Anatomy & Physiology

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Fifth Edition

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Anatomy & Physiology

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made in credibly *Easy!*



Fifth Edition

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Dedication

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Dedicated to my family: without you I could not have made it to where I am and I am so grateful. You are my joy!

Laura M. Willis

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Foreword

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If you're like me, you're too busy to wade through a foreword that uses pretentious terms and umpteen dull paragraphs to get to the point. So let's cut right to the chase! Here's why this book is so terrific:

- 1. It will teach you all the important things you need to know about anatomy and physiology without tons of tiny details to wade through.
- 2. It will help you remember what you've learned.
- 3. It will make you smile as it enhances your knowledge and skills. Don't believe me? Try these recurring logos on for size:



Zoom in-provides a close look at anatomic structures



Body shop—helps explain how body systems and structures work together



Now I get it!—converts complex physiology into easy-to-digest explanations



Senior moment—pinpoints the effects of aging on anatomy and physiology



Memory jogger—reinforces learning through acronyms and other tools that aid recall



Just for fun—reinforces understanding of anatomical terminology and pathophysiological concepts

See? I told you! And that's not all. Look for me and my friends in the margins throughout this book. We'll be there to explain key concepts, provide important care reminders, and offer reassurance. Oh, and if you don't mind, we'll be spicing up the pages with a bit of humor along the way, to teach and entertain in a way that no other resource can.

I hope you find this book helpful. Best of luck throughout your career!

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Chapter 1

The human body

Just the facts

In this chapter, you'll learn:

 anatomic terms for direction, reference planes, body cavities, and body regions to help describe the locations of various body structures

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- the structure of cells
- cell reproduction and energy generation
- four basic tissue types and their characteristics.

Anatomic terms

Anatomic terms describe directions within the body as well as the body's reference planes, cavities, and regions.

Directional terms

When navigating the body, directional terms help you determine the exact location of a structure.

Couples at odds

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Generally, directional terms can be grouped in pairs of opposites:

- *Superior* and *inferior* mean above and below, respectively. For example, the shoulder is superior to the elbow, and the hand is inferior to the wrist.
- *Anterior* means toward the front of the body, and *posterior* means toward the back. *Ventral* is sometimes used instead of anterior, and *dorsal* is sometimes used instead of posterior.
- *Medial* means toward the body's midline and *lateral* means away from it.
- *Proximal* and *distal* mean closest and farthest, respectively, to the point of origin (or to the trunk).
- *Superficial* and *deep* mean toward or at the body surface and farthest from it.

Locating body structures starts with directional terms, reference planes, cavities, and regions.



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To remember the meanings of **proximal** and **distal**, keep in mind that when something is in **proximity**, it's nearby. When something is **distant**, it's far away.

2 The human body

Reference planes

Reference planes are imaginary lines used to section the body and its organs. These lines run longitudinally, horizontally, and angularly. The four major body reference planes are:

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1. median sagittal

- 2. frontal
- 3. transverse
- 4. oblique. (See Picturing body reference planes.)

Body shop

[©]Picturing body reference planes

Body reference planes are used to indicate the locations of body structures. Shown here are the median sagittal, frontal, and transverse planes. An oblique plane—a slanted plane that lies between a horizontal plane and a vertical plane—isn't shown.



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Body cavities

Body cavities are spaces within the body that contain the internal organs. The *dorsal* and *ventral cavities* are the two major closed cavities—cavities without direct openings to the outside of the body. (See *Locating body cavities*.)

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Dorsal cavity

The dorsal cavity is located in the posterior region of the body.

The think tank and backbone of the operation

The dorsal cavity is further subdivided into two cavities:

- The *cranial cavity* (also called the *calvaria*), formed by the skull, encases the brain.
- The *vertebral cavity* (also called the *spinal cavity* or *vertebral canal*), formed by the vertebrae, encloses the spinal cord.



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Locating body cavities

The dorsal cavity, in the posterior region of the body, is divided into the cranial and vertebral cavities. The ventral cavity, in the anterior region, is divided into the thoracic and abdominopelvic cavities. These regions are shown in the illustration given below.



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Ventral cavity

The ventral cavity occupies the anterior region of the trunk. This cavity is subdivided into the *thoracic cavity* and the *abdominopelvic cavity*.

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Treasure chest

Surrounded by the ribs and chest muscles, the thoracic cavity refers to the space located superior to the abdominopelvic cavity. It's subdivided into the *pleural cavities* and the *mediastinum*:

- Each of the two *pleural cavities* contains a lung.
- The *mediastinum* houses the heart, large vessels of the heart, trachea, esophagus, thymus, lymph nodes, and other blood vessels and nerves.

The bread basket and below

The abdominopelvic cavity has two regions, the *abdominal cavity* and the *pelvic cavity*:

- The *abdominal cavity* contains the stomach, intestines, spleen, liver, and other organs.
- The *pelvic cavity*, which lies inferior to the abdominal cavity, contains the bladder, some of the reproductive organs, and the rectum.

Other cavities

The body also contains an *oral cavity* (the mouth), a *nasal cavity* (located in the nose), *orbital cavities* (which house the eyes), *middle ear cavities* (which contain the small bones of the middle ear), and the *synovial cavities* (enclosed within the capsules surrounding freely moveable joints).

Body regions

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Body regions are used to designate body areas that have special nerves or vascular supplies or those that perform special functions.

The guts of the matter

The most widely used body region terms are those that designate the sections of the abdomen. (See *Abdominal regions exposed*.) The abdomen has nine regions:

- The *umbilical region*, the area around the umbilicus, includes sections of the small and large intestines, inferior vena cava, and abdominal aorta.
- The *epigastric region*, superior to the umbilical region, contains most of the pancreas and portions of the stomach, liver, inferior vena cava, abdominal aorta, and duodenum.



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• The *hypogastric region* (or pubic area), inferior to the umbilical region, houses a portion of the sigmoid colon, the urinary bladder and ureters, the uterus and ovaries (in females), and portions of the small intestine.

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- The right and left *iliac regions* (or inguinal regions) are situated on either side of the hypogastric region. They include portions of the small and large intestines.
- The right and left *lumbar regions* (or loin regions) are located on either side of the umbilical region. They include portions of the small and large intestines and portions of the kidneys.
- The right and left *hypochondriac regions,* which reside on either side of the epigastric region, contain the diaphragm, portions of the kidneys, the right side of the liver, the spleen, and part of the pancreas.

Remember, each region has a specific nerve or vascular supply or performs a special function.



Body shop

Abdominal regions exposed

Here's an anterior view of the abdominal regions.



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A look at the cell

The *cell* makes up the body's structure and serves as the basic unit of living matter. Human cells vary widely, ranging from the simple squamous epithelial cell to the highly specialized neuron.

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The greatest regeneration

Generally, the simpler the cell, the greater its power to regenerate. The more specialized the cell, the weaker its regenerative power. Cells with greater regenerative power have shorter life spans than do those with less regenerative power.

Cell structure

Cells are made up of three basic components:

- 1. protoplasm
- 2. plasma membrane
- 3. nucleus. (See Inside the cell.)

Protoplasm

Protoplasm, a viscous, translucent, watery material, is the primary component of plant and animal cells. It contains a large percentage of water, inorganic ions (such as potassium, calcium, magnesium, and sodium), and naturally occurring organic compounds (such as proteins, lipids, and carbohydrates).

Getting charged

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The inorganic ions within protoplasm are called *electrolytes*. They regulate acid-base balance and control the amount of intracellular water. When these ions lose electrons (minute particles with a negative charge), they acquire a positive electrical charge. When they gain electrons, they acquire a negative electrical charge. The most common electrolytes in the body are sodium (Na⁺), potassium (K⁺), and chloride (Cl⁻)

A pair of "plasms"

Nucleoplasm is the protoplasm of the cell's nucleus. It plays a part in reproduction. *Cytoplasm* is the protoplasm of the cell body that surrounds the nucleus. It converts raw materials to energy. It's also the site of most synthesizing activities. In the cytoplasm, you'll find *cytosol*, *organelles*, and *inclusions*.



Sometimes simpler is better. The simpler I am, the more I can regenerate!





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This cross section shows the components and structures of a cell. As noted, each component plays a part in maintaining the health of the cell.

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A cytosol sea

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Cytosol is a viscous, semitransparent fluid that's 70% to 90% water. It contains proteins, salts, and sugars.

A lot to metabolize

Organelles are the cell's metabolic units. Each organelle performs a specific function to maintain the life of the cell:

- *Mitochondria* are structures within the cytoplasm that provide most of the body's adenosine triphosphate—the enzyme that fuels many cellular activities.
- *Ribosomes* are the sites of protein synthesis.
- The *endoplasmic reticulum* is an extensive network of membrane-enclosed tubules. *Rough endoplasmic reticulum* is covered with ribosomes and produces certain proteins. *Smooth endoplasmic reticulum* contains enzymes that synthesize lipids.



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• Each *Golgi apparatus* synthesizes carbohydrate molecules. These molecules combine with the proteins produced by rough endoplasmic reticulum to form secretory products such as lipoproteins.

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- *Lysosomes* are digestive bodies that break down foreign or damaged material in cells. (See *Lysosomes at work*.)
- *Peroxisomes* contain *oxidases*, enzymes capable of reducing oxygen to hydrogen peroxide and hydrogen peroxide to water.
- Cytoskeletal elements form a network of protein structures.
- *Centrosomes* contain *centrioles*, short cylinders that are adjacent to the nucleus and take part in cell division.

Temps that don't do any work

Inclusions are nonfunctioning units in the cytoplasm that are commonly temporary. The pigment *melanin* in epithelial cells and the stored nutrient *glycogen* in liver cells are both examples of nonfunctioning units.

Now I get it!

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Lysosomes at work

Lysosomes are the organelles responsible for digestion within a cell. Phagocytes assist in this process. Here's how lysosomes work.

Function of lysosomes

Lysosomes are digestive bodies that break down foreign or damaged material in cells. A membrane surrounds each lysosome and separates its digestive enzymes from the rest of the cytoplasm.

Breaking it down

The lysosomal enzymes digest matter brought into the cell by *phagocytes*, special cells that surround and engulf matter outside the cell and then transport it through the cell membrane. The membrane of the lysosome fuses with the membrane of the cytoplasmic spaces surrounding the phagocytized material; this fusion allows the lysosomal enzymes to digest the engulfed material.

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Plasma membrane

The *plasma membrane* (cell membrane) is the gatekeeper of the cell. It serves as the cell's external boundary, separating it from other cells and from the external environment.

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Checkpoint

Nothing gets by this semipermeable membrane without authorization from the nucleus. The semipermeable membrane consists of a double layer of phospholipids with protein molecules.

Nucleus

The *nucleus* is the cell's mission control. It plays a role in cell growth, metabolism, and reproduction.

A nucleus may contain one or more *nucleoli*—a dark-staining structure that synthesizes *ribonucleic acid* (RNA). The nucleus also contains *chromosomes*. Chromosomes control cellular activity and direct protein synthesis through ribosomes in the cytoplasm. (For more information on chromosomes, see chapter 2, Genetics.)

DNA and RNA

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Protein synthesis is essential for the growth of new tissue and the repair of damaged tissue. *Deoxyribonucleic acid* (DNA) carries genetic information and provides the blueprint for protein synthesis. RNA transfers this genetic information to the ribosomes, where protein synthesis occurs.

Touching all the bases

The basic structural unit of DNA is a *nucleotide*. Nucleotides consist of a phosphate group that's linked to a five-carbon sugar, *deoxyribose*, and joined to a nitrogen-containing compound called a *base*. Four

different DNA bases exist:

- 1. adenine (A)
- 2. guanine (G)
- 3. thymine (T)
- 4. cytosine (C).

That means we fit together perfectly.

complementary.

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10 The human body

Identifying rings

Adenine and *guanine* are double-ring compounds classified as *purines. Thymine* and *cytosine* are single-ring compounds classified as *pyrimidines.*

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The chain gangs

DNA chains exist in pairs held together by weak chemical attractions between the nitrogen bases on adjacent chains. Because of the chemical shape of the bases, adenine bonds only with thymine and guanine bonds only with cytosine. Bases that can link with each other are called *complementary*.

Insider trading

RNA consists of nucleotide chains that differ slightly from the nucleotide chains found in DNA. Several types of RNA are involved in the transfer (to the ribosomes) of genetic information essential to protein synthesis. (See *Types of RNA*.)



Types of RNA

There are three types of ribonucleic acid (RNA): ribosomal, messenger, and transfer. Each has its own specific function.

Ribosomal RNA

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Ribosomal RNA is used to make ribosomes in the endoplasmic reticulum of the cytoplasm, where the cell produces proteins.

Messenger RNA

Messenger RNA directs the arrangement of amino acids to make proteins at the ribosomes. Its single strand of nucleotides is complementary to a segment of the deoxyribonucleic acid chain that contains instructions for protein synthesis. Its chains pass from the nucleus into the cytoplasm, attaching to ribosomes there.

Transfer RNA

Transfer RNA consists of short nucleotide chains, each of which is specific for an individual amino acid. Transfer RNA transfers the genetic code from messenger RNA for the production of a specific amino acid.

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Cell reproduction

Cells are under a constant call to reproduce; it's either that or die. Cell division is how cells reproduce (or replicate) themselves; they achieve this through the process of *mitosis* or *meiosis*.

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DNA does its thing

Before a cell divides, its chromosomes are duplicated. During this process, the double helix separates into two DNA chains. Each chain serves as a template for constructing a new chain. Individual DNA nucleotides are linked into new strands with bases complementary to those in the original.



Double double

In this way, two identical double helices are formed, each containing one of the original strands and a newly formed



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Z DNA up close

Linked deoxyribonucleic acid (DNA) chains form a spiral structure, or double helix.

A spiral staircase

To understand linked DNA chains, imagine a spiral staircase. The deoxyribose and phosphate groups form the railings of the staircase, and the nitrogen base pairs (adenine and thymine, guanine and cytosine) form the steps.

Cell division

Each chain serves as a template for constructing a new chain. When a cell divides, individual DNA nucleotides are linked into new strands with bases complementary to those in the originals. In this way, two identical double helices are formed, each containing one of the original strands and a newly formed complementary strand. These double helices are duplicates of the original DNA chain.



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complementary strand. These double helices are duplicates of the original DNA chain. (See DNA up close, page 11.)

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Mitosis

Mitosis is the equal division of material in the nucleus (*karyokinesis*) followed by division of the cell body (*cytokinesis*). It's the preferred mode of replication by all cells in the human body, except the gametes. Cell division occurs in five phases, an inactive phase called *interphase*, and four active phases:

- 1. prophase
- 2. metaphase
- 3. anaphase
- 4. telophase.

Two daughters equal 46

Mitosis results in two daughter cells (exact duplicates), each containing 23 pairs of chromosomes—or 46 individual chromosomes. This number is the *diploid number*. (See *Divide and conquer: five stages of mitosis*.)

Meiosis

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Meiosis is reserved for gametes (ova and spermatozoa). This process intermixes genetic material between homologous chromosomes, producing four daughter cells, each with the *haploid number* of chromosomes (23, or half of the 46). Meiosis has two divisions separated by a resting phase.

First division

The first division has six phases and begins with one parent cell. When the first division ends, the result is two daughter cells—each containing the haploid (23) number of chromosomes.

Division 2, the sequel

The second division is a four-phase division that resembles mitosis. It starts with two new daughter cells, each containing the haploid number of chromosomes, and ends with four new haploid cells. In each cell, the two chromatids of each chromosome separate to form new daughter cells. However, because each cell entering the second division has only 23 chromosomes, each daughter cell formed has only 23 chromosomes. (See *Meiosis: Step-by-step*, page 14.)



To help you remember the difference between haploid and diploid think of the prefix **di**- in diploid. **Di**- means **double**, so diploid cells have double the number of chromosomes in a haploid cell.

He may think he's better than me, but we have identical DNA!

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Now I get it!

Divide and conquer: five stages of mitosis

Through the process of mitosis, the nuclear content of all body cells (except gametes) reproduces and divides. The result is the formation of two new daughter cells, each containing the diploid (46) number of chromosomes.

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Interphase

During *interphase*, the nucleus and nuclear membrane are well defined, and the nucleolus is visible. As chromosomes replicate, each forms a double strand that remains attached at the center by a centromere.

Prophase

In *prophase*, the nucleolus disappears and the chromosomes become distinct. *Chromatids*, halves of each duplicated chromosome, remain attached by the centromere. Centrioles move to opposite sides of the cell and radiate spindle fibers.

Metaphase

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Metaphase occurs when chromosomes line up randomly in the center of the cell between the spindles, along the *metaphase plate*. The centromere of each chromosome then replicates.

Anaphase

Anaphase is characterized by centromeres moving apart, pulling the separate chromatids (now called *chromosomes*) to opposite ends of the cell. The number of chromosomes at each end of the cell equals the original number.

Telophase

During *telophase*, the final stage of mitosis, a nuclear membrane forms around each nucleus and spindle fibers disappear. The cytoplasm compresses and divides the cell in half. Each new cell contains the diploid (46) number of chromosomes.



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<u> Now I g</u>et it!

Meiosis: Step-by-step

Meiosis has two divisions that are separated by a resting phase. By the end of the first division, two daughter cells exist that each contains the haploid (23) number of chromosomes. When the second division ends, each of the two daughter cells from the first division divides, resulting in four daughter cells, each containing the haploid number of chromosomes.

First division

The first division has six phases. Here's what happens during each one.

Interphase

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- 1. Chromosomes replicate, forming a double strand attached at the center by a centromere.
- 2. Chromosomes appear as an indistinguishable matrix within the nucleus.

3. Centrioles appear outside the nucleus.

Prophase I

- 1. The nucleolus and nuclear membrane disappear.
- 2. Chromosomes are distinct, with chromatids attached by the centromere.

3. Homologous chromosomes move close together and intertwine;

exchange of genetic information (genetic recombination) may occur. 4. Centrioles separate and spindle fibers appear.

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Metaphase I

1. Pairs of synaptic chromosomes line up randomly along the meta-phase plate.

2. Spindle fibers attach to each chromosome pair.

Anaphase I

1. Synaptic pairs separate.

2. Spindle fibers pull homologous, double-stranded chromosomes to opposite ends of the cell.

3. Chromatids remain attached.

Telophase I

1. The nuclear membrane forms.

2. Spindle fibers and chromosomes disappear.

3. Cytoplasm compresses and divides the cell in half.

4. Each new cell contains the haploid (23) number of chromosomes.

Interkinesis

1. The nucleus and nuclear membrane are well defined.

2. The nucleolus is prominent, and each chromosome has two chromatids that don't replicate.

Second division

The second division closely resembles mitosis and is characterized by these four phases.

Prophase II

1. The nuclear membrane disap-

- pears.
- 2. Spindle fibers form.

3. Double-stranded chromosomes appear as thin threads.

Metaphase II

1. Chromosomes line up along the metaphase plate.

2. Centromeres replicate.

Anaphase II

1. Chromatids separate (now a

single-stranded chromosome).

2. Chromosomes move away from each other to the opposite ends of the cell.

Telophase II

1. The nuclear membrane forms.

2. Chromosomes and spindle fibers disappear.

3. Cytoplasm compresses, dividing the cell in half.

4. Four daughter cells are created, each of which contains the haploid (23) number of chromosomes.

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Cellular energy generation

All cellular function depends on energy generation and transportation of substances within and among cells.

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Cellular power

Adenosine triphosphate (ATP) serves as the chemical fuel for cellular processes. ATP consists of a nitrogen-containing compound (adenine) joined to a five-carbon sugar (ribose), forming adenosine. Adenosine is joined to three phosphate (or triphosphate) groups. Chemical bonds between the first and second phosphate groups and between the second and third phosphate groups contain abundant energy.

The three R's

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ATP needs to be converted to *adenosine diphosphate* (ADP) to produce energy. To understand this conversion, remember the three R's:

- Rupture—ATP is converted to ADP when the terminal high-energy phosphate bond ruptures.
- *Release*—Because the third phosphate is liberated, energy stored in the chemical bond is released.
- *Recycle*—Mitochondrial enzymes then reconvert ADP and the liberated phosphate to ATP. To obtain the energy needed for this reattachment, mitochondria oxidize food nutrients. This makes recycled ATP available again for energy production.

Movement within cells

Each cell interacts with body fluids through the interchange of substances.

Modes of transportation

Several transport methods—*diffusion, osmosis, active transport,* and *endocytosis*—move substances between cells and body fluids. In another method, *filtration,* fluids and dissolved substances are transferred across capillaries into *interstitial fluid* (fluid in the spaces between cells and tissues).

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Diffusion

In *diffusion*, solutes move from an area of higher concentration to one of lower concentration. Eventually, an equal distribution of solutes between the two areas occurs.

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Go with the flow

Diffusion is a form of passive transport—no energy is required to make it happen; it just happens. It's kind of like fish traveling downstream. They just go with the flow. (See *Understanding passive transport*.)

Advancing and declining rates

Several factors influence the rate of diffusion:

• *concentration gradient*—the greater the concentration gradient (the difference in particle concentration on either side of the plasma membrane), the faster the rate of diffusion No energy is required for diffusion so I'll just go with the flow.



Now I get it!

Understanding passive transport

No energy is required for passive transport. It occurs through two mechanisms: diffusion and osmosis.

Diffusion

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In diffusion, substances move from an area of higher concentration to an area of lower concentration. Movement continues until distribution is uniform.

Osmosis

In osmosis, fluid moves from an area of higher concentration to one of lower concentration.



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• *particle size*—the smaller the particles, the faster the rate of diffusion

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• *lipid solubility*—the more lipid-soluble the particles are, the more rapidly they diffuse through the lipid layers of the cell membrane.

Osmosis

Osmosis is the passive transport of fluid across a membrane, from an area of lower solute concentration (comparatively *more* fluid) into an area of higher solute concentration (comparatively *less* fluid).

Enough is enough

Osmosis stops when enough fluid has moved through the membrane to equalize the solute concentration on both sides of the membrane.

Active transport

Active transport requires energy. Usually, this mechanism moves a substance across the cell membrane against the concentration gradient—from an area of lower concentration to one of higher concentration. Think of active transport as swimming upstream. When a fish swims upstream, it has to expend energy.

ATP at it again

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The energy required for a solute to move against a concentration gradient comes from ATP. ATP is stored in all cells and supplies energy for solute movement in and out of cells. (See *Understanding active transport*, page 18.)

It goes both ways

However, active transport also can move a substance with the concentration gradient. In this process, a carrier molecule in the cell membrane combines with the substance and transports it through the membrane, depositing it on the other side.

Endocytosis

Endocytosis is an active transport method in which, instead of passing through the cell membrane, a substance is engulfed by the cell. The cell surrounds the substance with part of the cell membrane. This part separates to form a *vacuole* (cavity) that moves to the cell's interior. Several factors influence the rate of diffusion concentration gradient, particle size, and lipid solubility.



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Now I get it!

Understanding active transport

Active transport moves molecules and ions against a concentration gradient from an area of lower concentration to one of higher concentration. This movement requires energy, usually in the form of adenosine triphosphate (ATP). The sodium-potassium pump and pinocytosis are examples of active transport mechanisms.

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Sodium-potassium pump

The sodium-potassium pump moves sodium from inside the cell to outside, where the sodium concentration is greater; potassium moves from outside the cell to inside, where the potassium concentration is greater.

Pinocytosis

In pinocytosis, tiny vacuoles take droplets of fluid containing dissolved substances into the cell. The engulfed fluid is used in the cell.



Gobbling up particles

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Endocytosis involves either *phagocytosis* or *pinocytosis*. Phagocytosis refers to engulfment and ingestion of particles that are too large to pass through the cell membrane. Pinocytosis occurs only to engulf dissolved substances or small particles suspended in fluid.

Filtration

Fluid and dissolved substances also may move across a cell membrane by *filtration*.

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Pressure is the point

In filtration, pressure (provided by capillary blood) is applied to a solution on one side of the cell membrane. The pressure forces fluid and dissolved particles through the membrane. The rate of filtration (how quickly substances pass through the membrane) depends on the amount of pressure. Filtration promotes the transfer of fluids and dissolved materials from the blood across the capillaries into the interstitial fluid.

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A look at human tissue

Tissues are groups of cells that perform the same general function. The human body contains four basic types: *epithelial, connective, muscle,* and *nervous tissue*.

Epithelial tissue

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Epithelial tissue (epithelium) is a continuous cellular sheet that covers the body's surface, lines body cavities, and forms certain glands. Imagining a mummy wrapped in strips of cloth will give you some idea of how epithelial tissue covers the human body. (See *Distinguishing types of epithelial tissue*, page 20.)

Patrolling the borders

Some columnar epithelial cells in the lining of the intestines have vertical striations, forming a *striated border*. In the tubules of the kidneys, borders of columnar epithelial cells have tiny, brushlike structures (microvilli) called a *brush border*.

This hair isn't just for looks

Two common types of cells that form epithelial tissue are *stereociliated* and *ciliated epithelial cells*. The former line the epididymis and have long, piriform (pear-shaped) tufts. The latter possess *cilia*, fine hairlike protuberances. Cilia are larger than microvilli and move fluid and particles through the cavity of an organ.

Endothelium

Epithelial tissue with a single layer of squamous cells attached to a basement membrane is called *endothelium*. Such tissue lines the heart, lymphatic vessels, and blood vessels.

A crowd of us cells working together on the same function is called tissue.



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Z Distinguishing types of epithelial tissue

Epithelial tissue (epithelium) is classified by the number of cell layers and the shape of surface cells. Some types of epithelium go through a process of desquamation (shedding of debris) and regenerate continuously by transformation of cells from deeper layers.

Identified by number of cell layers

Classified by number of cell layers, epithelium may be *simple* (one-layered), *stratified* (multilayered), or *pseudostratified* (one-layered but appearing to be multilayered).

Classified by shape

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If classified by shape, epithelium may be *squamous* (containing flat surface cells), *columnar* (containing tall, cylindrical surface cells), or *cuboidal* (containing cubeshaped surface cells).

The top left illustration below shows how the basement membrane of simple squamous epithelium joins the epithelium to underlying connective tissues. The remaining illustrations show the five other types of epithelial tissue.



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Glandular epithelium

Organs that produce secretions consist of a special type of epithelium called *glandular epithelium*.

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The secret is in how it secretes

Glands are classified as endocrine or exocrine according to how they secrete their products.

- *Endocrine glands* release their secretions into the blood or lymph. For instance, the medulla of the adrenal gland secretes epinephrine and norepinephrine into the bloodstream.
- *Exocrine glands* discharge their secretions into ducts that lead to external or internal surfaces. For example, the sweat glands secrete sweat onto the surface of the skin.

Mixing it up

Mixed glands contain both endocrine and exocrine cells. The pancreas is a mixed gland. As an endocrine gland, it produces insulin and glucagon. As an exocrine gland, it introduces pancreatic juices into the intestines.

Connective tissue

Connective tissue—a category that includes bone, cartilage, and adipose (fatty) tissue—binds together and supports body structures. Connective tissue is classified as *loose* or *dense*.

Cut loose

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Loose (areolar) connective tissue has large spaces that separate the fibers and cells. It contains a lot of intercellular fluid.

Dense tissue issues

Dense connective tissue provides structural support and has greater fiber concentration. Dense tissue is further subdivided into dense regular and dense irregular connective tissue:

- *Dense regular* connective tissue consists of tightly packed fibers arranged in a consistent pattern. It includes tendons, ligaments, and *aponeuroses* (flat fibrous sheets that attach muscles to bones or other tissues).
- *Dense irregular* connective tissue has tightly packed fibers arranged in an inconsistent pattern. It's found in the dermis, submucosa of the GI tract, fibrous capsules, and fasciae.



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22 The human body

Suppose it's adipose

Commonly called *fat, adipose tissue* is a specialized type of loose connective tissue where a single lipid (fat) droplet occupies most of each cell. Widely distributed subcutaneously, it acts as insulation to conserve body heat, as a cushion for internal organs, and as a storage depot for excess food and reserve supplies of energy. Adipose tissue also functions as endocrine tissue because it produces the appetite-suppressing hormone leptin.



Muscle tissue consists of muscle cells with a generous blood supply. Muscle cells measure up to several centimeters long and have an elongated shape that enhances their *contractility* (ability to contract).

The tissues at issue

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There are three basic types of muscle tissue:

- 1. *Striated muscle tissue* gets its name from its striped, or striated, appearance; it contracts voluntarily.
- 2. *Cardiac muscle tissue* is sometimes classified as striated because it's also composed of striated tissue. However, it differs from other striated muscle tissue in two ways: its fibers are separate cellular units that don't contain many nuclei, and it contracts involuntarily.
- 3. *Smooth muscle tissue* consists of long, spindle-shaped cells and lacks the striped pattern of striated tissue. Its activity is stimulated by the autonomic nervous system and isn't under voluntary control.

Wall tissue paper

Smooth muscle tissue lines the walls of many internal organs and other structures, including the respiratory passages from the trachea to the alveolar ducts, the urinary and genital ducts, the arteries and veins, the larger lymphatic trunks, the intestines, the arrectores pilorum, and the iris and ciliary body of the eye.

Nervous tissue

The main function of *nervous tissue* is communication. Its primary properties are *irritability* (the capacity to react to various physical and



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chemical agents) and *conductivity* (the ability to transmit the resulting reaction from one point to another).

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Nervous tissue specialists

Neurons are highly specialized cells that generate and conduct nerve impulses. A typical neuron consists of a cell body with cytoplasmic extensions—numerous *dendrites* on one pole and a single *axon* on the opposite pole. These extensions allow the neuron to conduct impulses over long distances.

Protecting neurons

Neuroglia form the support structure of nervous tissue, insulating and protecting neurons. They're found only in the central nervous system.



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Quick quiz

1. The reference plane that divides the body lengthwise into right and left regions is the:

- A. frontal plane.
- B. sagittal plane.
- C. transverse plane.
- D. oblique plane.

Answer: B. Imaginary lines called *reference planes* are used to section the body. The sagittal plane runs lengthwise and divides the body into right and left regions.

- 2. The structure that plays the biggest role in cellular function is the:
 - A. nucleus.
 - B. Golgi apparatus.
 - C. ribosome.
 - D. mitochondrion.

Answer: A. Serving as the cell's control center, the nucleus plays a role in cell growth, metabolism, and reproduction.

- 3. The four basic types of tissue that the human body contains are:
 - A. muscle, cartilage, glandular, and connective tissue.
 - B. bone, cartilage, glands, and adipose tissue.
 - C. loose, dense connective, dense regular, and dense irregular tissue.
 - D. epithelial, connective, muscle, and nervous tissue.

Answer: D. Tissues are groups of cells with the same general function. The human body contains four basic types of tissue: epithelial, connective, muscle, and nervous tissue.



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- 4. Meiosis ends when:
 - A. two new daughter cells form, each with the haploid number of chromosomes.
 - B. one daughter cell forms and is an exact copy of the original.

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- C. four new daughter cells form, each with the haploid number of chromosomes.
- D. four new daughter cells form, each with the diploid number of chromosomes.

Answer: C. Meiosis comes to completion with the end of telophase II. The result is four daughter cells, each of which contains the haploid (23) number of chromosomes.

Scoring

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☆☆☆ If you answered all four questions correctly, fantastic! You're well on your way to a fantastic voyage through the human body.

- ☆☆ If you answered three questions correctly, all right! You're in for some smooth sailing. Pretty soon you'll know the body inside and out.
 - ☆ If you answered fewer than three questions correctly, buck up, camper. With plenty more quick quizzes to go, you'll conquer this body of knowledge in no time.

Selected References

Hall, J. (2015). *Guyton and Hall textbook of medical physiology* (13th ed.). Philadelphia, PA: Elsevier.

Saladin, K. (2014). Anatomy & physiology: The unity of form and function (7th ed.). New York, NY: McGraw Hill. ()

Chapter 2

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Genetics

Just the facts

In this chapter, you'll learn:

- the way traits are transmitted
- the role of chromosomes and genes in heredity
- factors that determine trait predominance
- causes of genetic defects.

A look at genetics

Genetics is the study of heredity—the passing of traits from biological parents to their children. People inherit not only physical traits, such as eye color, but also biochemical and physiologic traits, including the tendency to develop certain diseases or conditions. Genetic information is carried in *genes*, which are strung together on the *deoxyribonucleic acid* (DNA) double helix to form chromosomes.

Family inheritance

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Parents transmit inherited traits to their offspring in germ cells, or *gametes*. There are two types of human gametes: eggs (ova) and sperm (spermatozoa).

Chromosomes

The nucleus of each germ cell contains structures called *chromosomes*. Each chromosome contains a strand of genetic material called *DNA*. DNA is a long molecule that's made up of thousands of segments called *genes*. These genes carry the code for proteins that influence each trait a person inherits, ranging from blood type to toe shape. Chromosomes exist in pairs except in the germ cells.

Your genes determine how you look, how your body functions, and even whether you're prone to certain diseases.



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Counting chromosomes

A human ovum contains 23 chromosomes. A sperm also contains 23 chromosomes, each similar in size and shape to a chromosome in the ovum. When an ovum and a sperm unite, the corresponding chromosomes pair up. The result is a fertilized cell with 46 chromosomes (23 pairs) in its nucleus.

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Gen XX (or XY)

Of the 23 pairs of chromosomes in each living human cell, the two sex chromosomes of the 23rd pair determine a person's gender. The other 22 pairs are called *autosomes*.

In a female, both sex chromosomes are relatively large and each is designated by the letter X. In a male, one sex chromosome is an X chromosome and one is a smaller chromosome, designated by the letter Y.

Each gamete produced by a male contains either an X or a Y chromosome. Each gamete produced by a female contains an X chromosome. When a sperm with an X chromosome fertilizes an ovum, the offspring is female (two X chromosomes). When a sperm with a Y chromosome fertilizes an ovum, the offspring is male (one X and one Y chromosome).

Dividing the family assets

Ova and sperm are formed by a cell-division process called *meiosis*. In meiosis, each of the 23 pairs of chromosomes in a cell splits. The cell then divides, and each new cell (an ovum or sperm) receives one set of 23 chromosomes. (See chapter 1, The human body, page 14, for more information on meiosis.)

Genes

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Genes are segments of a DNA chain, arranged in sequence on a chromosome. This sequence determines the properties of an organism.

Locus pocus

The location of a specific gene on a chromosome is called the *gene locus*. The locus of each gene is specific and doesn't vary from person to person. This allows each of the thousands of genes in an ovum to join the corresponding genes from a sperm when the chromosomes pair up at fertilization.

How do I look?

The genetic information stored at a locus of a gene determines the genetic constitution—or *genotype*—of a person. The detectable, outward manifestation of a genotype is called the *phenotype*.

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The genome at a glance

The human genome is made up of a set of very long deoxyribonucleic acid (DNA) molecules, one for each chromosome. DNA has four different chemical building blocks, called *bases*. Each human genome contains about 3 billion of these bases, arranged in an order that's unique for each person. Increasing its complexity, arrayed along the DNA molecules are over 30,000 genes.

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Consider its size: If the DNA sequence of the human genome were compiled in books, the equivalent of 200 volumes the size of the Manhattan telephone book (at 1,000 pages each) would be needed to hold it all.

The human genome

One complete set of chromosomes, containing all the genetic information for one person, is called a *genome*. (See *The genome at a glance*.)

The 411 on genes

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For several years, scientists intensely studied the human genome to determine the entire sequence of each DNA molecule and the location and identity of all genes. The project was successfully completed in April 2003.

Catching the culprit

Genetic sequencing information allows practitioners to identify the causes of disease rather than simply treating symptoms. Other benefits include future development of more specific diagnostic tests, the formation of new therapies, and methods for avoiding conditions that trigger disease.

In addition, practitioners can now test patients for a gene error, present in 1 out of 500 people, that indicates an increased risk of developing colon cancer. Similarly, individuals with personal or family histories of breast or ovarian cancer can be tested for genetic predispositions to those diseases. Researchers are also seeking genes associated with dozens of other diseases, including chronic conditions such as asthma and diabetes.

Trait predominance

Each parent contributes one set of chromosomes (and therefore one set of genes) to his or her offspring. Therefore, every offspring has two genes for every locus (location on the chromosome) on the autosomal chromosomes. If I were to read out loud the 3 billion bases in just one genome, it would take 9½ years.



lt only takes one bad gene to spoil a gene pool party. ()

28 Genetics

Variation is the spice of life

Some characteristics, or traits, are determined by one gene that may have many variants. Variations of the same gene are called *alleles*. A person who has identical alleles on each chromosome is *homozygous* for that trait; if the alleles are different, they're said to be *heterozygous*. Other traits—called *polygenic traits*—require the interaction of more than one gene.

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Autosomal inheritance

On autosomal chromosomes, one allele may exert more influence in determining a specific trait. This is called the *dominant gene*. The less influential allele is called the *recessive gene*. Offspring express the trait of a dominant allele if both, or only one, chromosome in a pair carries it. For a recessive allele to be expressed, both chromosomes must carry recessive versions of the alleles. (See *How genes express themselves*.)

Sex-linked inheritance

The X and Y chromosomes are the sex chromosomes. The X chromosome is much larger than the Y. Therefore, males (who have XY chromosomes) have less genetic material than do females (who have

Now I get it!

How genes express themselves

Genes account for inherited traits. *Gene expression* refers to a gene's effect on cell structure or function; however, the effects vary with the gene.

Dominant genes

If genes could speak, dominant genes would be loud and garrulous, dominating every conversation! Dominant genes (such as the one for dark hair) can be expressed and transmitted to the offspring even if only one parent possesses the gene.

Recessive genes

Unlike dominant genes, recessive genes prefer to hide their light under a bushel basket. A recessive gene (such as the one for blond hair) is expressed only when both parents transmit it to the offspring.

Codominant genes

Firm believers in equality, codominant genes (such as the genes that direct specific types of hemoglobin synthesis in red blood cells) allow expression of both alleles.

Sex-linked genes

Sex-linked genes are carried on sex chromosomes. Almost all appear on the X chromosome and are recessive. In the male, sex-linked genes behave like dominant genes because no second X chromosome exists. ()

XX chromosomes), which means they have only one copy of most genes on the X chromosome. Inheritance of those genes is called *X-linked*, or *sex-linked*, *inheritance*.

Unequal X-change

A woman transmits one copy of each X-linked gene to each of her children, male or female. Because a man transmits an X chromosome only to his female children (male children receive a Y chromosome), he transmits X-linked genes only to his daughters, never his sons.

Multifactorial inheritance

Multifactorial inheritance reflects the interaction of at least two genes and the influence of environmental factors.

Raising the bar

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Height is a classic example of a multifactorial trait. In general, the height of an offspring peaks between the heights of the two parents. However, nutritional patterns, health care, and other environmental factors also influence development of such traits as height. A better-nourished, healthier child of two short parents may be taller than either parent.

When it all comes together

Some diseases also have genetic predispositions for multifactorial inheritance; that is, the gene for a disease might be expressed only under certain environmental conditions.

Factors that may contribute to multifactorial inheritance include:

- use of drugs, alcohol, or hormones by either parent
- maternal smoking
- maternal or paternal exposure to radiation
- maternal infection during pregnancy
- preexisting diseases in the mother
- nutritional factors
- general maternal or paternal health
- maternal-fetal blood incompatibility
- inadequate prenatal care.

Genetic defects

Genetic defects are defects that result from changes to genes or chromosomes. They're categorized as autosomal disorders, sex-linked disorders, or multifactorial disorders. Some defects arise spontaneously, Good nutrition, proper health care, and other environmental factors can influence how a gene expresses itself.

whereas others may be caused by environmental teratogens. *Teratogens* are environmental agents (such as infectious toxins, maternal diseases, drugs, chemicals, and physical agents) that can cause structural or functional defects in a developing fetus. They may also cause spontaneous abortion, complications during labor and delivery, hidden defects in later development (such as cognitive or behavioral problems), and benign or cancerous tumors.

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Permanent change in plans

A permanent change in genetic material is known as a *mutation*. Mutations can result from exposure to radiation, certain chemicals, or viruses. They may also happen spontaneously and can occur anywhere in the genome.

Every cell has built-in defenses against genetic damage. However, if a mutation isn't identified or repaired, it may produce a new trait that can be transmitted to offspring. Some mutations produce no effect, others change the expression of a trait, and others can even change the way a cell functions. Some mutations cause serious or deadly defects, such as congenital anomalies and cancer.

Autosomal disorders

In autosomal disorders, an error occurs at a single gene site on the DNA strand. Single-gene disorders are inherited in clearly identifiable patterns that are the same as those seen in inheritance of normal traits. Because every person has 22 pairs of autosomes and only 1 pair of sex chromosomes, most hereditary disorders are caused by autosomal defects.

The assertive type

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Autosomal dominant transmission involves transmission of an abnormal gene that's dominant. Autosomal dominant disorders usually affect male and female offspring equally. Children with one affected parent have a 50% chance of being affected.

Passive-aggressive behavior

Autosomal recessive inheritance involves transmission of a recessive gene that's abnormal. Autosomal recessive disorders also usually affect male and female offspring equally. If both parents are affected, all their offspring will be affected. If both parents are unaffected but carry the defective gene, each child has a 25% chance of being affected. If only one parent is affected and the other isn't a carrier, none of the offspring will be affected, but all will carry the defective gene. If one parent is affected and the other is a carrier, 50% of their children will be affected. Because of this transmission pattern, ()

autosomal recessive disorders may occur even when there's no family history of the disease.

Sex-linked disorders

Genetic disorders caused by genes located on the sex chromosomes are termed *sex-linked disorders*.

X calls the shots

Most sex-linked disorders are controlled by genes on the X chromosome, usually as recessive traits. Because males have only one X chromosome, a single X-linked recessive gene can cause disease to be exhibited in a male. Females receive two X chromosomes, so they may be homozygous for a disease allele (and exhibit the disease), homozygous for a normal allele (and neither have nor carry the disease), or heterozygous (carry, but not exhibit, the disease).

Most people who express X-linked recessive traits are males with unaffected parents. In rare cases, the father is affected and the mother is a carrier. All daughters of an affected male are carriers. An affected male never transmits the trait to his son. Unaffected male children of a female carrier don't transmit the disorder.

Evidence in history

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With X-linked dominant inheritance, evidence of the inherited trait usually exists in the family history. A person with the abnormal trait must have one affected parent. If the father has an X-linked dominant disorder, all of his daughters and none of his sons will be affected. If a mother has an X-linked dominant disorder, each of her children has a 50% chance of being affected.

Multifactorial disorders

Most multifactorial disorders result from a number of genes and environmental influences acting together. In polygenic inheritance, each gene has a small additive effect and the combination of genetic errors is unpredictable. Multifactorial disorders can result from a less-thanoptimum expression of many different genes, not from a specific error.

Mixing it up

Some multifactorial disorders are apparent at birth, such as cleft lip, cleft palate, congenital heart disease, anencephaly, clubfoot,

Most people who have X-linked recessive disorders are males. That's because males have only one X chromosome. Females have a second X chromosome that overpowers the "diseased" X. ۲

Now I get it!

Chromosomal disjunction and nondisjunction

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This illustration shows disjunction and nondisjunction of an ovum. When disjunction proceeds normally, fertilization with a normal sperm results in a zygote with the correct number of chromosomes. In nondisjunction, the duplicating chromosomes fail to separate; the result is one trisomic cell and one monosomic cell.



and myelomeningocele. Others, such as type II diabetes mellitus, hypertension, hyperlipidemia, most autoimmune diseases and many cancers, don't appear until later. Environmental factors most likely influence the development of multifactorial disorders during adulthood.

Chromosome defects

Aberrations in chromosome structure or number cause a class of disorders called *congenital anomalies*, or *birth defects*. Genetic aberrations include the loss, addition, or rearrangement of genetic material. ۲

Most clinically significant chromosome aberrations arise during meiosis, an incredibly complex process that can go wrong in many ways. Potential contributing factors include maternal age, radiation, and use of some therapeutic or recreational drugs.

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Unequally yoked

Translocation, the relocation of a segment of a chromosome to a nonhomologous chromosome, occurs when chromosomes split apart and rejoin in an abnormal arrangement. The cells still have a normal amount of genetic material, so often there are no visible abnormalities. However, the children of parents with translocated chromosomes may have serious genetic defects, such as monosomies or trisomies.

A *monosomy* is a condition in which the number of chromosomes present is one less than normal; an autosomal monosomy is incompatible with life. The presence of an extra chromosome is called a *trisomy*. A mixture of both abnormal and normal cells results in *mosaicism* (two or more cell lines in the same person). The effects of mosaicism depend on the number and location of abnormal cells. Some common translocation outcomes are Down's syndrome and trisomy 13.

Breaking up is hard to do

During both meiosis and mitosis, chromosomes normally separate in a process called *disjunction*. Failure to separate, called *nondisjunction*, causes an unequal distribution of chromosomes between the two resulting cells. If nondisjunction occurs soon after fertilization, it may affect all the resulting cells. The incidence of nondisjunction increases with parental age. (See *Chromosomal disjunction and nondisjunction*.)



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- 1. What's the total number of chromosomes in a fertilized cell?
 - A. 12
 - B. 23
 - C. 46
 - D. 52

Answer: C. There are 46 chromosomes (23 pairs) in the nucleus of a fertilized cell.

2. According to genetic theory, if a child has cystic fibrosis (CF), this must mean:

- A. both parents transmit the gene for CF.
- B. one parent transmits the gene for CF.
- C. one grandparent has CF.
- D. neither parent has the gene for CF.

Answer: A. Because the trait for CF is a recessive gene, it's only expressed when both parents transmit it to the offspring.

- 3. The presence of an extra chromosome is called:
 - A. monosomy.
 - B. trisomy.
 - C. mosaicism.
 - D. nondisjunction.

Answer: B. Trisomy is the presence of an extra chromosome; it results from nondisjunction.

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- 4. Which definition applies to the term *mutation*?
 - A. An environmental agent responsible for a genetic defect
 - B. A permanent change in genetic material
 - C. Interaction of at least two abnormal genes
 - D. Expression of a recessive gene in an offspring

Answer: B. A mutation is a permanent change in genetic material that may result from exposure to radiation, certain chemicals, or viruses. Mutations may also occur spontaneously.

5. A child has brown eyes and brown hair. This description reveals the child's:

- A. phenotype.
- B. genotype.
- C. genome.
- D. autosomes.

Answer: A. Phenotype refers to the outward, detectable manifestation of a person's genetic makeup or genotype.

Scoring

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☆☆☆ If you answered all five questions correctly, excellent! When it comes to genetics, you're a gene-ius.

☆☆ If you answered four questions correctly, good job! Your understanding of genetics is definitely dominant.

☆ If you answered fewer than four questions correctly, don't worry. Another look at the chapter may help to X-plain Y.

Selected References

Hall, J. (2015). *Guyton and Hall textbook of medical physiology* (13th ed.). Philadelphia, PA: Elsevier.

Saladin, K. (2014). Anatomy & physiology: The unity of form and function (7th ed.). New York, NY: McGraw Hill. ()

Chapter 3

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Chemical organization



In this chapter, you'll learn:

the chemical composition of the body

- the structure of an atom
- differences between inorganic and organic compounds.

A look at body chemistry

The human body is composed of chemicals; in fact, all of its activities are chemical in nature. To understand the human body and its functions, you must understand chemistry.

Comes down to chemistry

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The chemical level is the simplest and most important level of structural organization. Without the proper chemicals in the proper amounts, body cells—and eventually the body itself—would die.

Principles of chemistry

Every cell contains thousands of different chemicals that constantly interact with one another. Differences in chemical composition differentiate types of body tissue. Furthermore, the blueprints of heredity (deoxyribonucleic acid [DNA] and ribonucleic acid [RNA]) are encoded in chemical form.

What's the matter?

Matter is anything that has mass and occupies space. It may be a solid, liquid, or gas.

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Without chemicals in the proper amounts, I would die.

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Energetic types

Energy is the capacity to do work—to put mass into motion. It may be *potential energy* (stored energy) or *kinetic energy* (the energy of motion). Types of energy include chemical, electrical, and radiant.

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Chemical composition

An *element* is matter that can't be broken down into simpler substances by normal chemical reactions. All forms of matter are composed of chemical elements. Each of the chemical elements in the periodic table has a chemical symbol. For example, N is the chemical symbol for nitrogen. (See *Understanding elements and compounds*.)

It's elementary

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Carbon, hydrogen, nitrogen, and oxygen account for 96% of the body's total weight. Calcium and phosphorus account for another 2.5%. (See *What's a body made of*? page 38.)

Atomic structure

An *atom* is the smallest unit of matter that can take part in a chemical reaction. Atoms of a single type constitute an element.

Subatomic particles

Each atom has a dense central core called a *nucleus*, plus one or more surrounding energy layers called *electron shells*. Atoms consist of three basic subatomic particles: *protons, neutrons,* and *electrons*.

Weighing in

A proton weighs nearly the same as a neutron, and a proton and a neutron each weigh 1,836 times as much as an electron.

Protons

Protons (p+) are closely packed particles in the atom's nucleus that have a positive charge. Each element has a distinct number of protons.

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<u> Now I ge</u>t it!

Understanding elements and compounds

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It can be confusing to understand the difference between elements and compounds. The best way to remember the difference is to understand how atoms combine to form each.

Get at them atoms

A single atom constitutes an element. Thus, an atom of hydrogen is the element hydrogen. Now, here's the confusing part: An element can also be composed of more than one atom—a molecule.



Yep. I'm an atom—the smallest unit of matter that can take part in a chemical reaction.

We're atoms that are joined together to make a molecule. We're the same, so we're a molecule of an element.

Molecules: Two (or more) of the same A molecule is a combination of two or more atoms. If these atoms are the same—that is, all the same element

(such as all hydrogen atoms)—they're considered a *molecule* of that element (a molecule of hydrogen).



l know we just met, but l think we'll make a beautiful compound together. ()

Compounds: Where the different come together

If the atoms combined are different that is, different elements (such as a carbon atom and an oxygen atom) the molecule formed is a *compound* (such as the compound CO, or carbon monoxide).



Positive thinking

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An element's number of protons determines its *atomic number* and positive charge. For example, all carbon atoms—and *only* carbon atoms—have six protons; therefore, the atomic number of carbon is 6 (6p+).

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What's a body made of?

This chart shows the chemical elements of the human body in descending order from most to least plentiful.

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Neutrons

Neutrons (n) are uncharged, or neutral, particles in the atom's nucleus.

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Mass numbers

An atom's *atomic mass number* is distinct from its atomic number. The atomic mass number is the sum of the number of protons and neutrons in the nucleus of an atom. You can also think of the atomic mass number as the sum of the masses of protons and neutrons. For example, helium, with two protons and two neutrons, has an atomic mass number of 4.

Isolating the isotopes

Not all the atoms of an element necessarily have the same number of neutrons. An *isotope* is a form of an atom that has a different number of neutrons and, therefore, a different atomic weight.

A weighty matter

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Understanding isotopes is a key to another important concept, *atomic weight*. An atom's atomic weight is the average of the relative weights (atomic mass numbers) of all the element's isotopes. (Recall that isotopes are different atomic forms of the same element that vary in the number of neutrons they contain.)

Electrons

Electrons (e–) are negatively charged particles that orbit the nucleus in electron shells. They play a key role in chemical bonds and reactions.

Staying neutral

The number of electrons in an atom equals the number of protons in its nucleus. The electrons' negative charges cancel out the protons' positive charges, making atoms electrically neutral.

Shell games

Electrons circle the nucleus in *shells*, or concentric circles. Each electron shell can hold a maximum number of electrons and represents a specific energy level. The innermost shell can accommodate two electrons at most, whereas the outermost shells can hold many more.

An atom with single (unpaired) electrons orbiting in its outermost electron shell can be chemically *active*—that is, able to take part in chemical reactions. An atom with an outer shell that contains only pairs of electrons is chemically inactive, or *stable*.

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The value of valence

An atom's valence (its ability to combine with other atoms) equals the number of unpaired electrons in its outer shell. For example, sodium (Na⁺) has a plus-one valence because its outer shell contains an unpaired electron.

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Chemical bonds

A *chemical bond* is a force of attraction that binds a molecule's atoms together. Formation of a chemical bond usually requires energy. Breakup of a chemical bond usually releases energy.

The name's bond...

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Several types of chemical bonds exist:

- A *hydrogen bond* occurs when two atoms associate with a hydrogen atom. Oxygen and nitrogen, for instance, commonly form hydrogen bonds.
- An *ionic* (electrovalent) *bond* occurs when valence electrons transfer from one atom to another.
- A covalent bond forms when atoms share pairs of valence electrons. (See Picturing ionic and covalent bonds.)

Chemical reactions

A *chemical reaction* involves unpaired electrons in the outer shells of atoms. In this reaction, one of two events occurs:

- 1. Unpaired electrons from the outer shell of one atom transfer to the outer shell of another atom.
- 2. One atom shares its unpaired electrons with another atom.

How will they react?

Energy, particle concentration, speed, and orientation determine whether a chemical reaction will occur. The four basic types of chemical reactions are *synthesis*, *decomposition*, *exchange*, and *reversible reactions*. (See *Comparing chemical reactions*, page 42.) It takes energy to bind atoms together. Breaking the bond releases energy.

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Comparing chemical reactions

When chemical reactions occur, they involve unpaired electrons in the outer shells of atoms. Here are the four basic types of chemical reactions.

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Synthesis reaction (anabolism)

A synthesis reaction combines two or more substances (reactants) to form a new, more complex substance (product). This results in a chemical bond.

$A + B \rightarrow A B$

Decomposition reaction (catabolism)

In a decomposition reaction, a substance decomposes, or breaks down, into two or more simpler substances, leading to the breakdown of a chemical bond.

$A B \rightarrow A + B$

Exchange reaction

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An exchange reaction is a combination of a decomposition and a synthesis reaction. This reaction occurs when two complex substances decompose into simpler substances. The simple substances then join (through synthesis) with different simple substances to form new complex substances.

$A B + C D \rightarrow A + B + C + D \rightarrow A D + B C$

Reversible reaction

In a reversible reaction, the product reverts to its original reactants, and vice versa. Reversible reactions may require special conditions, such as heat or light.

 $A + B \leftrightarrow A B$

Inorganic and organic compounds

Although most biomolecules (molecules produced by living cells) form *organic compounds* (compounds containing carbon), some form *inorganic compounds* (compounds without carbon).

Inorganic compounds

Inorganic compounds are usually small and include water and *electro-lytes*—inorganic acids, bases, and salts.

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The body's reservoir

Water is the body's most abundant substance. It performs a host of vital functions, including:

- easily forming polar covalent bonds (which permits the transport of solvents)
- acting as a lubricant in mucus and other bodily fluids
- entering into chemical reactions, such as nutrient breakdown during digestion
- enabling the body to maintain a relatively constant temperature (by both absorbing and releasing heat slowly).

Recognizing ionizing

Acids, bases, and salts are *electrolytes*—compounds whose molecules consist of positively charged ions, *cations*, and negatively charged ions, *anions*, that *ionize* (separate into ions) in solution:

- *Acids* ionize into hydrogen ions (H⁺) and anions. In other words, acids separate into a positively charged hydrogen ion and a negatively charged anion.
- *Bases,* in contrast, ionize into hydroxide ions and cations. Bases separate into negatively charged hydroxide ions and positively charged cations.
- Salts form when acids react with bases. In water, salts ionize into cations and anions, but not hydrogen or hydroxide ions.

A balancing act

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Body fluids must attain acid-base balance to maintain *homeostasis* (the dynamic equilibrium of the body). A solution's acidity is determined by the number of hydrogen ions it contains. The more hydrogen ions present, the more acidic the solution. Conversely, the more hydroxide ions a solution contains, the more basic, or *alkaline*, it is.

Organic compounds

Most biomolecules form *organic compounds*—compounds that contain carbon or carbon-hydrogen bonds. *Carbohydrates, lipids, proteins,* and *nucleic acids* are all examples of organic compounds.

Carbohydrates

In the body, carbohydrates are sugars, starches, and glycogen.

The energy company

The main functions of carbohydrates are to release energy and store energy. There are three types of carbohydrates:





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- 1. *Monosaccharides*, such as ribose and deoxyribose, are sugars with three to seven carbon atoms.
- 2. *Disaccharides*, such as lactose and maltose, contain two monosaccharides.
- 3. *Polysaccharides*, such as glycogen, are large carbohydrates with many monosaccharides.

Lipids

Lipids are water-insoluble biomolecules. The major lipids are *triglycer-ides*, *phospholipids*, *steroids*, *lipoproteins*, and *eicosanoids*.

To insulate and protect

Triglycerides are the most abundant lipid in both food and the body. These lipids are neutral fats that insulate and protect. They also serve as the body's most concentrated energy source. Triglycerides contain three molecules of a fatty acid chemically joined to one molecule of glycerol.

Bars on the cell

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Phospholipids are the major structural components of cell membranes and consist of one molecule of glycerol, two molecules of a fatty acid, and a phosphate group.

No fat in cholesterol?

Steroids are simple lipids with no fatty acids in their molecules. They fall into four main categories, each of which performs different functions.

- *Bile salts* emulsify fats during digestion and aid absorption of the fat-soluble vitamins (vitamins A, D, E, and K).
- *Hormones* are chemical substances that have a specific effect on other cells.
- *Cholesterol*, a part of animal cell membranes, is needed to form all other steroids.
- *Vitamin D* helps regulate the body's calcium concentration.

Porters and other hardworking lipids

Lipoproteins help transport lipids to various parts of the body. *Eicosanoids* include *prostaglandins*, which, among other functions modify hormone responses, promote the inflammatory response, and open the airways, and *leukotrienes*, which also play a part in allergic and inflammatory responses.



To recall the distinction between the three types of carbohydrates, remember the prefixes:

Mono- means one.

Di- means two; therefore, disaccharides contain two monosaccharides.

Poly- means many, so you can expect that polysaccharides contain many monosaccharides.

Bile salts aid

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Proteins

Proteins are the most abundant organic compound in the body. They're composed of building blocks called *amino acids*. Amino acids are linked together by *peptide bonds*—chemical bonds that join the carboxyl group of one amino acid to the amino group of another.

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Building up the blocks

Many amino acids linked together form a *polypeptide*. One or more polypeptides form a protein. The sequence of amino acids in a protein's polypeptide chain dictates its shape. A protein's shape determines which of its many functions it performs:

- providing structure and protection
- promoting muscle contraction
- transporting various substances
- regulating processes
- serving as an enzyme (the largest group of proteins, which act as catalysts for crucial chemical reactions).

Nucleic acids

Nucleic acids' main function is to store and transmit genetic information. The nucleic acids DNA and RNA are composed of nitrogenous bases, sugars, and phosphate groups. The primary hereditary molecule, DNA, contains two long chains of deoxyribonucleotides, which coil into a double-helix shape.

Holding it together

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Deoxyribose and phosphate units alternate in the "backbone" of the chains. Holding the two chains together are base pairs of adenine-thymine and guanine-cytosine.

RNA and its special function

Unlike DNA, RNA has a single-chain structure. It contains ribose instead of deoxyribose and replaces the base thymine with uracil. RNA transmits genetic information from the cell nucleus to the cytoplasm. In the cytoplasm, it guides protein synthesis from amino acids.





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