in the body.
  - **Surface anatomy:** This is the examination of internal structures related to overlying skin surfaces. Surface anatomy is used, for example, to locate the correct blood vessels used for phlebotomy.
- **Microscopic anatomy:** The study of small body structures that require a microscope to be seen.

This requires making thin slices of tissues, which are then stained and affixed (mounted) to glass slides for microscopic examination.
  - **Cytology:** A subdivision of microscopic anatomy that focuses on body cells.
  - **Histology:** A subdivision of microscopic anatomy that focuses on body tissues.
- **Developmental anatomy:** The study of structural changes in anatomy throughout the life span.
  - **Embryology:** A subdivision of developmental anatomy that focuses on developmental changes occurring before birth.

For medical diagnosis, scientific research, and other highly specialized needs, *pathological* or *radiographic* anatomy may be used. Pathological anatomy focuses on disease and the structural changes that are a result of the disease, whereas radiographic anatomy focuses on internal structures via the use of X-rays or specialized scanning equipment such as magnetic resonance imaging (MRI) or computed tomography (CT). *Molecular anatomy* focuses on the structure of chemical substances (biological molecules). Although formally considered a branch of *biology*, molecular anatomy is still part of the overall study of anatomy as it focuses on subcellular particles of the body.

Anatomical studies require a combination of many different skills. These include anatomic terminology, observation, *auscultation* (using a stethoscope to listen to organ sounds), manipulation, and *palpation* (feeling body organs for normal or abnormal conditions by using the hands).

Classifications of Physiology

Physiology is concerned with how the body functions, often focusing on cellular or molecular activities. There are also many subdivisions of physiology, which are primarily focused on certain organ systems. Examples of physiology classifications are as follows:

- *Respiratory physiology:* Focuses on the functions of the respiratory system
- *Cardiovascular physiology*: Focuses on the heart and blood vessels
- *Neurophysiology*: Focuses on the nervous system
- *Renal physiology*: Focuses on the functions of the kidneys, including urine production

The physiology of the human body is based on chemical reactions that affect the actions of cells at the molecular level. Physiology is also linked to the study of physics, which takes into account body functions such as blood pressure, electrical currents, and muscular movement.

TEST YOUR UNDERSTANDING

1. Describe the difference between gross anatomy and developmental anatomy.
2. Compare cytology and histology.
3. Explain the classifications of physiology.

Organization Levels of the Body

Every body structure is made up of smaller structures, which are, likewise, made up of even smaller components. Chemicals compose every material found in the human body. They contain microscopic **atoms** combined into structures known as **molecules**. Many molecules may be combined into macromolecules. These macromolecules, in turn, form **organelles**, which help to complete the intended functions of a **cell**, the basic unit of both structure and function in the human body.

Cells are microscopic structures that can vary in size, shape, and function. Cells are grouped together to form **tissues**, which, in turn, are grouped together to form **organs**. Groups of similarly functioning organs form **organ systems**, which then combine to form a living **organism** (**FIGURE 1-1**). Body parts are organized into different levels of complexity, including the *atomic level*, *molecular level*, and *cellular level*. Atoms are the most simple in structure, with complexity increasing in molecules, organelles, tissues, and organs.

At *tissue level*, the tissues work together and perform one or more specific functions. For example, cardiac muscle cells interact within the heart with other types of cells. They also interact with extracellular materials, forming cardiac muscle tissue. At the *organ level*, the tissues of an organ work together, performing several functions. For example, layers of cardiac muscle tissue work with connective tissue to form most of the wall of the heart, which is a hollow, three-dimensional organ. At the *organ system level*, groups of organs interact and perform particular functions. For example, each contraction of the heart pushes blood into the blood vessel network. The heart, along with the blood and blood vessels, comprise the cardiovascular system. This is one of the 11 organ systems of the human body.

TEST YOUR UNDERSTANDING

1. Describe the major levels of organization of the body.
2. Identify the tissue level and the organ system level.

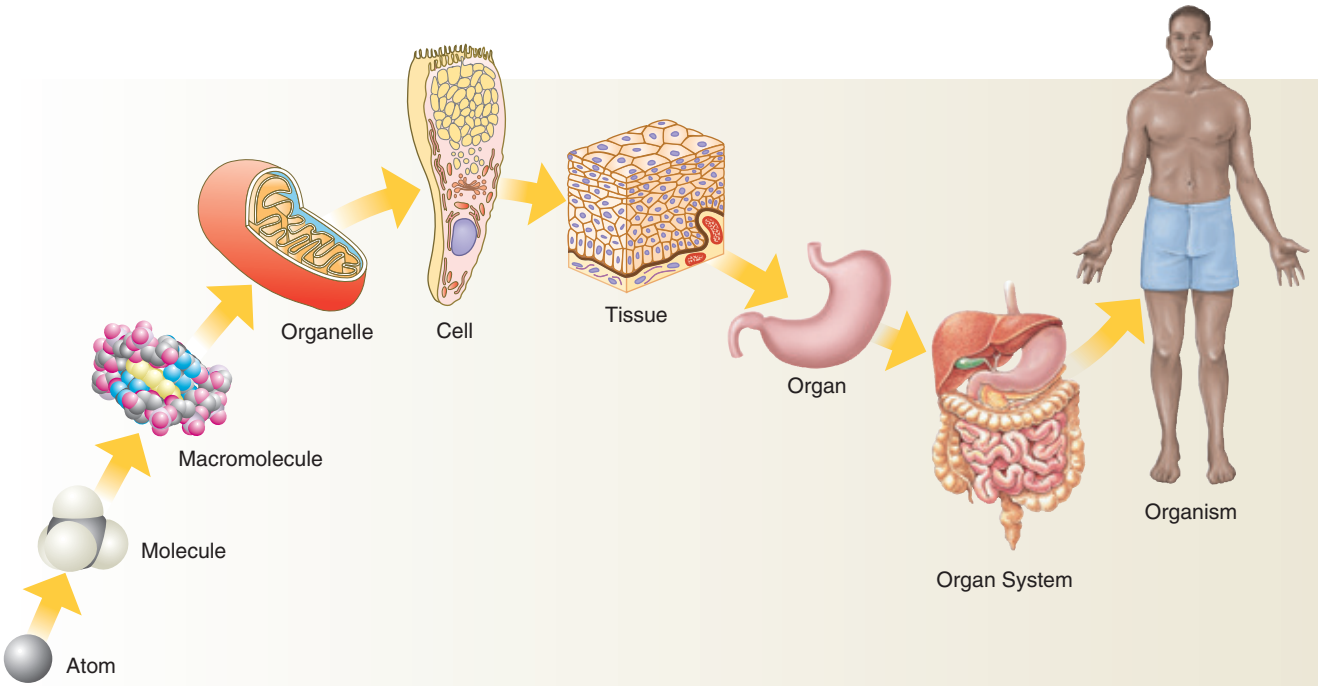


FIGURE 1-1 Organization levels of the body.

Adapted from Shier, D.N., Butler, J.L., and Lewis, R. Hole's Essentials of Human Anatomy & Physiology, Tenth edition. McGraw Hill Higher Education, 2009.

Essentials for Life

Humans and other animals share many similar traits. All body cells are interdependent as we are multicellular organisms. Vital body functions occur over various organ systems, which contribute to overall body health.

Boundaries

The body's boundaries are maintained to keep the internal environment distinct from the external environment. All body cells are surrounded by selectively permeable membranes. The skin encloses and protects the body as a whole from factors such as dryness, bacteria, heat, sunlight, and chemicals.

Movement

Movement of the body is achieved via the muscular and skeletal systems. Inside the body, the cardiovascular, digestive, and urinary systems too use movement to transport blood, food materials, and urine. Even cells move, such as when muscle cells move by shortening, which is known as *contractility*.

Responsiveness

The ability to sense and respond to environmental stimuli (changes) is known as *responsiveness*, which is also referred to as *excitability*. An example is the way we quickly withdraw our hands from a hot saucepan. Nerve cells are highly excitable. They communicate with rapid electrical impulses, and, therefore, the nervous system is the most responsive of all body systems. However, all body systems have some degree of excitability.

Digestion

Humans require specific nutrients to remain healthy and to grow and develop normally. Energy is gained from the breakdown, digestion, absorption, and assimilation of food. Digestion breaks down food materials to simple, more easily absorbed molecules. Absorbed nutrients move throughout the body's circulation. Nutrient-rich blood is distributed, via the cardiovascular system, to the entire body. Respiration brings in oxygen that works with nutrients to grow and repair body parts. The unusable parts of these processes are then excreted as waste.

Metabolism

The body's **metabolism** controls all these processes. It includes all chemical reactions inside body cells, the breaking down of substances into simpler forms (*catabolism*), creating more complex cellular components from simpler substances (*anabolism*), and the use of nutrients and oxygen to produce energy-rich adenosine triphosphate (ATP) molecules (via *cellular respiration*).

In metabolism, nutrients and oxygen from the digestive and respiratory systems are circulated to all body cells. Hormones from the endocrine system glands have strong regulatory control over metabolism.

Excretion

The process of removing wastes from the body is known as *excretion*. Nonessential substances that are produced during digestion and metabolism must be removed. The digestive system removes food components that cannot be digested via the feces. The urinary system removes urea and other metabolic wastes containing nitrogen via the urine. The blood carries carbon dioxide to the lungs for it to be exhaled.

Reproduction

Reproduction is a process that occurs at several levels. At the cellular level, reproduction means cell division. Cells divide to produce two identical daughter cells, which the body uses for growth and repair. At the organism level, the human reproduction system unites a sperm with an egg. A fertilized egg is formed, developing into a baby inside the body of the mother. The function of the production of offspring is controlled by endocrine system hormones. Reproductive structures differ between the sexes, with the female structures providing a fertilization site for the male sperm cells. The female reproductive structures protect the developing fetus and nurture its growth until birth.

Growth

An increase in the size of an organism or its body parts is called *growth*. Most often, growth is achieved by an increase in the amount of cells. In fact, even when the cells do not divide, they can increase in size. True growth occurs when constructive activities occur more quickly than destructive activities. The various characteristics of life are listed in **TABLE 1-1**.

Survival

Human beings need several substances for survival: food (nutrients), water, oxygen, pressure, and heat in specific quantities and with specific qualities.

Nutrients

Food provides nutrients for energy, growth, and regulation of the chemical reactions in the body. Some of these chemicals are used as energy sources or supply the raw materials needed for building new living matter; other chemicals help to regulate vital chemical reactions. Plant-based foods contain high levels of carbohydrates, vitamins, and minerals. Carbohydrates are the primary energy fuel for body cells. Certain vitamins and minerals are needed for chemical reactions inside cells and for oxygen transport in the blood. Calcium is a mineral that assists in making bones harder and is needed for blood clotting. Animal-based foods contain high levels of proteins and fats. Proteins

TABLE 1-1 Characteristics of Human Life

Characteristic	Examples
Movement	Change in positions of the body or its parts; motion of internal organs
Responsiveness	Reaction to changes, inside or outside the body
Digestion	Breakdown of foods into simpler forms for absorption and usage
Excretion	Removal of wastes from metabolic reactions
Growth	Increases in body size, without changes in shape
Respiration	Obtaining oxygen and removing carbon dioxide; releasing energy from foods
Absorption	Movement of substances through membranes into body fluids
Circulation	Movement of substances in body fluids
Assimilation	Change of absorbed substances into various chemical forms

are the most essential component required for building cell structures. Fats assist in this process and are a great source of energy-providing fuel for the body.

Water

Water is required for metabolic processes and makes up most of the body's actual structure, transporting substances and regulating temperature. It accounts for 60% to 80% of body weight and is the most abundant chemical in the body. Water allows chemical reactions to occur and is also the fluid base for secretions and excretions. Water is mostly obtained from ingested liquids or foods, and is lost in the urine, by evaporation from the lungs and skin, and also in other body excretions.

Oxygen

Oxygen is a gas that drives metabolic processes by releasing energy from food that is consumed and by bringing nutrients to cells throughout the body. This energy release involves *oxidative* reactions, for which oxygen is required. Therefore, all nutrients require oxygen for them to be effectively used. Human cells only survive for a few minutes without oxygen. Oxygen makes up approximately 20% of the air that we breathe. It is made available to the blood and body cells by both the respiratory and cardiovascular systems. Appropriate amounts of oxygen sustain life, but even oxygen may be toxic in excessive quantities.

Atmospheric Pressure

Appropriate pressure, specifically atmospheric pressure, is essential for breathing and gas exchange. Blood pressure is a form of hydrostatic pressure that forces the blood through the veins and arteries. Atmospheric pressure may be defined as the force that air exerts upon the body's surface. Gas exchange, in higher altitudes, may be insufficient to support cellular metabolism because at these altitudes, atmospheric pressure is lower and the air is thinner. At sea level, the average atmospheric pressure is 760 mm of mercury (Hg).

Body Temperature

Heat energy is produced from metabolic reactions, influencing their speed. The muscular system generates the most body heat. Body heat is measured as temperature. Normal body temperature must be maintained if chemical reactions are to sustain life continually. If the temperature is too high, chemical reactions occur very quickly, and proteins in the body change shape and cease functioning. If body temperature drops below 98.6°F (37°C), metabolic reactions slow down and eventually stop. Death may occur also because of either variation in temperature.

TEST YOUR UNDERSTANDING

1. What factors are necessary to sustain life in humans?
2. What elements are needed by the body for survival?
3. Describe metabolism and the effect of atmospheric pressure on the body.

Homeostasis

The internal environment of the human body must stay relatively stable for the person to survive. **Homeostasis** is a term that describes a stable internal body environment. It requires a constant balance. There must be normal concentrations of nutrients, oxygen, and water. Heat and pressure must be regulated at tolerable levels. **Homeostatic mechanisms** regulate the body by negative or **positive feedback.**

Homeostatic Control

For homeostasis to occur, the body primarily uses the nervous and endocrine systems. These systems allow forms of communication that control homeostasis to occur. The nervous system uses neural electrical impulses for these activities, whereas the endocrine system uses blood borne hormones. The nervous system handles rapid, short-term, extremely specific responses. The endocrine system responds more slowly, but its effects last for a longer duration. The event or factor that is being controlled (regulated) is referred to as the *variable*.

The two basic components of homeostatic control are as follows:

- *Autoregulation*: Also known as *intrinsic regulation*, this occurs when a body structure or system adjusts its activities because of some change in its environment. For example, declining tissue oxygen levels cause cells to release chemicals that widen local blood vessels, thereby increasing blood flow rate, which provides more oxygen to the local area of the body.
- *Extrinsic regulation*: Related to nervous or endocrine system activity, these systems influence activities of many other body systems simultaneously. For example, increased exercise causes the nervous system to increase the heart rate and circulate

blood more quickly. It also reduces blood flow to the digestive tract and other less active organs. Oxygen in the circulating blood is then available to the active muscles, which need it the most.

All mechanisms used for homeostatic control involve at least three components:

- ***Receptors***: These are "sensors" that monitor the internal body environment and respond to stimuli. Receptors send information to the *control center* along the *afferent pathway*. You can remember this more easily because the afferent pathway carries information that is "approaching" the control center.
- *Control center*: This is a point in the body that determines the *set point* (the range or level at which a variable must be maintained). It analyzes the input from the receptors to determine appropriate responses or actions. It then sends information to *effectors* via the *efferent pathway*. You can remember this more easily because the efferent pathway carries information that is "exiting" the control center (**FIGURE 1-2**). The set point for the average body temperature, for example, is 98.6°F (37°C). Another set point is normal adult blood pressure, which is ideally below 120 (systolic) and under 80 (diastolic).
- *Effectors*: These are components of homeostatic control that allow the control center to respond to stimuli. The control center's response involves negative (reducing) or positive (enhancing) feedback. Basically, negative feedback shuts off the control process, whereas positive feedback makes it occur at a faster rate.

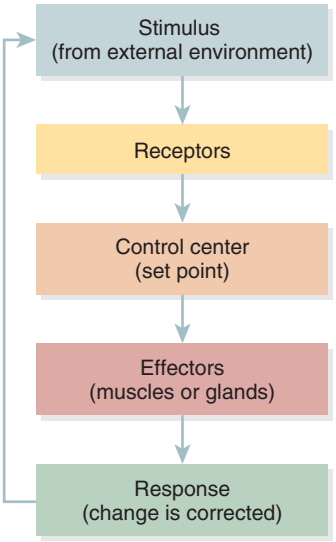


FIGURE 1-2 Homeostatic mechanism.

Negative Feedback

A **negative feedback** mechanism is one that prevents the correction of deviations from doing too much (which could possibly harm the body). Most of the feedback mechanisms of the human body use negative feedback. Examples of negative feedback are blood pressure regulation, erythropoiesis (the production of red blood cells), body temperature regulation (thermoregulation), and control of blood glucose levels. The *hypothalamus* of the brain maintains homeostatic control of body temperature. Information is received from temperature receptors in the skin and within the hypothalamus itself. The normal *set point* of body temperature is approximately 98.6°F (37°C). When temperature rises above this normal, hypothalamic activity targets muscle tissue in walls of blood vessels that supply the skin, and also targets the sweat glands. This causes blood flow to increase near the body surface, and acceleration of sweat gland secretion. The skin loses heat to the environment, and sweat evaporation speeds up this process. As temperature lowers back to normal, hypothalamic activity declines, and all processes reverse.

Negative feedback is the main controller of homeostasis, providing long-term control over internal systems and body conditions. Minor variations are usually ignored, while normal body ranges are maintained instead of exact, fixed values. The regulatory process works dynamically since set points vary with changes in environment and activity. While sleeping, thermoregulation has a lower set point than when you are awake and active. Therefore, temperature varies because of small fluctuations around the set point, or change in the set point. Similar variations occur throughout all body physiology.

Set points differ between individuals based on age, gender, genetic factors, overall health, and the environment. There are no actual *precise* homeostatic conditions. Basically, homeostatic values are either based on average between large amounts of people, or as a range including 95% (or more) of those people being sampled. While most healthy adults have body temperature between 98.1°F and 98.9°F (between 36.7°C and 37.2°C), 5% have resting body temperatures above or below this range.

Positive Feedback

A **positive feedback** mechanism is one that makes conditions move away from the normal state to stimulate further changes. They are usually short-lived and extremely specific actions, producing extreme responses. A positive feedback mechanism is defined as one that results in or responds in an enhanced way

to the original stimulus, accelerating the result or response. Examples of positive feedback are the onset of contractions before childbirth, the process of blood clotting, lactation, the secretion of estrogen during the follicular phase of menstruation, and the generation of nerve signals.

In positive feedback, cycles *escalate* and are often referred to as being part of a **positive feedback loop**. These loops are usually found when a possibly stressful or dangerous body process must be completed quickly prior to homeostasis being restored. One example is a severe laceration, which may lower blood pressure and reduce the heart's effectiveness. As the clotting process attempts to combat the loss of blood, a positive feedback loop occurs, which increases the clotting activities.

Homeostatic Imbalance

Most diseases occur because of *homeostatic imbalance* (meaning the disturbance of homeostasis). Aging causes body systems to become less efficient and more uncontrollable, resulting in instability in the internal body environment and increasing the risk for illness. Also, when helpful negative feedback mechanisms become overwhelmed, certain destructive positive feedback mechanisms can dominate (such as those seen in some forms of heart failure). Additional examples of homeostatic imbalance include abdominal injury due to physical trauma (and lack of protective bones in this body region), sepsis (resulting in severe pain, such as in **peritonitis**), and metabolic acidosis or alkalosis (which can affect all body systems and lead to death). Trauma may involve hemorrhage and perforation of abdominal organs. Any cause of homeostatic imbalance can result in death if untreated.

FOCUS ON PATHOLOGY

Abdominopelvic organs are frequently damaged because of physical trauma, such as in a vehicular accident. The organs of the upper chest or pelvis are protected by bones, but only the abdominal muscles form the walls of the abdominal cavity.

TEST YOUR UNDERSTANDING

1. Define homeostasis.
2. Why is homeostasis essential to survival?
3. Describe two homeostatic mechanisms.

Organization of the Body

The human body is composed of distinct body parts, cavities, membranes, and organ systems that include various body systems. All of these are discussed in greater detail in the following sections.

Body Cavities and Membranes

The body is divided into two main cavities, the dorsal cavity and the ventral cavity. These two main cavities are divided into smaller subcavities. The dorsal cavity protects the organs of the nervous system. Its two subdivisions include the *cranial cavity* of the skull, which encases the brain, and the *vertebral (spinal) cavity*, located inside the vertebral column, which encases the spinal cord. The vertebral cavity is also referred to as the *vertebral canal.* The cranial and spinal cavities are in continuation with each other. The ventral cavity contains most of the body's organs. More anterior and larger than the dorsal cavity, it houses the **viscera** (visceral organs). The ventral cavity is divided into the *thoracic cavity* and the *abdominopelvic cavity*.

Thoracic Cavity

The thoracic cavity is surrounded by the chest muscles and ribs, and contains the lungs and heart; organs of the cardiovascular, respiratory, and lymphatic systems; inferior esophagus; and the thymus. It is subdivided into lateral **pleural cavities**, which surround each lung, and the medial **mediastinum**, which is a tissue mass that separates the cavities. Each pleural cavity is lined by a **serous membrane**, which is shiny and slippery, and functions to reduce friction as the lung expands and recoils during breathing. The *pleura* is the serous membrane lining a pleural cavity. The **visceral** *pleura* covers the outer lung surfaces. The **parietal** *pleura* covers the inner body wall and mediastinal surface.

The mediastinum is a mass of connective tissue surrounding and protecting the esophagus, trachea, thymus, and major blood vessels originating or ending at the heart. It also contains a small chamber surrounding the heart, which is called the **pericardial cavity**. The attached portion of the heart is called the *base*. The serous membrane of the heart is the *pericardium*, subdivided into the *visceral pericardium* (covering the heart) and its opposing surface, the *parietal pericardium*. As the heart changes size and shape while beating, the pericardial cavity also changes. Friction is prevented by the slipper