CHAPTER 1: INTRODUCTION TO THE HUMAN BODY

At the end of this chapter, student will be able to:

a) Define the terms *anatomy*, *physiology*, and *pathophysiology*. Use an example to explain how they are related.
b) Name the levels of organization of the body from simplest to most complex, and explain each.
c) Define the terms *metabolism*, *metabolic rate*, and *homeostasis*, and use examples to explain.
d) Explain how a negative feedback mechanism works, and how a positive feedback mechanism differs.
e) Describe the anatomic position.
f) State the anatomic terms for the parts of the body.
g) Use proper terminology to describe the location of body parts with respect to one another.
h) Name the body cavities, their membranes, and some organs within each cavity.
i) Describe the possible sections through the body or an organ.
j) Explain how and why the abdomen is divided into smaller areas. Be able to name organs in these areas.
k) Name the organic molecules that make up cell membranes and state their functions.
l) Describe the functions of the cell organelles.
m) Define each of these cellular transport mechanisms and give an example of the role of each in the body: diffusion, osmosis, facilitated diffusion, active transport, filtration, phagocytosis, and pinocytosis.
n) Describe the triplet code of DNA.
o) Explain how the triplet code of DNA is transcribed and translated in the synthesis of proteins.
p) Describe what happens in mitosis and in meiosis.

1.1. INTRODUCTION TO HUMAN ANATOMY & PHYSIOLOGY

Knowledge of the structure and the function of the human body are essential for those planning a career in the health sciences. It is the basis for understanding disease. In this unit, anatomy and physiology are defined, body's structural and functional organization is explained and an overview of the characteristics of life and homeostasis is provided. Finally, terminology and body plan are presented.

Anatomy is the scientific discipline that investigates the body's structure. It describes the shape and size; it examines the relationship between the structures of the body parts and its function. The structure of specific body part allows it to perform a particular function. Understanding the relationship between structure and function makes it easier to understand and appreciate Anatomy.

Developmental anatomy is the study of the structural changes that occur between conception and adulthood.

Embryology is a subspecialty of developmental anatomy that considers changes from conception to the end of 8^{th} week of development.

Cytology examines the structural features of cells and **histology** examines tissues, which are cells and the materials surrounding them.

Gross anatomy is the study of structures that can be examined without the aid of a microscope. It can be approached from either a systemic or a regional perspective. In **systemic anatomy**, the body is studied system by system, which the approach taken in most introductory textbooks. In **regionally anatomy**, the body is studied area by area. Within each region, such as the head, abdomen, or arm, all systems are studied simultaneously.

Surface anatomy is the study of the external form of the body and its relation to deeper structures.

Physiology is the scientific investigation of the processes or functions of living things. The major goals of physiology are to understand and predict the body's responses to stimuli and to understand how the body maintains conditions within a narrow range of values in a constantly changing environment.

The study of the human body must encompass both anatomy and physiology because structures, functions, and processes are interlinked.

1.2. HUMAN BODY ORGANIZATION

Human beings are arguably (questionably, perhaps) the most complex organisms on this planet. Imagine billions of microscopic parts, each with its own identity, working together in an organized manner for the benefit of the total being. The human body is a single structure but it is made up of billions of smaller structures organized at six levels:

- **Chemical level:** involves interactions between **atoms**, which are tiny building blocks of matter.

- **Cell level**: Cells are the basic structural and functional units of organisms. Atoms combine to form molecules. Molecules can combine to form organelles, such as the nucleus and mitochondria which make up cells. Cell has long been recognized as the simplest units of living matter that can maintain life and reproduce themselves.

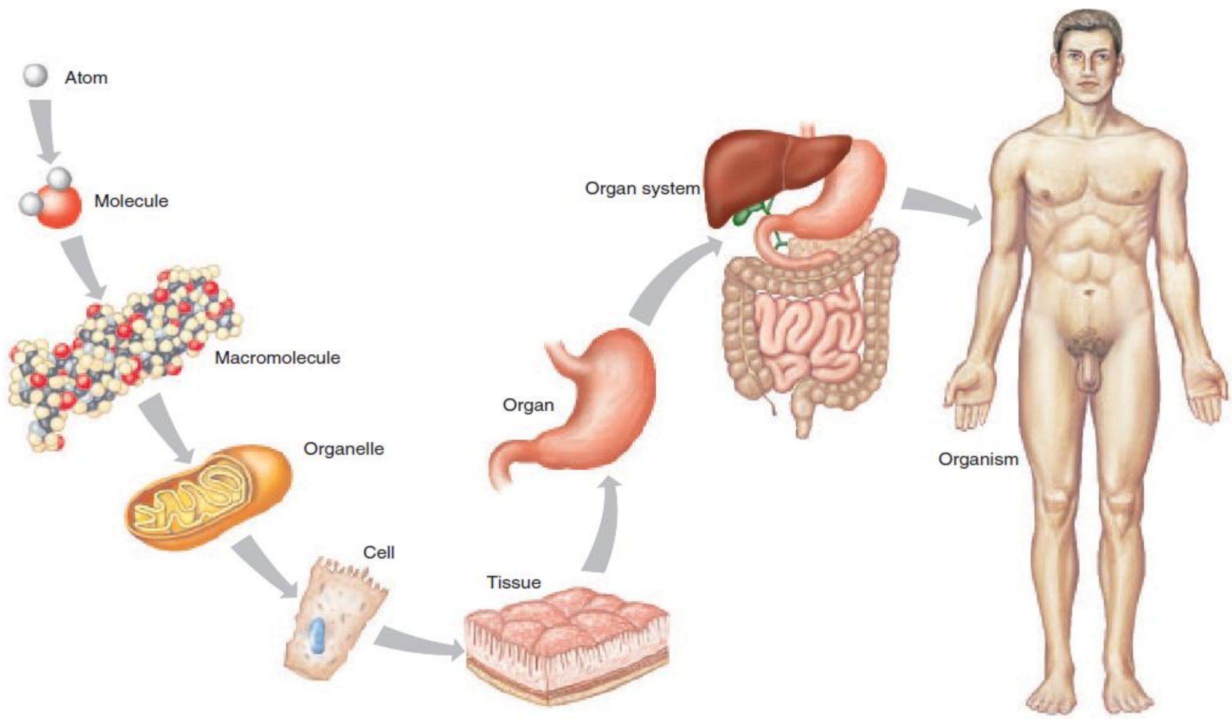
- **Tissue level:** Tissues are somewhat more complex units than cells. By definition, a tissue is an organization of a great many similar cells with varying amounts and kinds of nonliving, intercellular substance between them. The numerous different tissues that make up the body are classified into four basic types: epithelial, connective, muscle and nervous.

- **Organ level:** Organs are more complex units than tissues. An organ is an organization of several different kinds of tissues (at least two types of tissues) so arranged that together they can perform a special function. For example, the stomach is an organization of muscle, connective, epithelial, and nervous tissues. Muscle and connective tissues form its wall, epithelial and connective tissues form its lining, and nervous tissue extends throughout both its wall and its lining.

- **Organ system level:** An organ system is a group of organs that have a common function or set of functions and are therefore viewed as a unit. e.g: urinary system (Kidneys, ureter, Urinary bladder and urethra). Eleven major organ systems compose the human body:
  - ✓ Skeletal
  - ✓ Muscular
  - ✓ Nervous
  - ✓ Endocrine

- ✓ Cardiovascular
- ✓ Lymphatic
- ✓ Respiratory
- ✓ Digestive
- ✓ Urinary
- ✓ Reproductive and
- ✓ Integumentary system (skin and accessories)

- **Organism level:** An organism is any living thing considered as a whole whether composed of one cell, such as a bacterium, or of trillions of cells, such as human. The human organism is a complex organ systems, all mutually dependent on one another.



1.3. BODY FUNCTIONS AND LIFE PROCESS

1.3.1. Body Functions

Body functions are the physiological or psychological functions of body systems. The body's functions are ultimately its cells' functions. Survival is the body's most important business. Survival depends on the body's maintaining or restoring homeostasis, a state of relative constancy, of its internal environment.

More than a century ago, French physiologist, Claude Bernard (1813-1878), made a remarkable observation. He noted that "*body cells survived in a healthy condition only when the temperature, pressure, and chemical composition of their environment remained relatively constant* ".

Homeostasis is the existence and maintenance of a relatively constant environment within the body. For cells to function normally, the volume, temperature and chemical content conditions known as variables (because their values can change) must remain within a narrow range. Homeostasis mechanisms, such as sweating or shivering, normally maintain body temperature near an ideal normal value or set point.

NB: Homeostasis mechanisms are not able to maintain body temperature precisely at the set point. Instead, body temperature increase and decrease slightly around the set point to produce a normal range of values. And as long as body temperature remains within this normal range, homeostasis is maintained. A homeostatic mechanism has three components: a sensor, a regulatory center, and an effector. The sensor detects a change in the internal environment; the regulatory center activates the effector; the effector reverses the change and brings conditions back to normal again. Now, the sensor is no longer activated.

The organ systems help control the body's internal environment so that it remains relatively constant. Most systems of the body are regulated by negative-feedback mechanisms, which maintain homeostasis.

Negative feedback mechanisms keep body conditions within a set normal range by *reversing* any upward or downward shift. A **positive feedback system** tends to *strengthen* or *reinforce* a change in one of the body's controlled conditions.

The body's ability to perform many of its functions changes gradually over the years. In general, the body performs its functions least well at both ends of life - in infancy and in old age.

During childhood, body functions gradually become more and more efficient and effective. During late maturity and old age the opposite is true. They gradually become less and less efficient and effective. During young adulthood, they normally operate with maximum efficiency and effectiveness.

1.3.2. Life Process

All living organisms have certain characteristics that distinguish them from non-living forms. The basic processes of life include organization, metabolism, responsiveness, movements, and reproduction.

In humans, who represent the most complex form of life, there are additional requirements such as growth, differentiation, respiration, digestion, and excretion. All of these processes are interrelated. No part of the body, from the smallest cell to a complete body system, works in isolation.

All function together, in fine-tuned balance, for the well being of the individual and to maintain life. Disease such as cancer and death represent a disruption of the balance in these processes.

The following is a brief description of the life process:

1. Organization :

At all levels of the organizational scheme; there is a division of labour. Each component has its own job to perform in cooperation with others. Even a single cell, if it loses its integrity or organization, will die.

2. Metabolism

Metabolism is a broad term that includes all the chemical reactions that occur in the body. One phase of metabolism is catabolism in which complex substances are broken down into simpler building blocks and energy is released. Anabolism is said when two or more atoms, ions, or molecules combine to form new and larger molecules.

3. Responsiveness

Responsiveness or irritability is concerned with detecting changes in the internal or external environments and reacting to that change. It is the act of sensing a stimulus and responding to it.

4. Movement

There are many types of movement within the body. On the cellular level, molecules move from one place to another. Blood moves from one part of the body to another. The diaphragm moves with every breath. The ability of muscle fibers to shorten and thus to produce movement is called contractility.

5. Reproduction

For most people, reproduction refers to the formation of a new person, the birth of a baby. In this way, life is transmitted from one generation to the next through reproduction of the organism. In a broader sense, reproduction also refers to the formation of new cells for the replacement and repair of old cells as well as for growth. This is cellular reproduction. Both are essential to the survival of the human race.

6. *Growth*

Growth refers to an increase in size either through an increase in the number of cells or through an increase in the size of each individual cell. In order for growth to occur, anabolic processes must occur at a faster rate than catabolic processes.

7. Differentiation

Differentiation is a developmental process by which unspecialized cells change into specialized cells with distinctive structural and functional characteristics. Through differentiation, cells develop into tissues and organs.

8. Respiration

Respiration refers to all the processes involved in the exchange of oxygen and carbon dioxide between the cells and the external environment. It includes ventilation, the diffusion of oxygen and carbon dioxide, and the transport of the gases in the blood. Cellular respiration deals with the cell's utilization of oxygen and release of carbon dioxide in its metabolism.

9. Digestion

Digestion is the process of breaking down complex ingested foods into simple molecules that can be absorbed into the blood and utilized by the body.

10. Excretion

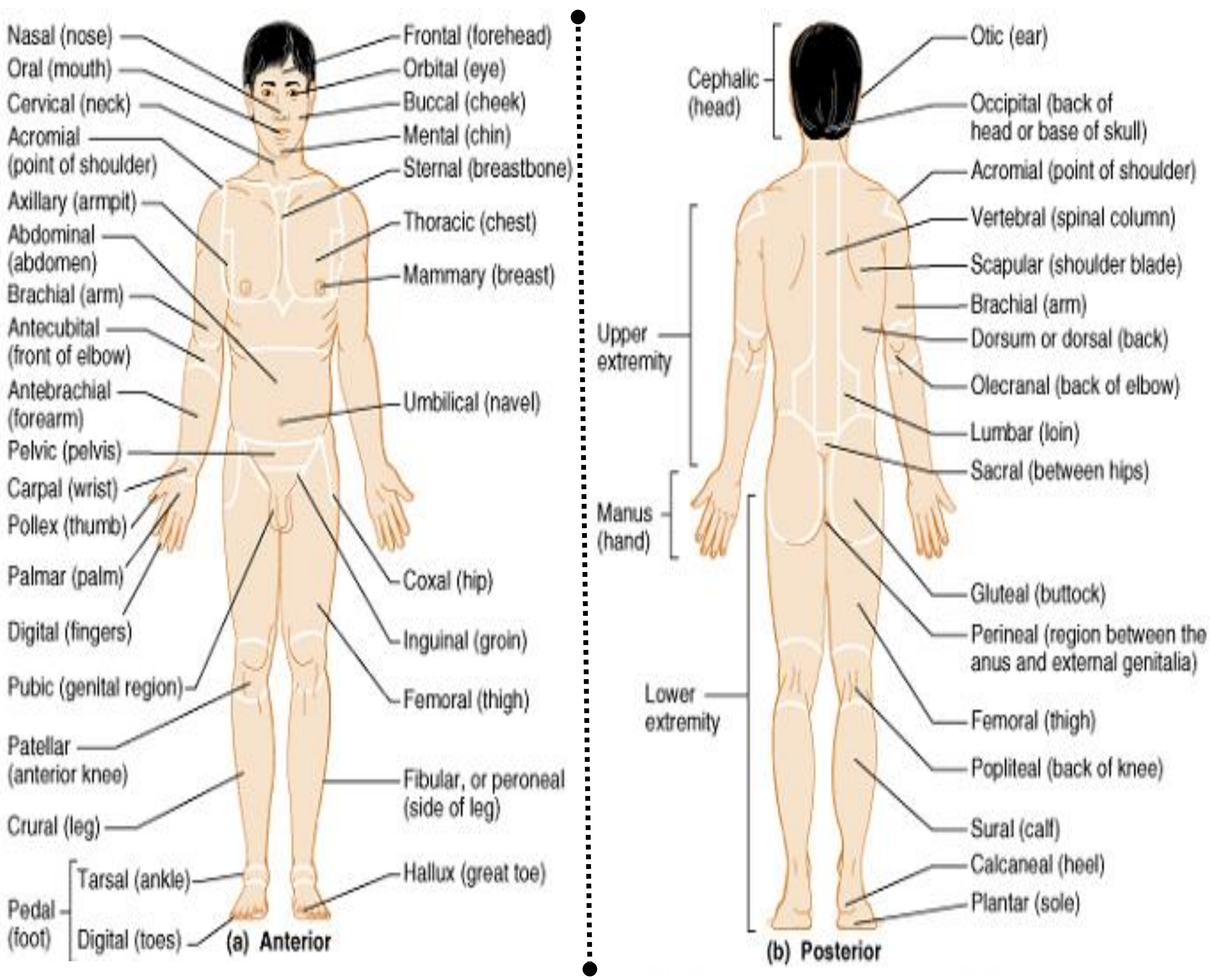
Excretion is the process that removes the waste products of digestion and metabolism from the body. It gets rid of by-products that the body is unable to use, many of which are toxic and incompatible with life.

The ten life processes described above are not enough to ensure the survival of the individual. In addition to these processes, life depends on certain physical factors from the environment. These include water, oxygen, nutrients, heat, and pressure.

1.4. ANATOMICAL TERMINOLOGY

Before we get into the following learning units, which will provide more detailed discussion of topics on different human body systems, it is necessary to learn some useful terms for describing body structure. Knowing these terms will make it much easier for us to understand the content of the next learning units. Three groups of terms are introduced here: directional terms, terms describing planes of the body, and terms describing body cavities.

1.4.1. Descriptive terms for body parts and areas

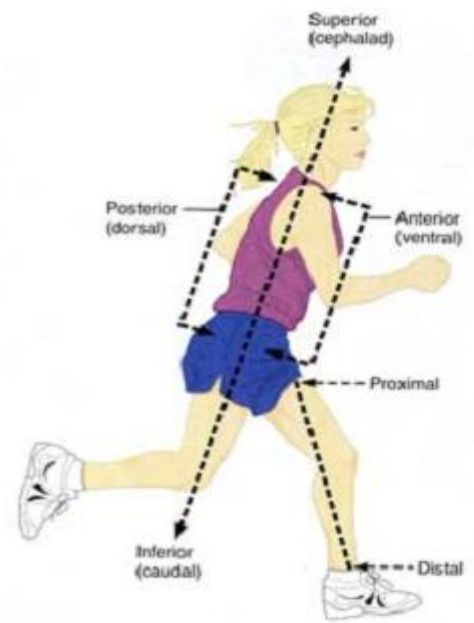


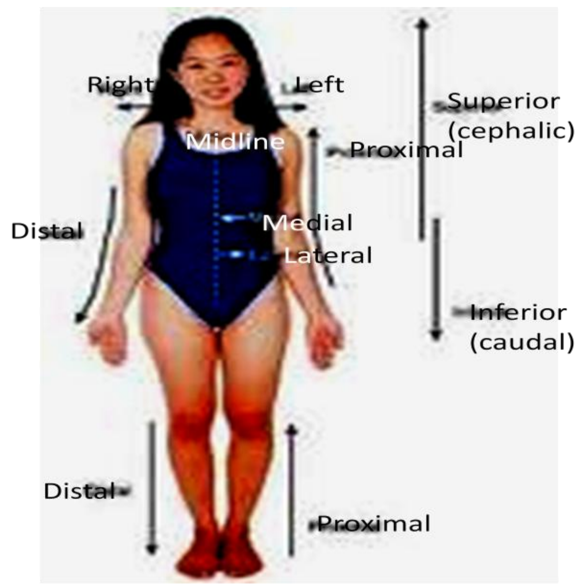
1.4.2. Body positions:

Anatomical position refers to a person standing erect with the face directed forward, the upper limbs hanging to the sides, and the palms of hands facing forward. A person is **supine** when lying face upward and **prone** when lying face downward.

1.4.3. Axes and Directional Terms

An organism has three axes: the *cephalocaudal axis*, the *anteroposterior axis*, and the *rightleft axis*. Directional terms describe the positions of structures relative to other structures or locations in the body.





- ✓ ***Superior or cranial*****:** toward the head end of the body; upper (example, the hand is part of the superior extremity).
- ✓ ***Inferior or caudal*****:** away from the head; lower (example, the foot is part of the inferior extremity).
- ✓ ***Anterior or ventral*****:** front (example, the kneecap is located on the anterior side of the leg).
- ✓ ***Posterior or dorsal*****:** back (example, the shoulder blades are located on the posterior side of the body).
- ✓ ***Medial*****:** toward the midline of the body (example, the middle toe is located at the medial side of the foot).

- ✓ ***Lateral:*** away from the midline of the body (example, the little toe is located at the lateral side of the foot).
- ✓ ***Proximal***: toward or nearest the trunk or the point of origin of a part (example, the proximal end of the femur joins with the pelvic bone).
- ✓ ***Distal*****:** away from or farthest from the trunk or the point or origin of a part (e.g., the hand is located at the distal end of the forearm).
- ✓ *Superficial*: Toward or on the surface (e.g., the skin is superficial to muscle.)
- ✓ *Deep*: Away from the surface, internal (e.g., the lungs are deep to the ribs.)

Note: The directional terms **superior**, **inferior**, **anterior** and **posterior** are useful for humans only, since these surfaces are different in quadrupeds. The remaining terms listed above are preferable because they can be used universally.

1.4.4. Planes of the Body

Medical professionals often refer to sections of the body in terms of anatomical planes (flat surfaces). These planes are imaginary lines - vertical or horizontal - drawn through an upright body. The terms are used to describe a specific body part.

A **sagittal** or **lateral plane** is a vertical plane that divides the body or an organ into right and left sides. More specifically, when such a plane passes through the midline of the body or an organ and divides it into *equal* right and left sides, it is called a **midsagittal plane** or a **median plane.** The *midline* is an imaginary vertical line that divides the body into equal left and right sides. If the sagittal plane does not pass through the midline but instead divides the body or an organ into *unequal* right and left sides, it is called a **parasagittal plane**.

A **frontal** or **coronal plane** divides the body or an organ into anterior (front) and posterior (back) portions. A **transverse plane** divides the body or an organ into superior (upper) and inferior (lower) portions.

Other names for a transverse plane are a **cross-sectional, axial** or **horizontal plane.** Sagittal, frontal, and transverse planes are all at right angles to one another. An **oblique plane,** by contrast, passes through the body or an organ at an angle between a transverse plane and a sagittal plane or between a transverse plane and a frontal plane.

When you study a body region, you often view it in section. A **section** is a cut of the body or one of its organs made along one of the planes just described. It is important to know the plane of the section so you can understand the anatomical relationship of one part to another.

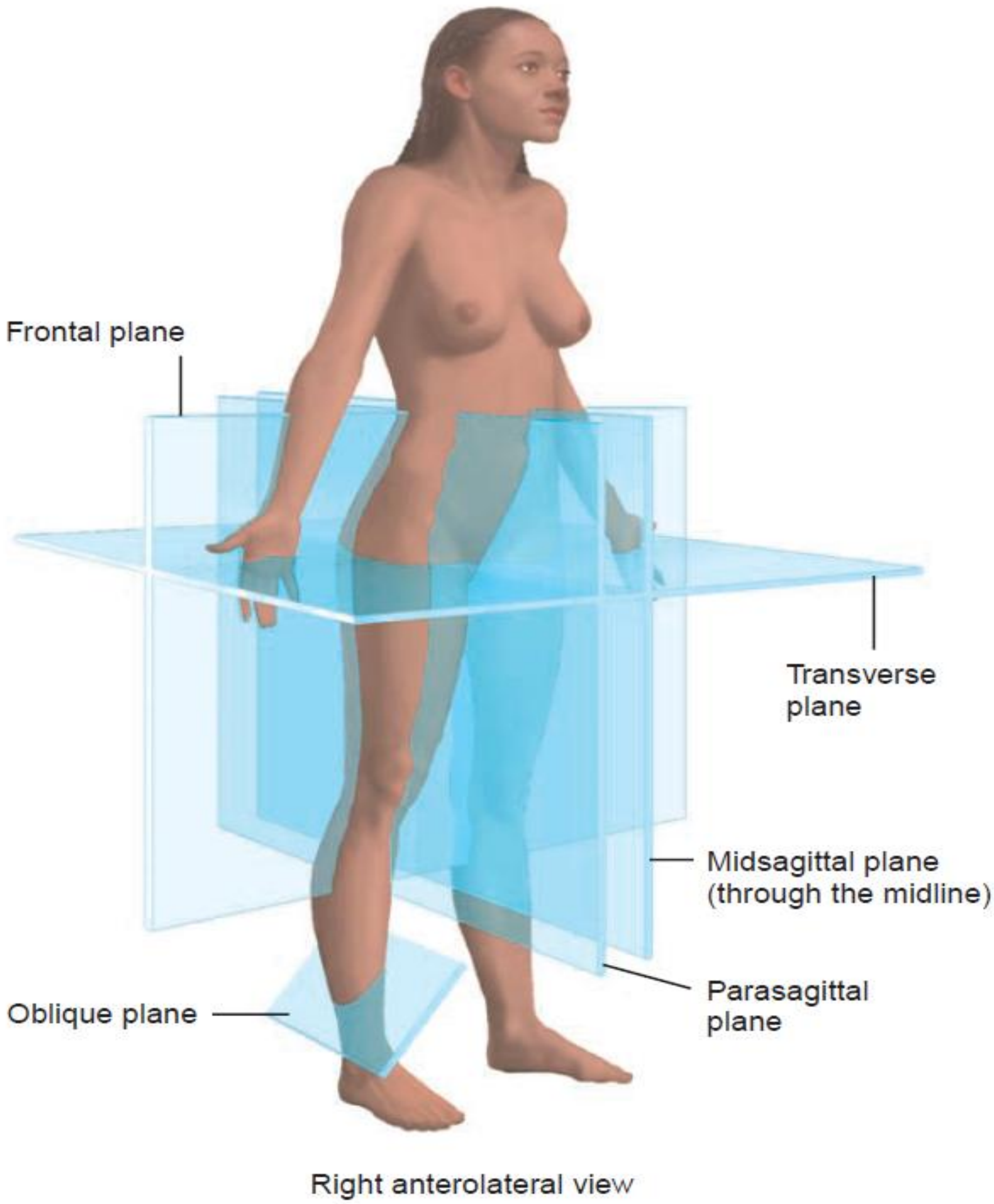


Figure: Frontal, transverse, sagittal, and oblique planes divide the body in specific ways.

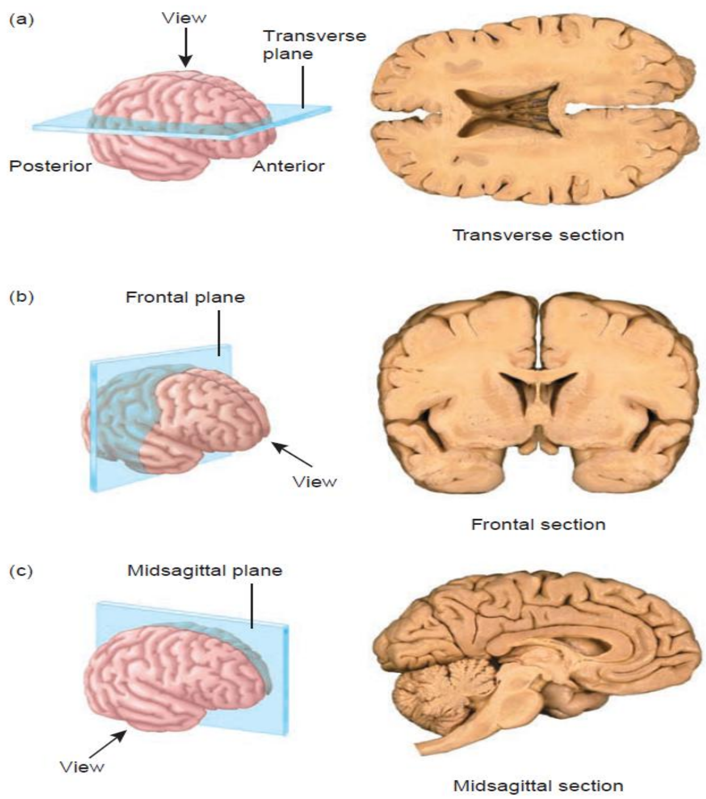
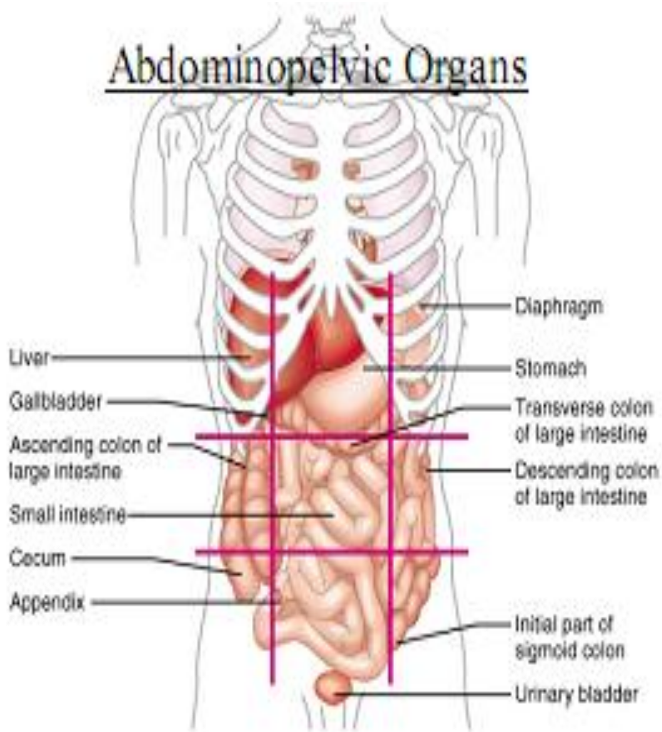
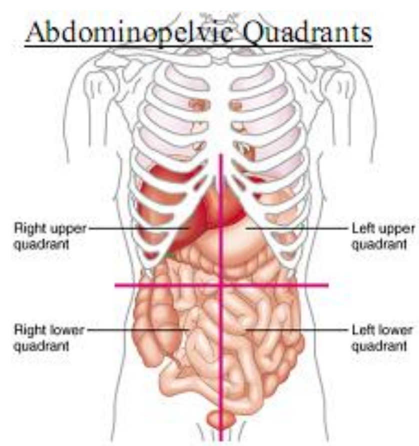
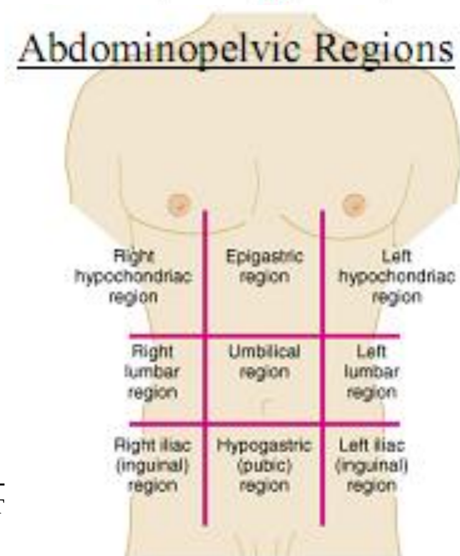


Figure: Planes divide the body in various ways to produce sections.

1.4.5 Abdominopelvic organs, regions and quadrants

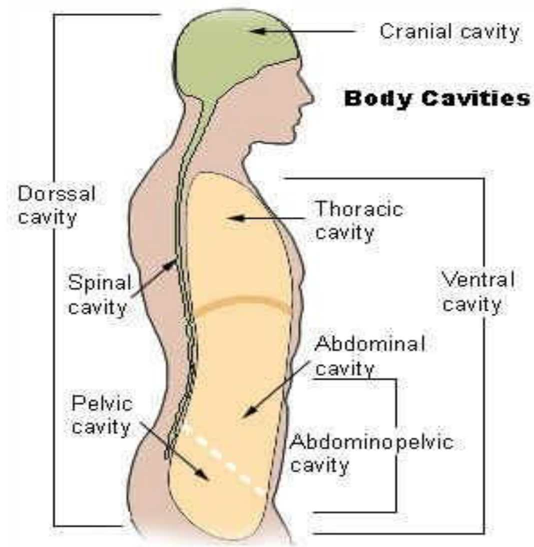






1.4.6. Body Cavities

The cavities, or spaces, of the body contain the internal organs, or viscera. The two main cavities are called the ventral and dorsal cavities. The ventral is the larger cavity and is subdivided into two parts (*thoracic* and *abdominopelvic* cavities) by the diaphragm, a dome-shaped respiratory muscle.

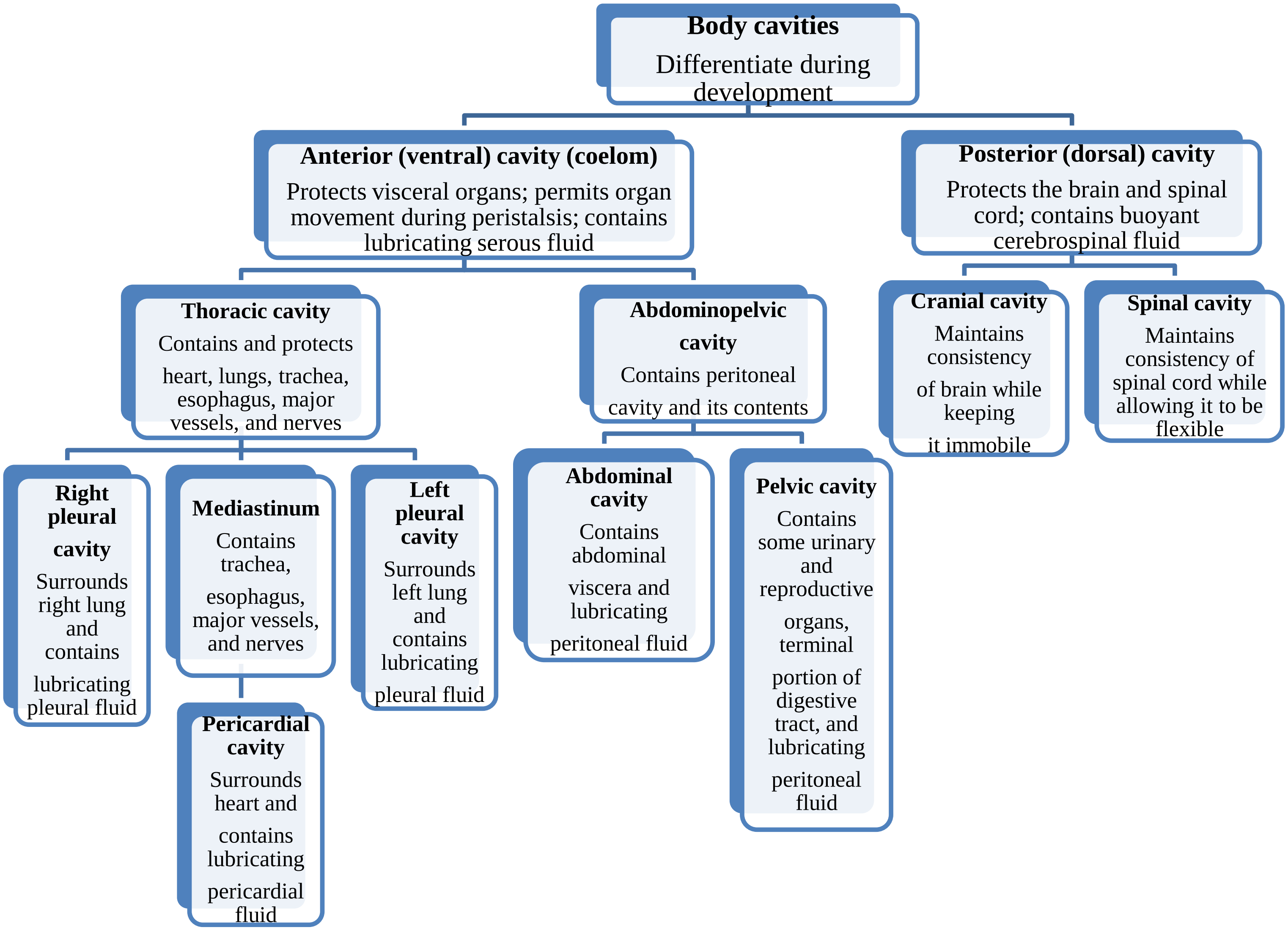


Thoracic cavity: The upper ventral, thoracic, or chest cavity contains the heart, lungs, trachea, esophagus, large blood vessels, and nerves. The thoracic cavity is bound laterally by the ribs (covered by costal pleura) and the diaphragm caudally (covered by diaphragmatic pleura).

- **Abdominal and pelvic cavity:** The lower part of the ventral (abdominopelvic) cavity can be further divided into two portions: abdominal portion and pelvic portion. The abdominal cavity contains most of the gastrointestinal tract as well as the kidneys and adrenal glands. The abdominal cavity is bound cranially by the diaphragm, laterally by the body wall, and caudally by the pelvic cavity.

  The pelvic cavity contains most of the urogenital system as well as the rectum. The pelvic cavity is bounded cranially by the abdominal cavity, dorsally by the sacrum, and laterally by the pelvis.
- **Dorsal cavity**: The smaller of the two main cavities is called the dorsal cavity. As its name implies, it contains organs lying more posterior in the body. The dorsal cavity, again, can be divided into two portions. The upper portion, or the cranial cavity, houses the brain, and the lower portion, or vertebral canal houses the spinal cord.

Organization of body cavities:



1.5. CELL STRUCTURE AND FUNCTIONS

Cells, the smallest structures capable of maintaining life and reproducing, compose all living things, from single-celled plants to multibillion-celled animals. The human body, which is made up of numerous cells, begins as a single, newly fertilized cell. Almost all human cells are microscopic in size. To give you an idea how small a cell is, one average-sized adult body, according to one estimate, consists of 100 trillion (100,000,000,000,000) cells!

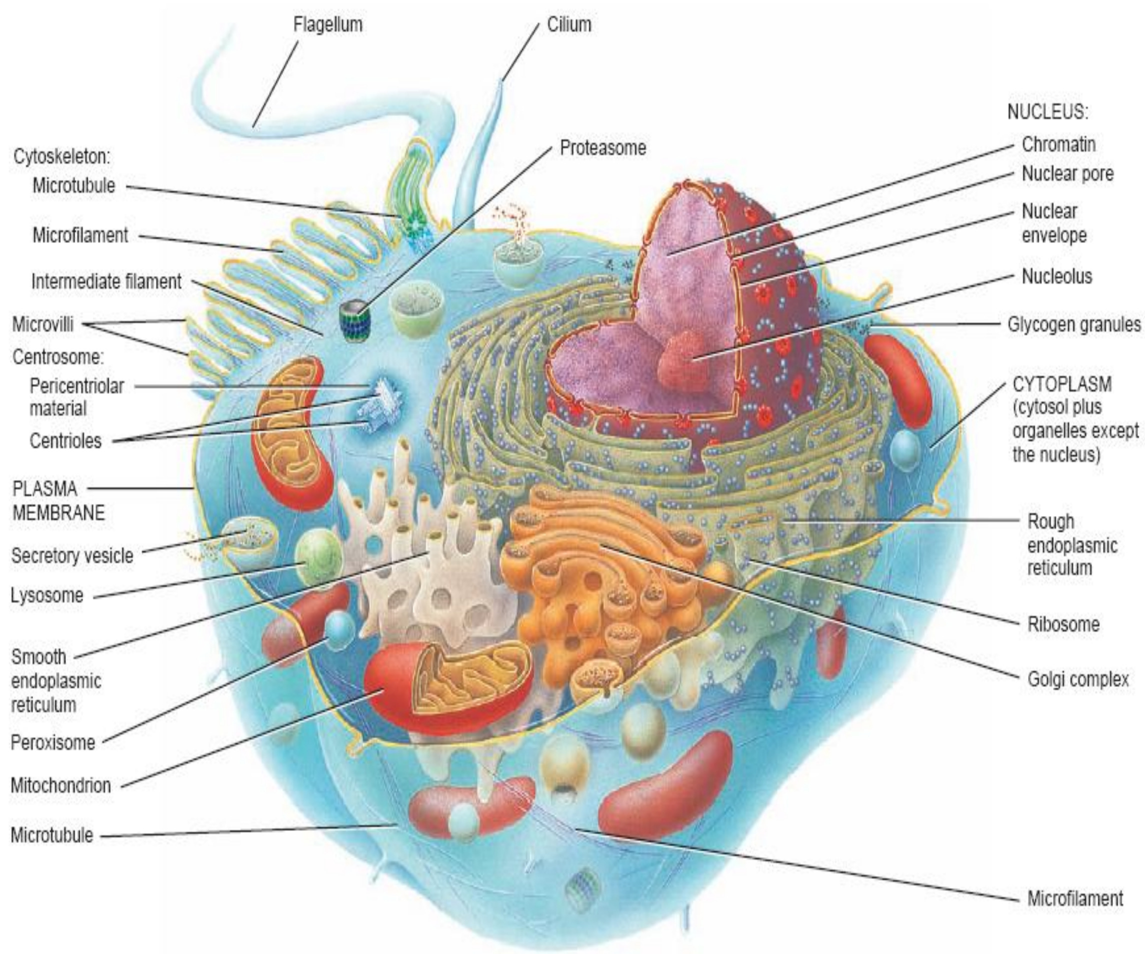
Microorganisms, such as amoebas and bacteria, are single cells that function independently. Human cells, however, must work together and function interdependently. Human cells vary in size, shape, and function. Most human cells are so small they can only be seen with the aid of a microscope and are measured in units called **micrometers**. One exception is the human ovum or egg cell, which is about 1 millimeter in diameter, just visible to the unaided eye.

1.5.1. Cell Structure

Ideas about cell structure have changed considerably over the years. Early biologists saw cells as simple membranous sacs containing fluid and a few floating particles. Despite their many differences, human cells have several similar structural features: a cell membrane, a nucleus, and cytoplasm and cell organelles.

Red blood cells are an exception because they have no nuclei when mature. The cell membrane forms the outer boundary of the cell and surrounds the cytoplasm, organelles, and nucleus.

There are many different types, sizes, and shapes of cells in the body. For descriptive purposes, the concept of a "generalized cell" is introduced. It includes features from all cell types.



Sectional view

Cell parts and Functions

Cell parts	Structure	Function
Plasma Membrane	Lipid bilayer composed of phospholipids and cholesterol with proteins that extend across or are embedded in either surface of the lipid bilayer.	- Outer boundary of cells that controls entry and exit of substances; - Receptor molecules function in intercellular communication; - Marker molecules enable cells to recognize one another.
Cytoplasm : Cytosol		
Fluid part	Water with dissolved ions and molecules; colloid with suspended proteins.	- Contains enzymes that catalyze decomposition and synthesis reactions; - ATP is produced in glycolysis reactions.
Cytoskeleton Microtubules	Hollow cylinders composed of the protein tubulin; 2,5nm in diameter.	- Support the cytoplasm and form centrioles, spindle fibers, cilia, and flagella; - Responsible for movement of structures

		in the cell.
Actin filaments	Small fibrils of the protein actin; 8 nm in diameter.	- Provide structural support to cells, - Support microvilli, - Responsible for cell movements.
Intermediate filaments	Protein fibers; 10 nm in diameter.	-Provider structural support to cells.
Cytoplasmic inclusions	Aggregates of molecules manufactured or ingested by the cell; may be membrane-bound.	- Function depends on the molecules: energy storage (lipid, glycogen), oxygen transport (haemoglobin), skin color (melanin), and others.
Organelles		
Nucleus - **Nuclear envelope**	Double membrane enclosing the nucleus; the outer membrane is continuous with the endoplasmic reticulum; nuclear pores extend through the nuclear envelope.	-Separates nucleus from cytoplasm and regulates movement of materials into and out of the nucleus.
Chromatin	Dispersed, thin strands of DNA, histones, and other proteins; condenses to form chromosomes during cell division	-DNA regulates protein (e.g., enzyme) synthesis and therefore the chemical reactions of the cell; -DNA is the genetic, or hereditary, material.
Nucleolus	One or more dense bodies consisting of ribosomal RNA and proteins.	-Assembly site of large and small ribosomal subunits.
Cytoplasmic organelles		
Ribosome	Ribosomal RNA and proteins form large and small subunits; attached to endoplasmic reticulum or free ribosomes are distributed throughout the cytoplasm.	-Site of protein synthesis.
Rough endoplasmic reticulum	Membranous tubules and flattened sacs with no attached ribosomes	- Protein synthesis and transport to Golgi apparatus.
Smooth endoplasmic reticulum	Membranous tubules and flattened sacs with no attached ribosomes.	- Manufactures lipids and carbohydrates; - Detoxifies harmful chemicals; - Stores calcium.
Golgi apparatus	Flattened membrane sacs stacked on each other.	- Modifies, packages, and distributes proteins and lipids for secretion or

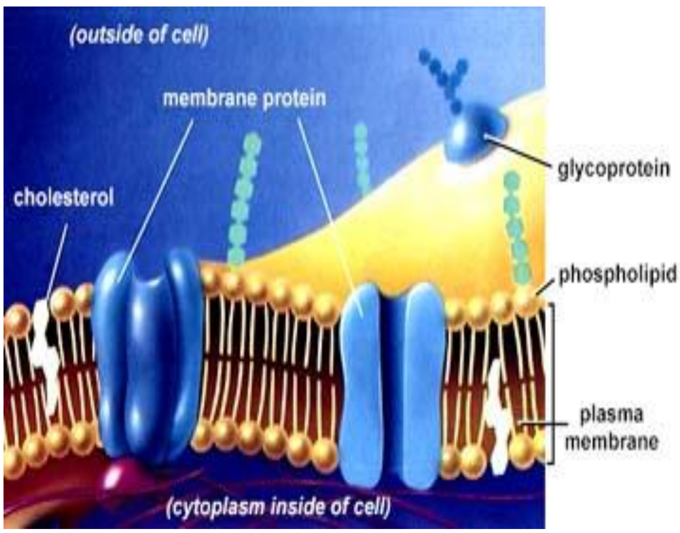
		internal use.
Secretory vesicle	Membrane-bound sac pinched off Golgi apparatus	- Carries proteins and lipids to cell surface for secretion.
Lysosome	Membrane-bound vesicle pinched off Golgi apparatus	- Contain enzymes to digest ingested material or damaged tissue
Peroxisome	Membrane-bound vesicle.	- One site of lipid and amino acid degradation; breaks down hydrogen peroxide.
Proteasomes	Tubelike protein complexes in the cytoplasm	- Site of destruction of old or damaged proteins
Mitochondria	Spherical, rod-shaped, or threadlike structures; enclosed by double membrane; inner membrane forms projections called cristae.	- Major sites of ATP synthesis when oxygen is available.
Centrioles	Pair of cylindrical organelles in the centrosome, consisting of triplets of parallel microtubules.	- Centers for microtubule formation; - Determine cell polarity during cell division; - Form the basal bodies of cilia and flagella.
Spindle Fibers	Microtubules extending from the centrosome to chromosomes and other parts of the cell (e.g., aster fibers)	- Assist in the separation of chromosomes during cell division.
Cilia	Extensions of the plasma membrane containing doublets of parallel microtubules; 10 um in length.	- Move materials over the surface of cells.
Flagellum	Extensions of the plasma membrane containing doublets of parallel microtubules, 55um in length.	- Enables a cell to move
Microvilli	Extension of the plasma membrane containing microfilaments.	- Increase surface area of the plasma membrane for absorption and secretion; modified to form sensory receptors.

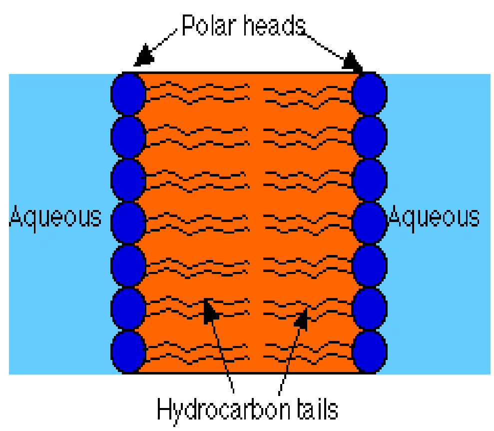
1.5.2. Cell Function

The structural and functional characteristics of different types of cells are determined by the nature of the proteins present. Cells of various types have different functions because cell structure and function are closely related. It is apparent that a cell that is very thin is not well suited for a **protective function**. Bone cells do not have an appropriate structure for nerve impulse conduction. Just as there are many cell types, there are varied cell functions. The generalized cell functions include *movement of substances across the cell membrane*, *cell division to make new cells*, and *protein synthesis*.

1.5.2.1. Movement of substances across the cell membrane

The cell membrane consists of 2 primary building blocks include protein (about 60% of the membrane) and lipid, or fat (about 40% of the membrane).





The primary lipid is called **<u>phospholipid</u>** , and molecules of phospholipid form a **'phospholipid bilayer'** (two layers of phospholipid molecules).

This bilayer forms because the two 'ends' of phospholipid molecules have very different characteristics: one end is polar (or hydrophilic) and one (the hydrocarbon tails below) is non-polar (or hydrophobic). The survival of the cell depends on maintaining the difference between extracellular and intracellular material.

Transport of materials into and out of cells:

TRANSPORT PROCESS	DESCRIPTION	SUBSTANCES TRANSPORTED
Passive Processes	Movement of substances down a concentration gradient until equilibrium is reached; do not require cellular energy in the form.	
Diffusion	The movements of molecules or ions down a concentration gradient due to their kinetic energy until they reach equilibrium.	
Simple diffusion	Passive movement of a substance down its concentration gradient, through the lipid bilayer of the plasma membrane without the help of membrane transport proteins.	Nonpolar, hydrophobic solutes: oxygen, carbon dioxide, and nitrogen gases; fatty acids; steroids; and fat-soluble vitamins.
Facilitated diffusion	Passive movement of a substance down its concentration gradient through the lipid bilayer by transmembrane proteins that function as channels or carriers	Polar or charged solutes: glucose, fructose, galactose, some vitamins, and ions such as K^+, Cl^-, Na^+ and Ca^{2+}
Osmosis	Passive movement of water molecules across a selectively permeable membrane from an area of higher water concentration to an area of lower water concentration	Solvent: water in living systems.
Active Processes	Movement of substances against a concentration gradient; requires cellular energy in the form of ATP.	
Active Transport	Active process in which a cell expends energy to move a substance across the membrane against its concentration gradient by transmembrane proteins that function as carriers.	Polar or charged solutes.
Primary active transport	Active process in which a substance moves across the membrane against its concentration gradient by pumps (carriers) that use energy supplied by hydrolysis of ATP.	Na^+, K^+, Ca^{2+}, H^+, I^-, Cl^-, and other ions.
Secondary active transport	Coupled active transport of two substances across the membrane using energy supplied by a Na^+ or H^+ concentration gradient	Antiport: Ca^{2+} H^+ out of cells. Symport: glucose, amino acids into cells.

	maintained by primary active transport pumps. Antiporters move Na^+ (or H^+) and another substance in opposite directions across the membrane; symporters move Na^+ (or H^+) and another substancse in the same direction across the membrane.	
Transport in Vesicles	Active process in which substances move into or out of cells in vesicles that bud from the plasma membrane; requires energy supplied by ATP.	
Endocytosis	Movement of substances into a cell in vesicles.	
Receptor-mediated endocytosis	Ligand–receptor complexes trigger infolding of a clathrin-coated endocytosis pit that forms a vesicle containing ligands.	Ligands: transferrin, low-density lipopro teins (LDLs), some vitamins, certain hormones, and antibodies.
Phagocytosis	"Cell eating"; movement of a solid particle into a cell after pseudopods engulf it to form a phagosome.	Bacteria, viruses, and aged or dead cells.
Bulk-phase endocytosis	"Cell drinking"; movement of extracellular fluid into a cell by infolding of plasma membrane to form a vesicle.	Solutes in extracellular fluid.
Exocytosis	Movement of substances out of a cell in secretory vesicles that fuse with the plasma membrane and release their contents into the extracellular fluid.	Neurotransmitters, hormones, and digestive enzymes
Transcytosis	Movement of a substance through a cell as a result of endocytosis on one side and exocytosis on the opposite side.	Substances, such as antibodies, across endothelial cells. This is a common route for substances to pass between blood plasma and interstitial fluid.

1.5.3 The genetic code and protein synthesis

DNA is a double strand of nucleotides in the form of a **double helix**, very much like a spiral ladder. The rungs of the ladder are made of the four nitrogenous bases, always found in complementary pairs: adenine with thymine (A–T) and guanine with cytosine (G–C).

Although DNA contains just these four bases, the bases may be arranged in many different sequences (reading up or down the ladder). It is the sequence of bases, the sequence of A, T, C, and G, that is the **genetic code**. The DNA of our 46 chromosomes may also be called our **genome**, which is the term for the total genetic information in a particular species.

The human genome is believed to contain about 3 billion base pairs, and the number of our genes is now estimated to be between 20,000 and 25,000. The code for a single amino acid consists of three bases in the DNA molecule; this **triplet** of bases may be called a **codon**. There is a triplet of bases in the DNA for each amino acid in the protein. If a protein consists of 100 amino acids, the gene for that protein would consist of 100 triplets, or 300 bases.

Some of the triplets will be the same, since the same amino acid may be present in several places within the protein. Also part of the gene are other triplets that start and stop the process of making the protein, rather like capital letters or punctuation marks start and stop sentences.

- **RNA and protein synthesis**

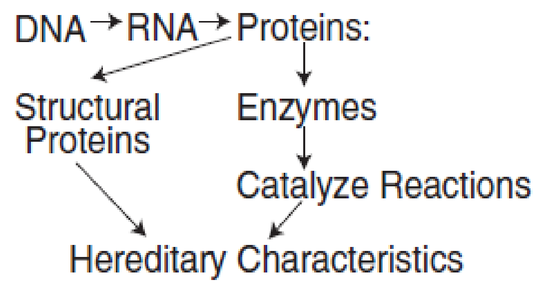
RNA, the other nucleic acid, has become a surprising molecule, in that it has been found to have quite a few functions. It may be involved in the repair of DNA, and it is certainly involved in **gene expression**. The expression of a gene means that the product of the gene is somehow apparent to us, in a way we can see or measure, or is not apparent when it should be.

Examples would be having brown eyes or blue eyes, or having or not having the intestinal enzyme lactase to digest milk sugar.

The transcription and translation of the genetic code in DNA into proteins require RNA. DNA is found in the chromosomes in the nucleus of the cell, but protein synthesis takes place on the ribosomes in the cytoplasm. **Messenger RNA** (**mRNA**) is the intermediary molecule between these two sites.

When a protein is to be made, the segment of DNA that is its gene uncoils, and the hydrogen bonds between the base pairs break. Within the nucleus are RNA nucleotides (A, C, G, U) and enzymes to construct a single strand of nucleotides that is a complementary copy of half the DNA gene (with uracil in place of thymine). This process is **transcription**, or copying, and the copy of the gene is mRNA, which now has the codons for the amino acids of the protein, and then separates from the DNA.

The gene coils back into the double helix, and the mRNA leaves the nucleus, enters the cytoplasm, and becomes attached to ribosomes. Thus, the expression of the genetic code may be described by the following sequence:



Each of us is the sum total of our genetic characteristics. Blood type, hair color, muscle proteins, nerve cells, and thousands of other aspects of our structure and functioning have their basis in the genetic code of DNA.

If there is a "mistake" in the DNA, that is, incorrect bases or triplets of bases, this mistake will be copied by the mRNA. The result is the formation of a malfunctioning or non-functioning protein. This is called a **genetic** or **hereditary disease**.

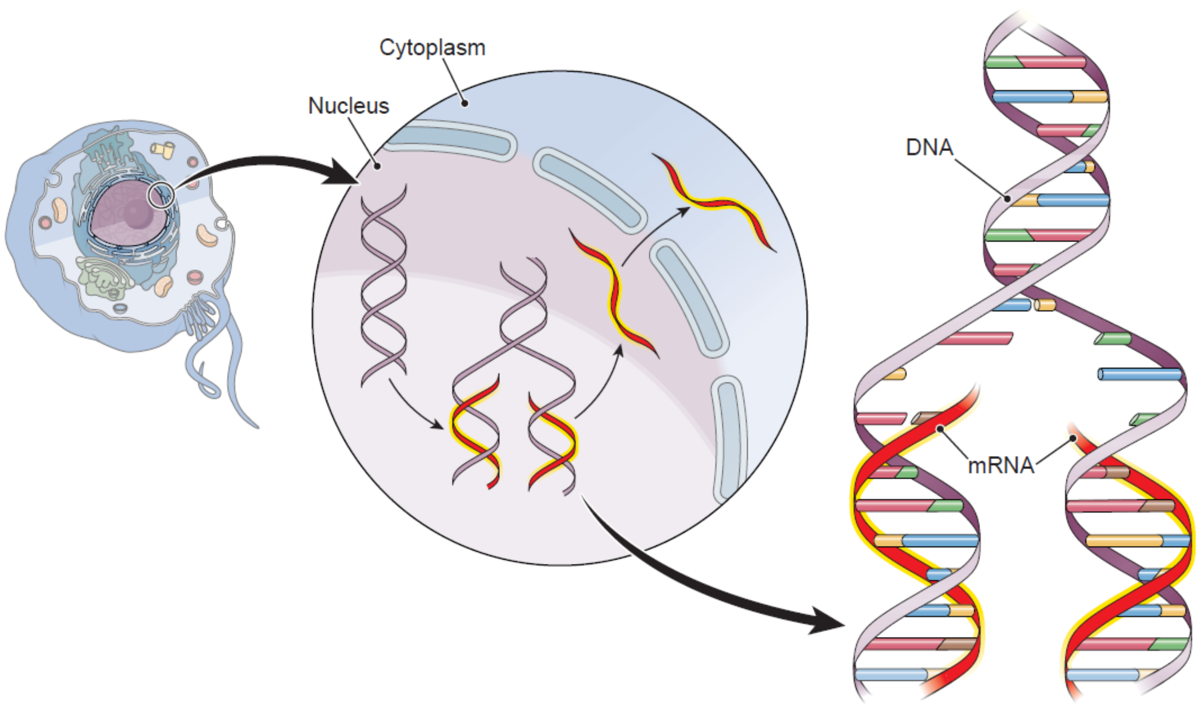


Figure: Transcription. In the first step of protein synthesis the DNA code is transcribed into messenger RNA (mRNA) by nucleotide base pairing.

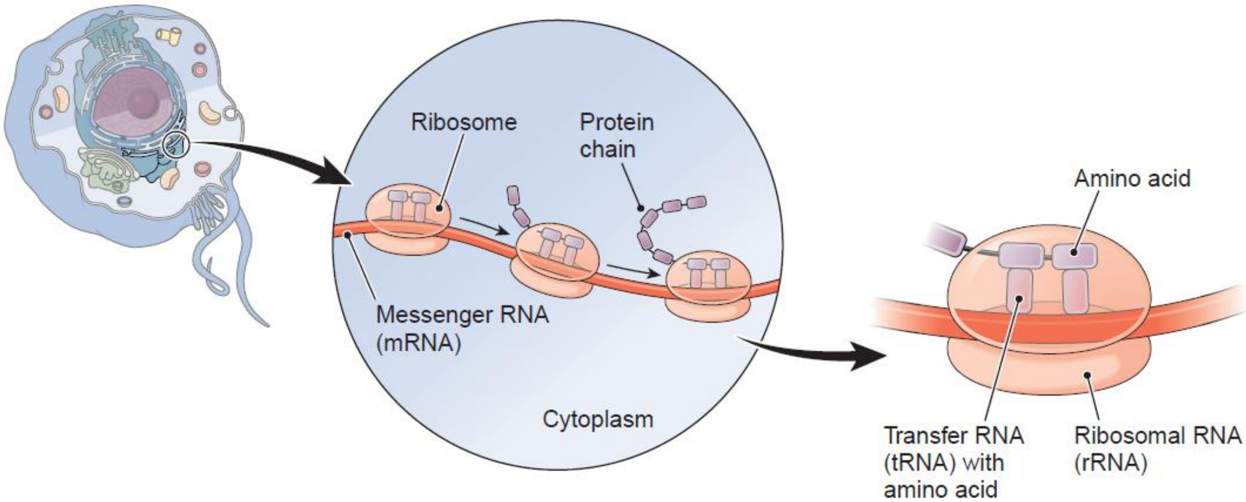


Figure: Translation. In protein synthesis, messenger RNA (mRNA) travels to the ribosomes in the cytoplasm. The information in the mRNA codes for the building of proteins from amino acids. Transfer RNA (tRNA) molecules bring amino acids to the ribosomes to build each protein.

Protein Synthesis

Molecule or Organelle	Function
DNA	• A double strand (helix) of nucleotides that is the genetic code in the chromosomes of cells. • A gene is the sequence of bases (segment of DNA) that is the code for one protein.
mRNA (messenger RNA)	• A single strand of nucleotides formed as a complementary copy of a gene in the DNA. • Now contains the triplet code: three bases is the code for one amino acid (a codon). • Leaves the DNA in the nucleus, enters the cytoplasm of the cell, and becomes attached to ribosomes.
Ribosomes	• The cell organelles that are the site of protein synthesis. • Attach the mRNA molecule. • Contain enzymes to form peptide bonds between amino acids
tRNA (transfer RNA)	• Picks up amino acids (from food) in the cytoplasm and transports them to their proper sites (triplets) along the mRNA molecule; has anticodons to match mRNA codons.

1.5.4 Cell division

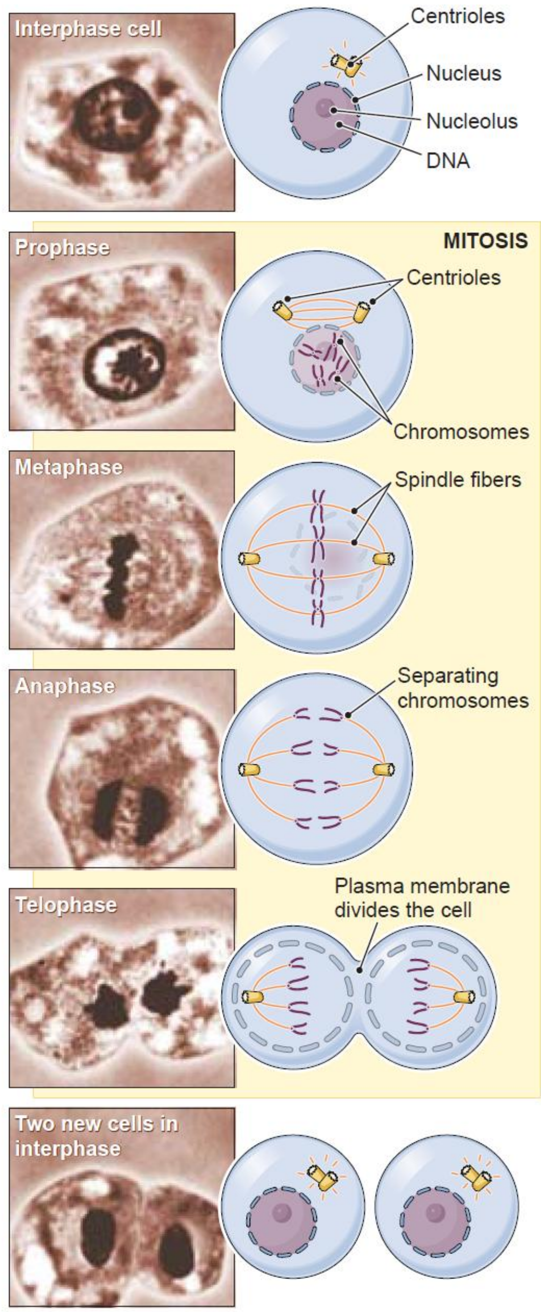
Cell division is the process by which a cell reproduces itself. There are two types of cell division: mitosis and meiosis. Although both types involve cell reproduction, their purposes are very different.

- **MITOSIS**

Each of us began life as one cell, a fertilized egg. Each of us now consists of billions of cells produced by the process of mitosis. In **mitosis**, one cell with the **diploid number** of chromosomes (the usual number, 46 for people) divides into two identical cells, each with the diploid number of chromosomes. This production of identical cells is necessary for the growth of the organism and for repair of tissues.

Before mitosis can take place, a cell must have two complete sets of chromosomes, because each new cell must have the diploid number. The process of **DNA replication** enables each chromosome (in the form of chromatin) to make a copy of itself.

The time during which this takes place is called **interphase**, the time between mitotic divisions. Although interphase is sometimes referred to as the resting stage, resting means "not dividing" rather than "inactive." The cell is quite actively producing a second set of chromosomes and storing energy in ATP.



(Figure: The stages of mitosis.)

The long, thin, and invisible chromatin molecules then begin to coil very precisely and extensively, and if we were looking at the nucleus of a living cell under a microscope, we would see the duplicated chromosomes appear. Each would look somewhat like the letter X, because the original DNA molecule and its copy (now called chromatids) are still attached.

The stages of mitosis are **prophase**, **metaphase**, **anaphase**, and **telophase**. Mitosis is essential for repair of tissues, to replace damaged or dead cells. One of the functions of red bone marrow is the production of red blood cells. Because red blood cells have a life span of only about 120 days, new ones are needed to replace the older ones that die. Very rapid mitosis in the red bone marrow produces approximately 2 million new red blood cells every second.

These dividing cells in the red bone marrow are among the stem cells present in the body. A **stem cell** is an unspecialized cell that may develop into several different kinds of cells.

The stages of mitosis

Stage	**Events**
Prophase	1. The chromosomes coil up and become visible as short rods. Each chromosome is really two chromatids (original DNA plus its copy) still attached at a region called the centromere. 2. The nuclear membrane disappears. 3. The centrioles move toward opposite poles of the cell and organize the spindle fibers, which extend across the equator of the cell.
Metaphase	1. The pairs of chromatids line up along the equator of the cell. The centromere of each pair is attached to a spindle fiber. 2. The centromeres now divide.
Anaphase	1. Each chromatid is now considered a separate chromosome; there are two complete and separate sets. 2. The spindle fibers contract and pull the chromosomes, one set toward each pole of the cell.
Telophase	1. The sets of chromosomes reach the poles of the cell and become indistinct as their DNA uncoils to form chromatin. 2. A nuclear membrane re-forms around each set of chromosomes.
Cytokinesis	1. The cytoplasm divides; new cell membrane is formed.

- **MEIOSIS**

Meiosis is a more complex process of cell division that results in the formation of **gametes**, which are egg and sperm cells. In meiosis, one cell with the diploid number of chromosomes divides twice to form four number of chromosomes.

In women, meiosis takes place in the ovaries and is called **oogenesis**. In men, meiosis takes place in the testes and is called **spermatogenesis**.

The differences between oogenesis and spermatogenesis will be discussed in Chapter entitled "The Reproductive System". The egg and sperm cells produced by meiosis have the haploid number of chromosomes, which is 23 for humans. Meiosis is sometimes called reduction division because the division process reduces the chromosome number in the egg or sperm. Then, during **fertilization**, in which the egg unites with the sperm, the 23 chromosomes of the sperm plus the 23 chromosomes of the egg will restore the diploid number of 46 in the fertilized egg. Thus, the proper chromosome number is maintained in the cells of the new individual.

- **Applications to the nursing care**

1. **Medical Uses of Isotonic, Hypertonic, and Hypotonic Solutions**

RBCs and other body cells may be damaged or destroyed if exposed to hypertonic or hypotonic solutions. For this reason, most **intravenous (IV) solutions,** liquids infused into the blood of a vein, are isotonic.

Examples: Isotonic saline (0.9% NaCl) and D5W, which stands for dextrose 5% in water. Sometimes infusion of a hypertonic solution such as Mannitol is useful to treat patients who have *cerebral edema,* excess interstitial fluid in the brain.

Infusion of such solution relieves fluid overload by causing osmosis of water from interstitial fluid into the blood.

The kidneys then excrete the excess water from the blood into the urine.

Hypotonic solutions, given either orally or through an IV, can be used to treat people who are dehydrated. The water in the hypotonic solution moves from the blood into interstitial fluid and then into body cells to rehydrate them. Water and most sports drinks that you consume to "rehydrate" after a workout are hypotonic relative to your body cells.

2. **Digitalis Increases Ca_ions in Heart Muscle Cells**

Digitalis often is given to patients with *heart failure*, a condition of weakened pumping action by the heart. Digitalis exerts its effect by slowing the action of the sodium-potassium pumps, which lets more Na^+ accumulates inside heart muscle cells.

The result is a decreased Na^+ concentration gradient across the plasma membrane, which causes the Na^+/Ca^{2+} antiporters to slow down. As a result, more Ca^{2+} remains inside heart muscle cells. The slight increase in the level of Ca^{2+} in the cytosol of heart muscle cells increases the force of their contractions and thus strengthens the force of the heartbeat.

3. **Viruses and Receptor-Mediated Endocytosis**

Although receptor-mediated endocytosis normally imports needed materials, some viruses are able to use this mechanism to enter and infect body cells.

For example, the human immunodeficiency virus (HIV), which causes acquired immunodeficiency syndrome (AIDS), can attach to a receptor called CD4.

This receptor is present in the plasma membrane of white blood cells called helper T cells. After binding to CD4, HIV enters the helper T cell via receptor-mediated endocytosis.

4. **Smooth ER and Drug Tolerance**

One of the functions of smooth ER, as noted earlier, is to detoxify certain drugs.

Individuals who repeatedly take such drugs, such as the sedative phenobarbital, develop changes in the smooth ER in their liver cells.

Prolonged administration of phenobarbital results in increased tolerance to the drug; the same dose no longer produces the same degree of sedation. With repeated exposure to the drug, the amount of smooth ER and its enzymes increases to protect the cell from its toxic effects.

As the amount of smooth ER increases, higher and higher dosages of the drug are needed to achieve the original effect.

5. Mitotic Spindle and Cancer

One of the distinguishing features of cancer cells is uncontrolled division.

The mass of cells resulting from this division is called a neoplasm or tumor. It may be benign or malignant. One of the ways to treat cancer is by chemotherapy, the use of anticancer drugs. Some of these drugs stop cell division by inhibiting the formation of the mitotic spindle. Unfortunately, these types of anticancer drugs also kill all types of rapidly dividing cells in the body, causing side effects such as nausea, diarrhea, hair loss, fatigue, and decreased resistance to disease.