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Разделы — цитология и генетика

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для студентов I курса, обучающихся на английском языке

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Chapters — cytology and genetics

Training aid in biology
for I course English-speaking students

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CHAPTER I. CYTOLOGY

Topic 1. Cells: the basic units of life. Main statements of the cell theory

Cytology (word comes from Greek: cytos — a cell, a cavity; logos — a science) — the section of biology that studies the cell structure and function.

The cell's discovery is related with the name of Robert Hooke (1635–1703). He was first who describe cell structure of plants. Developments in cytology in the 1800s had a strong influence on other sections of biology. Robert Brown (1773–1858) described the cell nucleus in 1833. Building on the work of others, Matthis Jacob Schleiden (1804–1881) and Theodor Schwann (1810–1882) proposed the concept of the **cell theory** in 1839. According to this theory: all life is composed of cells, cells arise only from preexisting cells, and the **cell** is the fundamental unit of structure and function in living organisms.

The cell theory has three important implications. First, it means that studying cell biology is in some sense the same as studying life. The principles that underlie the functions of the single cell in a bacterium are similar to those governing the 60 trillion cells of your body. Second, it means that life is continuous. Finally, it means that the origin of life on Earth was marked by the origin of the first cells.

Cells are so effective at capturing energy and replicating themselves — two fundamental characteristics of life — that since they evolved, cells have been the unit on which all life is built. A cell is not only the structural unit of a life, but also elementary functional unit of all living things because provides exchange of energy and substances, reproduction, growth and development, irritability and movement, heredity and diversity, homeostasis.

All living things are represented by **unicellular** organisms that have only one cell (for example amoeba, infusorians, and etc.) or **multicellular** organisms that composed of many cells. Fungi, plants, and animals including human are multicellular organisms.

Most cells are tiny and have different size and shape. The volume of cells ranges from 1 to 1,000 μm^3 and most of cells have size — from several to 100 μm (figure 1).

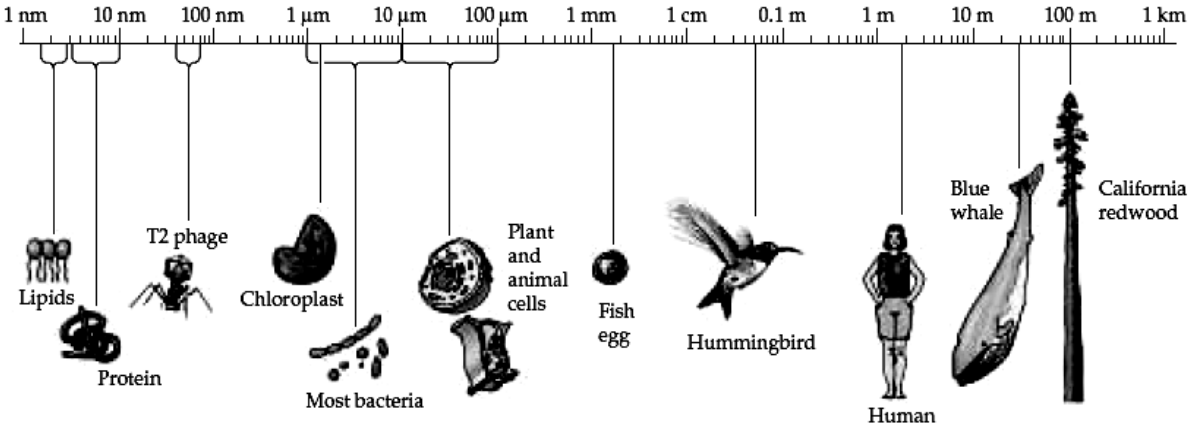

Figure 1 — This logarithmic scale shows the relative sizes of molecules, cells, and multicellular organisms

The eggs of some birds are enormous exceptions and also individual cells of several types of algae and bacteria are large enough. And although neurons (nerve cells) have a volume that is within the «normal» cell range, they often have fine projections that may extend for meters, carrying signals from one part of a large animal to another. It means that cell's shape can depend of their function (figure 2).

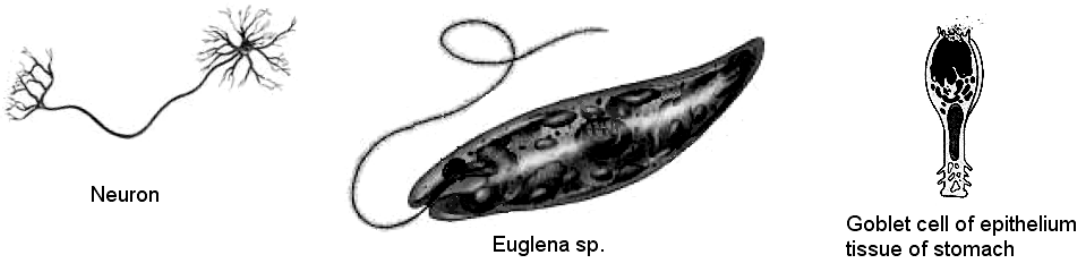

Figure 2 — Examples of different shape of cells

Biologists traditionally classify all living organisms into two major groups, the **prokaryotes** and the **eukaryotes**, depending of cell's organizational patterns. **Prokaryotic** cell organization is characteristic for unicellular organisms of the domains Bacteria and Archaea. Organisms in these domains are called prokaryotes. Their cells do not have membrane-enclosed internal compartments (nucleus and organelles). **Eukaryotic** cell organization is found in the domain Eukarya, which includes the protists, plants, fungi, and animals. Their cells have a special membrane-enclosed compartment called the nucleus. Eukaryotic cells also contain other membrane-enclosed compartments in which specific chemical reactions take place. Organisms with this type of cell organization are known as eukaryotes.

Cell membrane, cytoplasm, and nucleus are the structural elements of eukaryotic cells (figure 3).

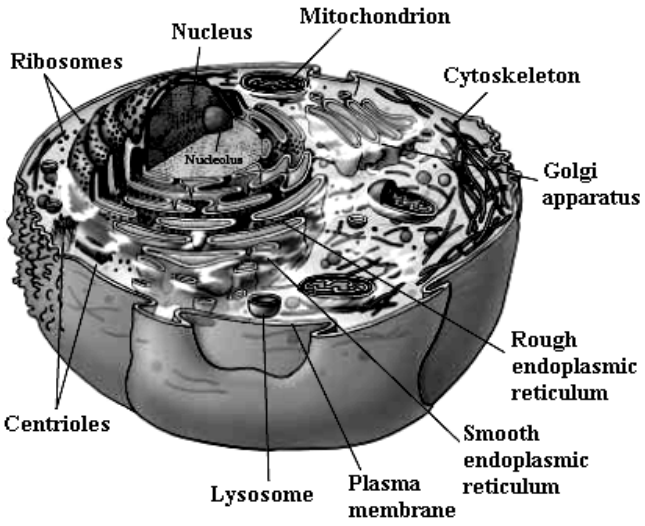

Figure 3 — Structure and organelles of animal cell

Comprehension questions

1. What cytology study?
2. Who discover a cell?
3. Who proposed the concept of the cell theory and when it was proposed?
4. Give main postulates of the cell theory.
5. What are important implications of the cell theory?
6. Why the cell is the structural and functional unit of life?
7. Give example of animals composed of only one cell.
8. What organisms are multicellular?
9. What have influence on cell's shape?
10. What volume and size can have cells?
11. Give example of biggest cell.
12. What cells are prokaryotic and eukaryotic?
13. What domains include prokaryotes?
14. What domain includes eukaryotes?
15. What are the structural elements of eukaryotic cells?

Definitions of the terms

Cell is the fundamental unit of structure and function in living organisms.

Cytology is the section of biology that studies the cell structure and function.

Eukaryotic cell — it is cell that has nucleus and other membrane-enclosed compartments in which specific chemical reactions take place.

Multicellular organisms — it is organisms that composed of many cells.

Prokaryotic cell — it is cell that does not has membrane-enclosed internal compartments.

Unicellular organisms — it is organisms that have only one cell in their structure.

Topic 2. Cell membrane

A **cell (plasma) membrane** separates each cell from its environment, creating a segregated (but not isolated) compartment.

The physical organization and functioning of all biological membranes depend on their constituents: lipids, proteins, and carbohydrates.

The *lipids* establish the physical integrity of the membrane and create an effective barrier to the rapid passage of hydrophilic materials such as water and ions. In addition, the phospholipid bilayer serves as a lipid «lake» in which a variety of proteins «float». This general design is known as the **FLUID MOSAIC MODEL** (figure 4).

Proteins embedded in the phospholipid bilayer have a number of functions, including moving materials through the membrane and receiving chemical signals from the cell's external environment. Each membrane has a set of proteins suitable to the specialized function of the cell or organelle it surrounds.

The *carbohydrates* associated with membranes are attached either to the lipids or to protein molecules. They are located on the outside of the plasma membrane, where they protrude into the environment, away from the cell. Like some of the proteins, carbohydrates are crucial in recognizing specific molecules.

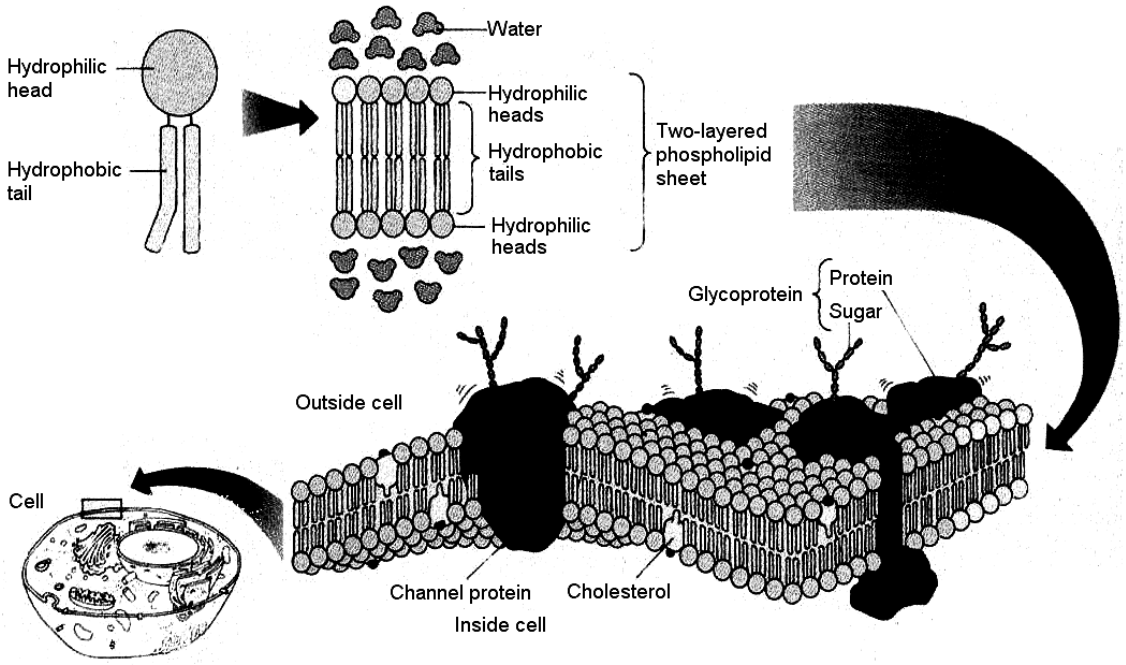

Figure 4 — The general molecular structure of biological membranes is a continuous phospholipid bilayer in which proteins are embedded

Structure of phospholipid bilayer

Most of the lipids in biological membranes are phospholipids. Phospholipids have both hydrophilic regions and hydrophobic regions:

• *Hydrophilic regions:* The phosphorus-containing «head» of the phospholipid is electrically charged and hence associates with polar water molecules.

• *Hydrophobic regions:* The long, nonpolar fatty acid «tails» of the phospholipid associate with other nonpolar materials, but they do not dissolve in water or associate with hydrophilic substances.

As a consequence of these properties, one way in which phospholipids can coexist with water is to form a *bilayer*, with the fatty acids of the two layers interacting with each other and the polar regions facing the outside aqueous environment.

Plasma membrane importance

The plasma membrane allows the cell to maintain a more or less *constant internal environment*.

The plasma membrane acts as a *selectively permeable barrier*, preventing some substances from crossing while permitting other substances to enter and leave the cell.

As the cell's boundary with the outside environment, the plasma membrane is important in *communicating with adjacent cells and receiving extracellular signals*.

The plasma membrane often has molecules protruding from it that are responsible for *binding and adhering* to adjacent cells.

Ways of receipt of substances in a cell

There are two fundamentally different kinds of processes by which substances cross biological membranes to enter and leave cells or organelles:

• **Passive transport** processes do not require any input of outside energy to drive them. The energy for these processes is in the substances themselves and the difference in their concentration on the two sides of the membrane. Passive transport processes include the different types of diffusion: simple diffusion through the phospholipids bilayer and facilitated diffusion through channel proteins or by means of carrier molecules. Water molecules are abundant enough and small enough that they move through membranes by a diffusion process called **osmosis**.

• **Active transport** processes, on the other hand, require the input of chemical energy (requires the direct participation of the energy-rich molecule ATP.). They do not use the intrinsic property of a concentration gradient.

Macromolecules such as proteins, polysaccharides, and nucleic acids are simply too large and too charged or polar to pass through membranes. This pass can be done by means of vesicles that either pinch off from the plasma membrane and enter the cell (endocytosis) or fuse with the plasma membrane and release their contents (exocytosis) (figure 5).

Endocytosis (figure 5a) is a general term for a group of processes that bring macromolecules, large particles, small molecules, and even small cells into the eukaryotic cell. There are two types of endocytosis: phagocytosis and pinocytosis. In all two, the plasma membrane invaginates (folds inward) around materials from the environment, forming a small pocket. The pocket deepens, forming a vesicle. This vesicle separates from the plasma membrane and migrates with its contents to the cell's interior.

In **phagocytosis** («cellular eating»), part of the plasma membrane engulfs large particles or even entire cells. Phagocytosis is used as a cellular feeding process by unicellular protists and by some white blood cells that defend the body by engulfing foreign cells and substances. The food vacuole or phagosome that forms usually fuses with a lysosome, where its contents are digested.

In **pinocytosis** («cellular drinking»), vesicles also form. However, these vesicles are smaller, and the process operates to bring small dissolved substances or fluids into the cell. Like phagocytosis, pinocytosis is relatively nonspecific as to what it brings into the cell. For example, pinocytosis goes on constantly in the *endothelium*, the single layer of cells that separates a tiny blood

capillary from its surrounding tissue, allowing the cells to rapidly acquire fluids from the blood.

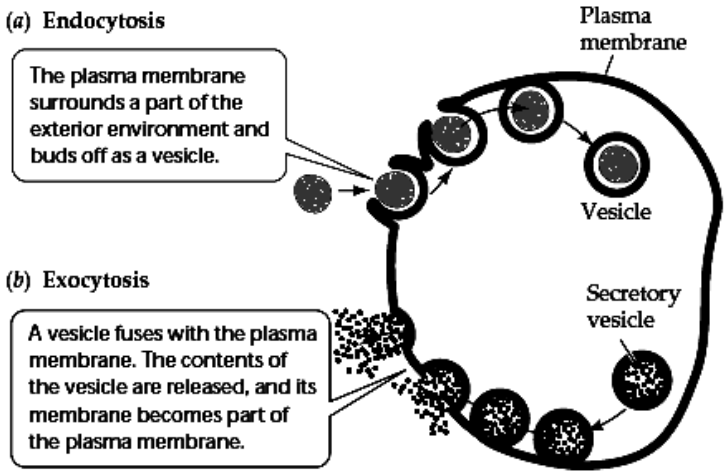

Figure 5 — Endocytosis (a) and exocytosis (b) are used by all eukaryotic cells to take up substances from and release substances to the outside environment

Exocytosis (Figure 5b) is the process by which materials packaged in vesicles are secreted from a cell when the vesicle membrane fuses with the plasma membrane. The initial event in this process is the binding of a membrane protein protruding from the cytoplasmic side of the vesicle with a membrane protein on the cytoplasmic side of the target site on the plasma membrane. The phospholipid regions of the two membranes merge, and an opening to the outside of the cell develops. The contents of the vesicle are released to the environment, and the vesicle membrane is smoothly incorporated into the plasma membrane.

Comprehension questions

1. What is cell membrane?
2. List the constituents of plasma membrane.
3. Describe function of lipids in plasma membrane.
4. Why the general design of cell membrane is known as the fluid mosaic model?
5. Describe function of proteins in plasma membrane.
6. Describe function of carbohydrates in plasma membrane.
7. Describe structure of phospholipid bilayer.
8. Why plasma membrane is important for the cell?
9. What is passive transport?
10. List the types of passive transport.
11. What is active transport? Does it need in ATP energy?
12. What is endocytosis and exocytosis?
13. What is phagocytosis?
14. What is pinocytosis?
15. Give examples of phagocytosis and pinocytosis.

Definitions of the terms

Active transport is the movement of a substance across a biological membrane *against* a concentration gradient *with* the expenses of chemical energy.

Cell (plasma) membrane is a membrane that separates each cell from its environment, creating a segregated (but not isolated) compartment.

Diffusion is the process of random movement of solute substances across membranes toward a state of equilibrium.

Osmosis is the diffusion of water across membranes.

Passive transport is the movement of a substance across a biological membrane *along* a concentration gradient *without* the expenses of chemical energy.

Phagocytosis is engulfing of large particles or even entire cells by plasma membrane.

Pinocytosis is a mergence of small dissolved substances or fluids by the cell.

Selective permeability is the plasma membrane property to prevent crossing of some substances and to permit other substances to enter and leave the cell.

Topic 3. Cell's nucleus and organelles

The **nucleus** is a constant structural component of all eukaryotic cells that contains most of the cell's genetic material (DNA). The nucleus consist of **nuclear envelope, nucleoplasm, nucleolus,** and **chromatin** (a complex of DNA and histon proteins) (figure 6).

The nucleus is surrounded by two membranes, which together form the **nuclear envelope**. The two membranes of the nuclear envelope are separated by 10–20 nm and are perforated by **nuclear pores**, which connect the interior of the nucleus with the cytoplasm. RNA and proteins pass through these pores to enter or leave the nucleus.

The **nucleolus** begins the assembly of ribosomes from specific proteins and RNA.

Surrounding the chromatin are water and dissolved substances collectively referred to as the **nucleoplasm**.

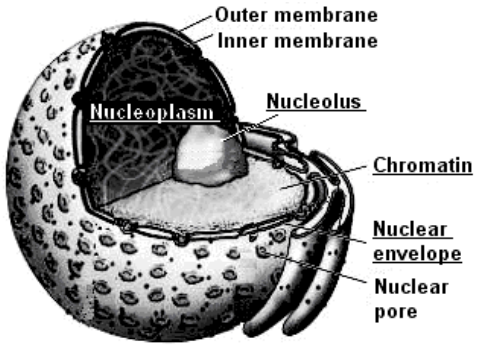

Figure 6 — The double-membraned nuclear envelope with nuclear pores, nucleolus, nucleoplasm, and chromatin are common features of all cell nuclei.
The pores are the gateways through which proteins from the cytoplasm enter the nucleus and genetic material (mRNA) from the nucleus enters the cytoplasm

At the start of cell division, the chromatin compact into chromosomes. The chromosome configuration is determined by presence of centromere. It divides a chromosome on 2 arms. The arrangement of centromere defines the basic forms of chromosomes (figure 7): metacentric (with equal arms); submetacentric (arms slightly different); acrocentric (arms strong different); telocentric (without arm).

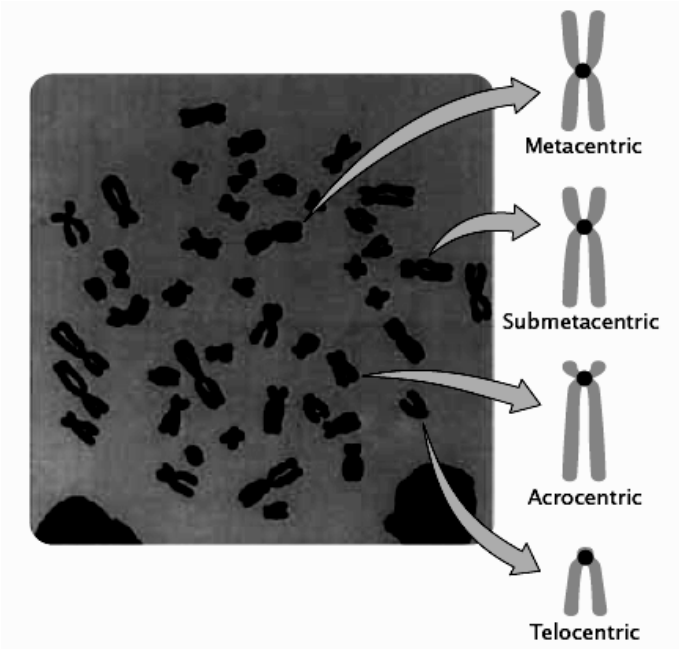

Figure 7 — On the basis of the location of the centromere, chromosomes are classified into four types: metacentric, submetacentric, acrocentric, and telocentric

Functions of a nucleus:

— storage of genetic information;

— transfer of genetic information;

— regulation of processes of a cell activity.

Organelles are the specialized constant components of the cytoplasm which are possess distinctive shapes and functions. Organelles shared on two groups:

✓ *General purpose:* mitochondria, Golgi apparatus, endoplasmic reticulum, ribosomes, lysosomes, centrioles, and also plastids and vacuoles which are characteristic for plant cells.

✓ *Special purpose*: cilia and flagella.

Cytoplasmic inclusions are changeable structures in the cytoplasm, representing metabolic products of cells.

In prokaryotic cells, **ribosomes** float freely in the cytoplasm. In eukaryotic cells they occur in two places: in the cytoplasm, where they may be free or attached to the surface of the endoplasmic reticulum; and inside the mitochondria and chloroplasts, where energy is processed. In each of these locations, the ribosomes are the sites where proteins are synthesized under the direction of nucleic acids. Chemically, ribosomes consist of a special type of RNA, called *ribosomal RNA* (*rRNA*), to which more than 50 different protein molecules are bound.

Endoplasmic reticulum is made up of a series of interrelated compartments enclosed by membranes. The rough endoplasmic reticulum has attached ribosomes that synthesize proteins. The smooth endoplasmic reticulum lacks ribosomes and is associated with the synthesis of lipids and steroids.

Golgi apparatus (named for its discoverer, *Camillo Golgi*) consists of flattened membranous sacs called *cisternae* and small membrane-enclosed vesicles. The cisternae appear to be lying together like a stack of saucers. Golgi apparatus concentrates, packages, and sorts proteins before they are sent to their cellular or extracellular destinations. It receives materials from the rough ER by means of vesicles that fuse with it. Some of these vesicles fuse with the plasma membrane and release their contents outside the cell. Golgi apparatus yet is the place where some polysaccharides for the plant cell wall are synthesized.

Originating in part from the Golgi apparatus are organelles called **lysosomes**. They contain many digestive enzymes which digest the engulfed materials. Undigested materials are secreted from the cell when the secondary lysosome fuses with the plasma membrane. Plant cells do not appear to contain lysosomes, but the central vacuole of a plant cell may function in an equivalent capacity because it, like lysosomes, contains many digestive enzymes.

Mitochondria are enclosed by an outer membrane and an inner membrane that folds inward to form cristae. Mitochondria contain the proteins needed for cellular respiration and production of ATP.

Centrioles, made up of triplets of microtubules, are involved in the distribution of chromosomes during cell division. Centrioles are found in all eukaryotes except the flowering and conifer plants, and some protists.

The cells of photosynthetic eukaryotes contain **chloroplasts**. These organelles are enclosed by double membranes and contain an internal system of thylakoids organized as grana. Thylakoids within chloroplasts contain the chlorophyll and proteins that harvest light energy for photosynthesis. Both mitochondria and chloroplasts contain their own DNA and ribosomes and are capable of making some of their own proteins.

Vacuoles are prominent in many plant cells and consist of a membrane-enclosed compartment full of water and dissolved substances. By taking in water, vacuoles enlarge and provide the pressure needed to stretch the cell wall and provide structural support for the plant.

Many eukaryotic cells possess **flagella** or **cilia**, or both. These whip like organelles may push or pull the cell through its aqueous environment, or they may move surrounding liquid over the surface of the cell.

Comprehension questions

1. What is cell nucleus?
2. What are the structural elements of nucleus?
3. Name the functions of nucleus.
4. What basic forms of chromosomes you are know?
5. What are organelles and cytoplasmic inclusions?

6. Name the base groups of organelles.
7. Describe the localization and function of ribosome.
8. Describe the structure and function of endoplasmic reticulum.
9. Describe the function of Golgi apparatus.
10. Describe the function of lysosome. What lysosomes contain?
11. Describe the structure and function of mitochondrion.
12. What organelle contains in animal cells only?
13. Describe the structure and function of centrioles.
14. What organelles contain in plant cells only?
15. Describe the structure and function of chloroplasts.
16. Describe the function of vacuole**.**
17. Describe the function of flagella and cilia.

Definitions of the terms

Centrioles are the cell's organelles that involved in the distribution of chromosomes during cell division.

Chloroplasts are the plant cell organelles that harvest light energy for photosynthesis.

Chromosome is a condensed chromatin that forms at the start of cell division.

Chromatin is a complex of DNA and histon proteins.

Cytoplasmic inclusions are changeable structures in the cytoplasm, representing metabolic products of cells.

Endoplasmic reticulum is the cell's organelle that takes part in synthesis of proteins, lipids and steroids.

Flagella and **cilia** are the cell's organelles that may push or pull the cell through its aqueous environment, or they may move surrounding liquid over the surface of the cell.

Golgi apparatus is the cell's organelle that concentrates, packages, and sorts proteins before they are sent to their cellular or extracellular destinations.

Lysosomes are the animal cell organelles that take part in breakdown of food and foreign objects taken up by the cell.

Mitochondria are the cell's organelles that need for cellular respiration and production of ATP.

Nucleus is a constant structural component of all eukaryotic cells that contains most of the cell's genetic material (DNA).

Organelles — are the specialized constant components of the cytoplasm which are possess distinctive shapes and functions.

Ribosomes are the cell's organelles where proteins are synthesized under the direction of nucleic acids.

Vacuoles provide the pressure needed to stretch the cell wall and provide structural support for the plant.

Topic 4. Cell reproduction

Reproduction is an ability of living things to engender they like. Cells are reproduced by division. All cells of organism except sex cells are named **so-**

matic. Set of chromosomes in somatic cells is named **diploid** (2n). Set of chromosomes in sex cells is named **haploid** (n) and equal to half of somatic cells set.

Unicellular organisms use cell division primarily to reproduce themselves, whereas in multicellular organisms cell division also plays important roles in growth and in the repair of tissues. Prokaryotes divide by fission. Eukaryotic cells divide by mitosis or meiosis.

The time between cell formation by mother cell division and it own division or death is called the **cell cycle**. **Mitotic cycle** has a time between two cell division ends.

Periods of cell cycle (figure 8)**:**

1. Between divisions — interphase when the cell grows, performs its functions, and prepared for division. Interphase has three subphases — G_1, S, and G_2.

2. Division of a cell — mitosis (table 1).

In mitosis, a single nucleus gives rise to two nuclei that are genetically identical to each other and to the parent nucleus. This process ensures the accurate distribution of the eukaryotic cell's multiple chromosomes to the daughter nuclei. In reality, mitosis is a continuous process in which each event flows smoothly into the next. For discussion, however, it is convenient to look at mitosis — the M phase of the cell cycle - as a series of separate events: prophase, prometaphase, metaphase, anaphase, and telophase.

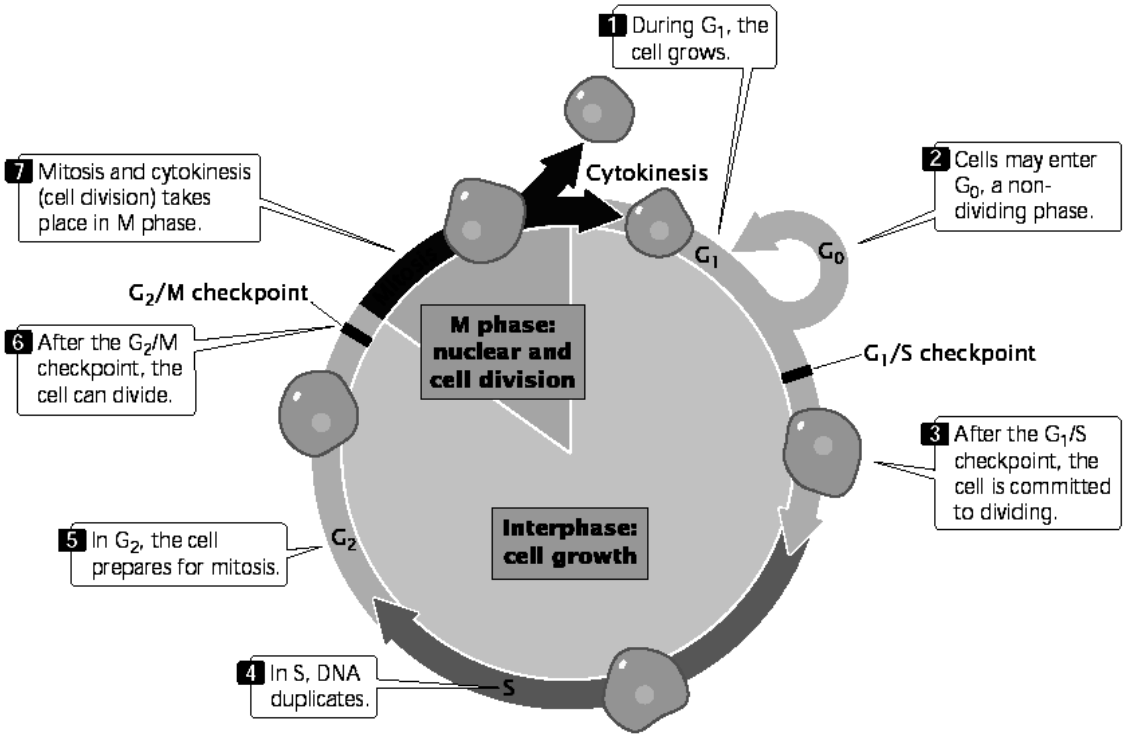

Figure 8 — The cell cycle consists of a mitotic (M) phase, during which first nuclear division (mitosis) and then cell division (cytokinesis) take place. The M phase is followed by a long period of growth known as interphase. Interphase has three subphases (G1, S, and G2) in cells that divide

Table 1 — Stages of interphase and mitosis

Stage	Major features
Interphase	
G_1 phase	Growth and development of the cell
S phase	Synthesis of DNA
G_2 phase	Preparation for division
M phase	
Prophase	Chromosomes condense and mitotic spindle forms
Prometaphase	Nuclear envelope disintegrates, spindle microtubules anchor to kinetochores
Metaphase	Chromosomes align on the metaphase plate
Anaphase	Sister chromatids separate, becoming individual chromosomes that migrate toward spindle poles
Telophase	Chromosomes arrive at spindle poles, the nuclear envelope re-forms and the condensed chromosomes relax
Cytokinesis	Cytoplasm divides; cell wall forms in plant cells

The basic unit of the eukaryotic chromosome is a double-stranded molecule of DNA complexed with many proteins to form a chromatin (figure 9). Before the S phase, each chromosome contains only one such double-stranded DNA molecule. However, after the DNA molecule replicates during the S phase, the two resulting DNA molecules, now called **chromatids**, are held together along most of their length. They stay this way until mitosis, when chromatids are separated.

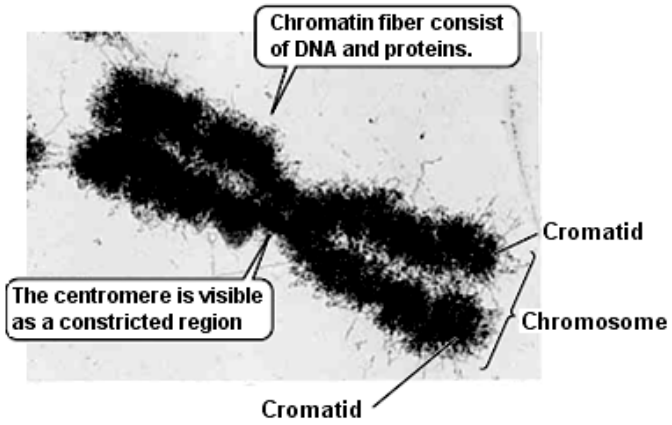

Figure 9 — A human chromosome, shown as the cell prepares to divide

Significance of mitosis:

In mitosis, a single nucleus gives rise to two nuclei that are genetically identical to each other and to the parent nucleus. This process ensures the accurate distribution of the eukaryotic cell's multiple chromosomes to the daughter nuclei (figure 10).

In multicellular organisms, each of somatic cells contains two sets of chromosomes, which are found in pairs. One chromosome of each pair of **homologous chromosomes** comes from each of the organism's two parents. Gametes, on the other hand, contain only a single set (haploid) of chromosomes. Two

haploid gametes fuse to form a new organism in a process called fertilization. The resulting zygote thus has two sets of chromosomes, just as somatic cells do.

Meiosis is the cell division that reduces the number of chromosomes to the haploid number in preparation for sexual reproduction (table 2, figure 10).

Functions of meiosis:

(a) To reduce the chromosome number from diploid to haploid;

(b) To ensure that each of the gametes has a complete set of chromosomes;

(c) To promote genetic diversity among the progeny.

Table 2 — Major events in each stage of meiosis

Stage	Major Events
Meiosis I	
Prophase I	Chromosomes condense, homologous pairs of chromosomes synapse, crossing over takes place, nuclear envelope breaks down, and mitotic spindle forms.
Metaphase I	Homologous pairs of chromosomes line up on the metaphase plate.
Anaphase I	The two chromosomes (each with two chromatids) of each homologous pair separate and move toward opposite poles.
Telophase I	Chromosomes arrive at the spindle poles.
Cytokinesis	The cytoplasm divides to produce two cells, each having half the original number of chromosomes.
Interkinesis	In some cells the spindle breaks down, chromosomes relax, and a nuclear envelope re-forms, but no DNA synthesis takes place.
Meiosis II	
Prophase II	Chromosomes condense, the spindle forms, and the nuclear envelope disintegrates.
Metaphase II	Individual chromosomes line up on the metaphase plate.
Anaphase II	Sister chromatids separate and migrate as individual chromosomes toward the spindle poles.
Telophase II	Chromosomes arrive at the spindle poles; the spindle breaks down and a nuclear envelope re-forms.
Cytokinesis	The cytoplasm divides.

Comprehension questions

1. What is reproduction?
2. What cells are named somatic?
3. What set of chromosomes have somatic cells?
4. What is called somatic cells division?
5. What is interphase?
6. Describe cell processes that take place during interphase.
7. Describe distribution of DNA molecules during S phase of interphase.
8. Name the phases of mitosis.
9. Describe the prophase of mitosis.
10. Describe the prometaphase of mitosis.
11. Describe the metaphase of mitosis.
12. Describe the anaphase of mitosis.

13. Describe the telophase of mitosis.
14. What is cytokinesis?
15. What is significance of mitosis?
16. What is meiosis?
17. What set of chromosomes have sex cells?
18. Describe the functions of meiosis.
19. Name the phases of meiosis I and II.
20. Describe the phases of meiosis I.
21. Describe the phases of meiosis II.
22. What is crossing over?
23. What is interkinesis?

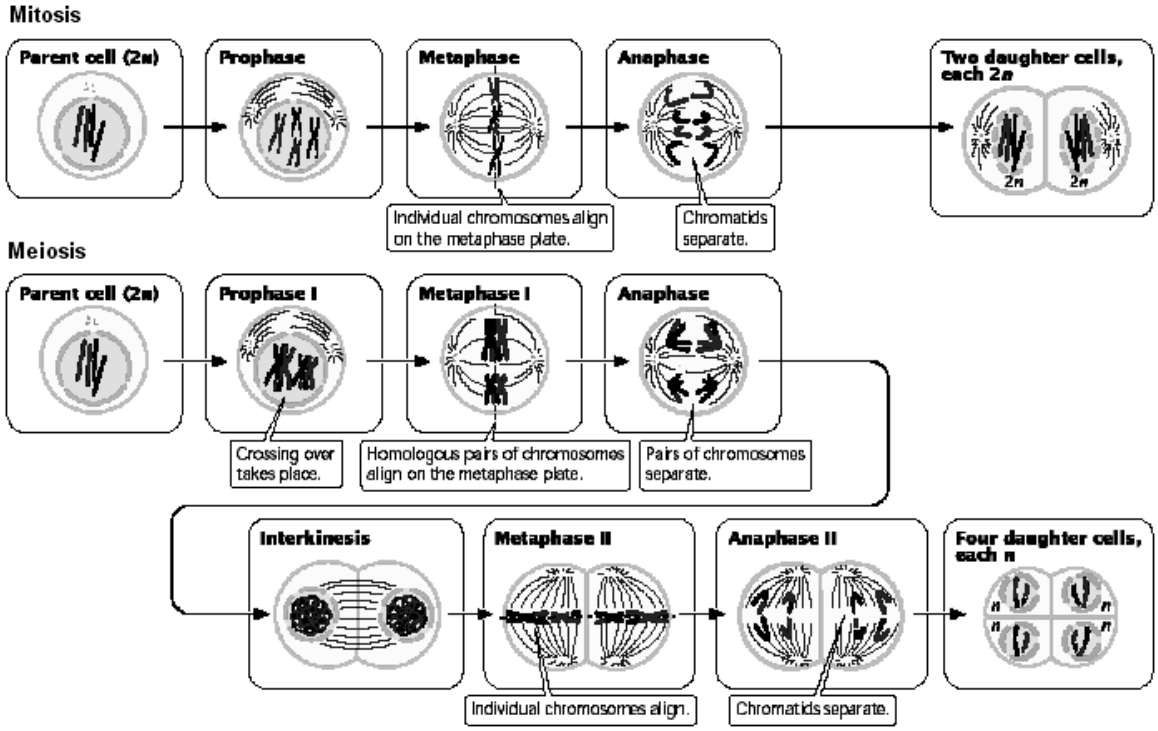

Figure 10 — Comparison of mitosis and meiosis

Definitions of the terms

Cell cycle is the time between cell formation by mother cell division and it own division or death.

Crossing over is an exchange of genetic material between homologous chromosomes.

Diploid set of chromosomes is a double set of chromosomes (2n) in somatic cells.

Haploid set of chromosomes is set of chromosomes in sex cells that equal to half of somatic cells set (n).

Homologous chromosomes are a couple of chromosomes that are similar in size and appearance.

Interkinesis is a brief interphase between meiosis I and II in which DNA does not replicate.

Interphase is a period between two mitotic divisions.

Meiosis is the cell division that reduces the number of chromosomes to the haploid number in preparation for sexual reproduction.

Mitosis is a division of somatic cell that produces two cells that are genetically identical to each other and to the parent cell.

Mitotic cycle is the time between two cell division ends.

Reproduction is an ability of living things to engender they like.

Somatic cells are all cells of organism except sex cells.

Sex cells are cells that take part in reproduction.

CHAPTER II. GENETICS

Topic 5. The chemical nature of DNA

A role of DNA in transfer of the genetic information had been proved by the discovering of the phenomenon of **transformation** and **transduction** in microorganisms. Transformation is ability of DNA of one bacterium to build into sites of DNA molecule of another bacterium and thus to transfer of inherited traits from one to another bacterium. The phenomenon was first observed in 1928 by Fred Griffith, an English physician whose special interest was the bacterium that causes pneumonia, *Streptococcus pneumonia.* Transduction is ability of bacteriophages to transfer fragments of DNA from one bacterium to another. The phenomenon was first observed in 1952 by Alfred Hershey and Martha Chase. They infected the bacterium *Escherichia coli* by bacteriophage T2.

Much of the basic chemistry of DNA was determined by Miescher, Kossel, Levene, Chargaff, and others, who had established that nucleic acids consisted of mononucleotides, and that each mononucleotide contained a sugar, nitrogen-containing base, and phosphate group (figure 11). Nucleic acids exist in two forms: **desoxyribonucleic acid (DNA)** and **ribonucleic acid (RNA)**. The nitrogenous bases in DNA are of four types: **adenine, guanine, cytosine**, and **thymine** (abbreviated A, G, C, and T), and the sequence of these bases encodes genetic information. Most organisms carry their genetic information in DNA, but a few viruses carry it in RNA. The four nitrogenous bases of RNA are abbreviated A, C, G, and U (**uracyl**).

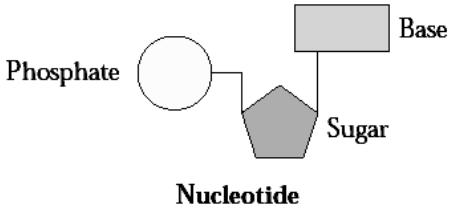

Figure 11 — Mononucleotide consists of a sugar, nitrogen-containing base, and phosphate group

The sugars of DNA and RNA are slightly different in structure also. RNA's ribose sugar has a hydroxyl group attached to the 2`-carbon atom, whereas DNA's sugar, called deoxyribose, has a hydrogen atom at this position (figure 12). This difference gives rise to the names ribonucleic acid (RNA) and deoxy-ribonucleic acid (DNA).

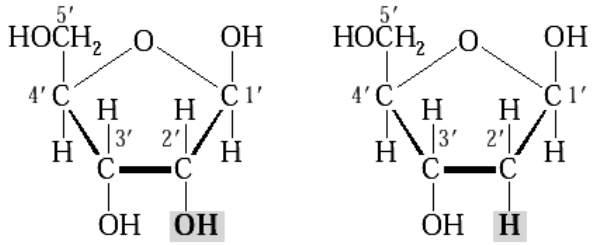

Figure 12 — Differences in sugar structures of RNA and DNA

In the 1940 s Erwin Chargaff and his colleagues carefully measured the amounts of the four bases in DNA from a variety of organisms and found that:

(a) The total amount of adenine is always equal to the amount of thymine (A = T).

(b) The amount of guanine is equal to cytosine (G = C).

(c) The amount of purines (A and G) is equal to pyrimidines (T and C).

(d) The number of the bases with 6-amino groups is equal to the number of the bases with 6-ketogroups (A+C = G+T).

(e)The ratio of bases A+T/G+C is species-specific.

These findings became known as Chargaff 's rules.

In 1947, William Ashbury began studying the three dimensional structure of DNA by using a technique called X-ray diffraction. Then, Watson and Crick investigated the structure of DNA, not by collecting new data but by using all available information about the chemistry of DNA to construct molecular models. The model developed by Watson and Crick showed that DNA consists of two strands of nucleotides wound around each other to form a right-handed helix, with the sugars and phosphates on the outside and the bases in the interior. These findings can be formulated as Watson and Crick's postulates:

(a) Each DNA molecule consist of two long antiparallel polynucleotide chains (strands) wind around each other making a double helix.

(b) Each nucleoside (sugar + the nitrogenous base) is located in the plane, which has a right angle with helix axis.

(c) Two strands are bounded to each other by the hydrogen bonds between bases.

(d) Pairing of the bases is strictly specific. There is only two possible pairs: A:T and G:C.

(e) *The sequence of the bases of one strand can considerably vary, but their sequence in other strand has to be complementary to it. I.e., the sequence of* the bases of one strand defines the complementary sequence in other.

DNA is made up of many nucleotides connected by covalent bonds, which join the 5`-phosphate group of one nucleotide to the 3`-carbon atom of the next nucleotide. These bonds, called phosphodiester linkages, are relatively strong covalent bonds; a series of nucleotides linked in this way constitutes a polynucleotide strand. RNA nucleotides also are connected by phosphodiester linkages to form similar single polynucleotide strand.

Levels of DNA packing

The DNA molecules in eukaryotic chromosomes are highly folded and condensed; if stretched out, some human chromosomes would be several centimeters long — thousands of times longer than the span of a typical nucleus.

In the course of the cell cycle, the level of DNA packaging changes — chromosomes progress from a highly packed state to a state of extreme condensation.

Eukaryotic DNA is closely associated with proteins, creating chromatin. The two basic types of chromatin are: **euchromatin**, which undergoes the normal process of condensation and decondensation in the cell cycle, and **heterochromatin,** which remains in a highly condensed state throughout the cell cycle, even during interphase.

The most abundant proteins in chromatin are the **histones**, positively charged proteins of five major types: H1, H2A, H2B, H3, and H4. The positive charges attract the negative charges on the phosphates of DNA and holds the DNA in contact with the histones which form **nucleosomes.** ***The nucleosome is the fundamental repeating unit of chromatin.*** The nucleosome core particle consists of two copies each of H2A, H2B, H3, and H4, around which DNA (about 145–147 bp of DNA) coils. Together, nucleosomes linked by linker DNA and H1 histone are called the *nucleosome string*. At a more complex level, the DNA molecule is associated with non histone proteins and is highly folded to produce a chromosome. Nucleosomes are folded into a 30-nm fiber that forms a series of 300-nm-long loops. The 300-nm loops are condensed to form a fiber that is 250 nm in diameter, which is itself tightly coiled to produce a 700-nm-wide chromatid (figure 13).

Types of RNA

(a) **Ribosomal RNA** (rRNA), along with ribosomal protein subunits, makes up the ribosome, the site of protein assembly.

(b) **Messenger or matrix RNA** (mRNA) carries the coding instructions for polypeptide chains from DNA to the ribosome. After attaching to a ribosome, an mRNA molecule specifies the sequence of the amino acids in a polypeptide chain and provides a template for joining amino acids. Large precursor molecules, which are termed *pre-messenger RNAs* (pre-mRNAs), are the immediate products of transcription in eukaryotic cells.

(c) **Transfer RNA** (tRNA) serves as the link between the coding sequence of nucleotides in the mRNA and the amino acid sequence of a polypeptide

chain. Each tRNA attaches to one particular type of amino acid and helps to incorporate that amino acid into a polypeptide chain.

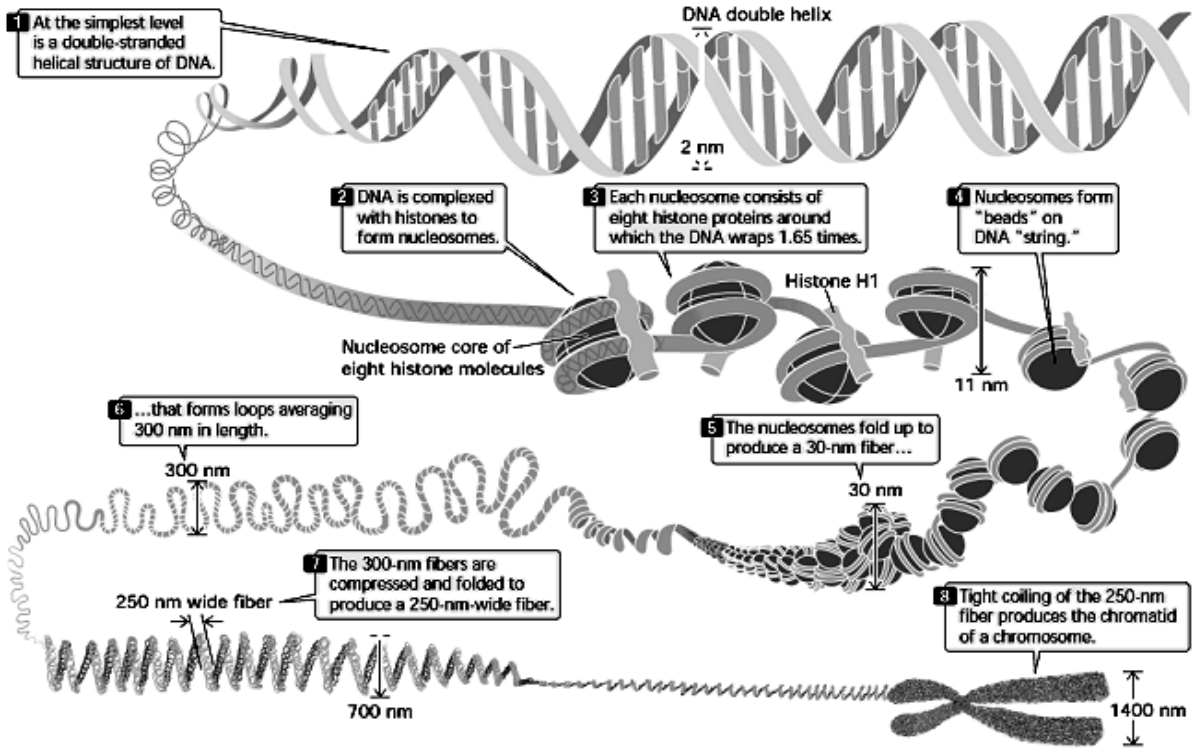

Figure 13 — Chromatin has a highly complex structure with several levels of organization

Comprehension questions

1. What experiments demonstrated that DNA is the genetic material?
2. Draw and label the three parts of a DNA nucleotide.
3. How does an RNA nucleotide differ from a DNA nucleotide?
4. List the Chargaff's rules.
5. List the Watson and Crick's postulates.
6. Describe the levels of DNA packing.
7. List types of RNA and describe their function.

Definitions of the terms

Transformation is ability of DNA of one bacterium to build into sites of DNA molecule of another bacterium and thus to transfer of inherited traits from one to another bacterium.

Transduction is ability of bacteriophages to transfer fragments of DNA from one bacterium to another.

DNA — desoxyribonucleic acid**.**

RNA — ribonucleic acid.

Euchromatin is the type of chromatin, which undergoes the normal process of condensation and decondensation in the cell cycle.

Heterochromatin is the type of chromatin**,** which remains in a highly condensed state throughout the cell cycle, even during interphase.

Histones are positively charged proteins associated with DNA.

Nucleosome is the fundamental repeating unit of chromatin.

Topic 6. From DNA to Protein: Genotype to Phenotype

We can identify three major pathways of information flow in the cell (figure 14**)**: in **replication,** information passes from one DNA molecule to other DNA molecules; in **transcription**, information passes from DNA to RNA; and, in **translation**, information passes from RNA to protein. This concept of information flow was formalized by Francis Crick in a concept that he called the **central dogma of molecular biology**. The central dogma states that genetic information passes from DNA to protein in a one-way information pathway. It indicates that genotype codes for phenotype but phenotype cannot code for genotype.

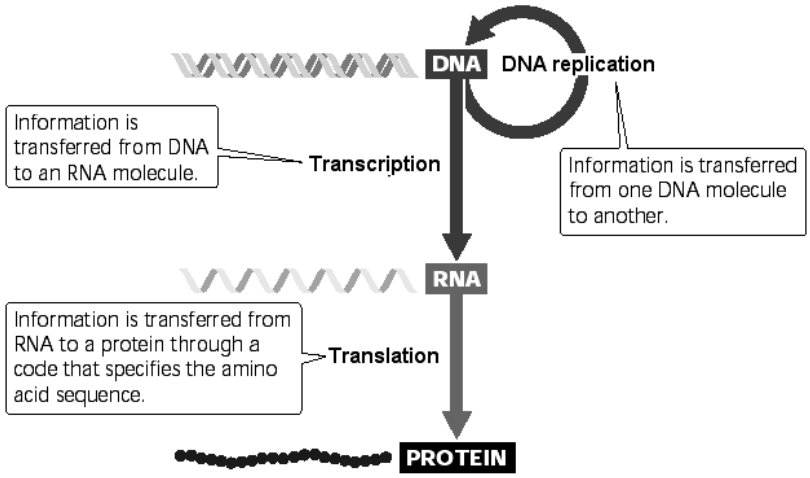

Figure 14 — The three major pathways of information transfer within the cell are replication, transcription, and translation

Replication

Initially, three alternative models were proposed for DNA replication.

• In **conservative replication**, the entire double-stranded DNA molecule serves as a template for a whole new molecule of DNA, and the original DNA molecule is *fully* conserved during replication.

• In **dispersive replication**, both nucleotide strands break down (disperse) into fragments, which serve as templates for the synthesis of new DNA fragments, and then somehow reassemble into two complete DNA molecules. In this model, each resulting DNA molecule is interspersed with fragments of old and new DNA; none of the original molecule is conserved.

• **Semiconservative replication** is intermediate between these two models; the two nucleotide strands unwind and each serves as a template for a new DNA molecule. *It is most experimentally proven.*

Prokaryotes have a single origin of replication; eukaryotes have many. Replication in both cases proceeds in both directions from an origin of replication. The DNA fragment from the point of replication start to the point of replication end forms replication unit – a **replicon**. DNA replication has 3 stages: initiation, elongation, and termination (Figure 15).

1. **Initiation**: DNA *helicase* unwinds the double helix, and the template strands are stabilized by single-strand binding proteins. An *RNA primase* catalyzes the synthesis a short RNA primer, to which nucleotides are added.

2. **Elongation**: Because the two nucleotide strands of DNA are antiparallel, replication takes place continuously on one strand (the leading strand) and discontinuously on the other (the lagging strand).Through the action of *DNA polymerase*, the leading strand grows continuously in the 5′-to-3′ direction until the replication of that section of DNA has been completed. On the lagging strand, DNA is still made in the 5′-to-3′ direction. But synthesis of the lagging strand is discontinuous: The DNA is added as short fragments (*Okasaki fragments*).

3. **Termination**: RNA primer is degraded and DNA is added in its place.

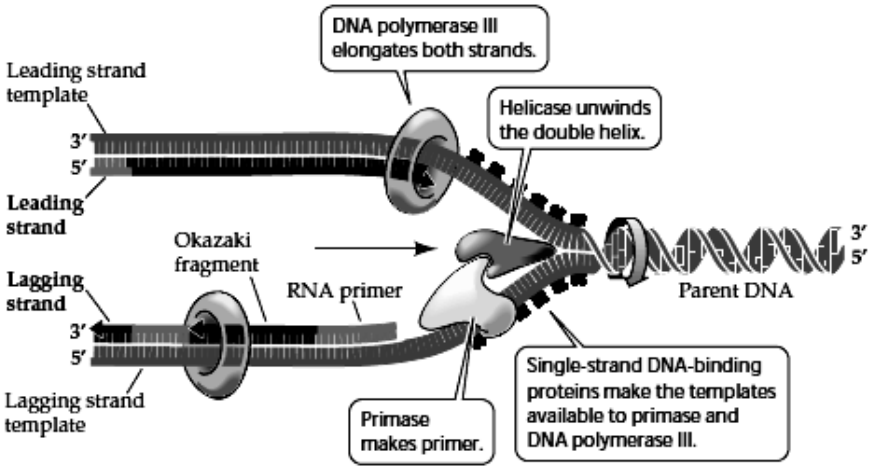

Figure 15 — Scheme of replication in eukariotes

The replication of DNA is not perfectly accurate, and the DNA of nondividing cells is subject to damage by environmental agents. DNA polymerases initially make a significant number of mistakes in assembling polynucleotide strands. The observed error rate of one for every 10^6 bases replicated would result in about 1,000 mutations every time a human cell divided. Our cells have at least three DNA repair mechanisms:

1. A *proofreading* mechanism corrects errors in replication as DNA polymerase makes them.

2. A *mismatch repair* mechanism scans DNA immediately after it has been replicated and corrects any base-pairing mismatches.

3. An *excision repair* mechanism removes abnormal bases that have formed because of chemical damage and replaces them with functional bases.

Transcription

Three distinct processes—initiation, elongation, and termination—constitute DNA transcription (figure 16).

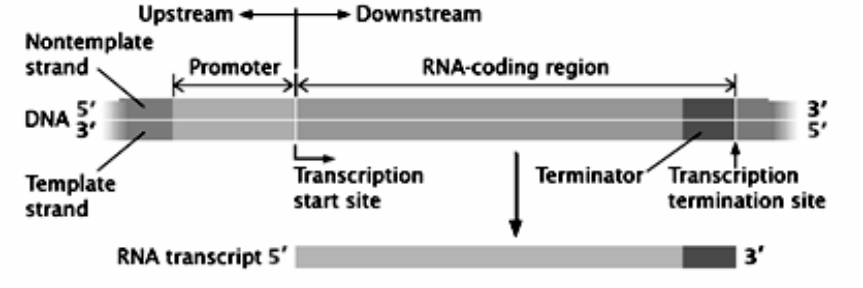

Figure 16 — Scheme of transcription in eukariotes

Initiation begins transcription, and requires a **promoter**, a special sequence of DNA to which *RNA polymerase* binds very tightly. A promoter, which is a specific sequence in the DNA that reads in a particular direction, orients the RNA polymerase and thus "aims" it at the correct strand to use as a template.

Once RNA polymerase has bound to the promoter, it begins the process of **elongation**. It unwinds the DNA about 20 base pairs at a time and reads the template strand in the 3′-to-5′ direction. Like DNA polymerase, RNA polymerase adds new nucleotides to the 3′ end of the growing strand, but does not require a primer to get started.

Just as initiation sites specify the start of transcription, particular base sequences in the DNA specify its **termination**. The mechanisms of termination are complex and of more than one kind. For some genes, the newly formed transcript simply falls away from the DNA template and the RNA polymerase. For others, a helper protein pulls the transcript away.

The question is: How do transcription and translation produce specific and functional protein products? These processes require a **genetic code** that relates genes (DNA) to mRNA and mRNA to the amino acids of proteins. The genetic code specifies which amino acids will be used to build a protein.

The basic postulates of the code are:

1. Genetic code has a triplet structure. The triplet of mRNA is called **codon**.
2. One amino acid corresponds, as a rule, more than one codon. In codon first two nucleotides are identical to one amino acid more often, and the third varies.
3. The nucleotide sequence is recognized in one direction, triplet by triplet.
4. AUG represents starting codon.
5. UAG, UAA, UGA are stop codons.
6. The genetic code is universal for all organisms.

The genetic code consists of 64 codons and the amino acids specified by these codons. The complete genetic code is shown in figure 17.

The last step of information flow is **translation**, the RNA-directed assembly of a protein. Like transcription, translation occurs in three steps: initiation, elongation, and termination (figure 18).

Initiation. The translation of mRNA begins with the formation of an **initiation complex**, which consists of the methionine-charged tRNA and small ribosomal subunit, both bound to the mRNA. The rRNA of the small ribosomal subunit binds to a complementary ribosome recognition sequence on the mRNA. This sequence is «upstream» (toward the 5′ end) of the actual start codon (AUG) that begins translation. After has bound to the mRNA, the large subunit of the ribosome joins the complex.

First base	Second base: U	Second base: C	Second base: A	Second base: G	Third base
U	UUU Phe	UCU Ser	UAU Tyr	UGU Cys	U
	UUC Phe	UCC Ser	UAC Tyr	UGC Cys	C
	UUA Leu	UCA Ser	UAA Stop	UGA Stop	A
	UUG Leu	UCG Ser	UAG Stop	UGG Trp	G
C	CUU Leu	CCU Pro	CAU His	CGU Arg	U
	CUC Leu	CCC Pro	CAC His	CGC Arg	C
	CUA Leu	CCA Pro	CAA Gln	CGA Arg	A
	CUG Leu	CCG Pro	CAG Gln	CGG Arg	G
A	AUU Ile	ACU Thr	AAU Asn	AGU Ser	U
	AUC Ile	ACC Thr	AAC Asn	AGC Ser	C
	AUA Ile	ACA Thr	AAA Lys	AGA Arg	A
	AUG Met	ACG Thr	AAG Lys	AGG Arg	G
G	GUU Val	GCU Ala	GAU Asp	GGU Gly	U
	GUC Val	GCC Ala	GAC Asp	GGC Gly	C
	GUA Val	GCA Ala	GAA Glu	GGA Gly	A
	GUG Val	GCG Ala	GAG Glu	GGG Gly	G

Figure 17 — Genetic information is encoded in mRNA in three — letter units — codons — made up of the bases uracil (U), cytosine (C), adenine (A), and guanine (G). To decode a codon, find its first letter in the left column, then read across the top to its second letter, then read down the right column to its third letter. The amino acid the codon specifies is given in the corresponding row. For example, AUG codes for methionine, and GUA codes for valine

Elongation. Consist of repeated steps on ribosome in which amino acids that bring by tRNA form a peptide bond with the amino acid chain.

Termination. The elongation cycle ends, and translation is terminated, when a stop codon—UAA, UAG, or UGA—enters in the site of ribosome.

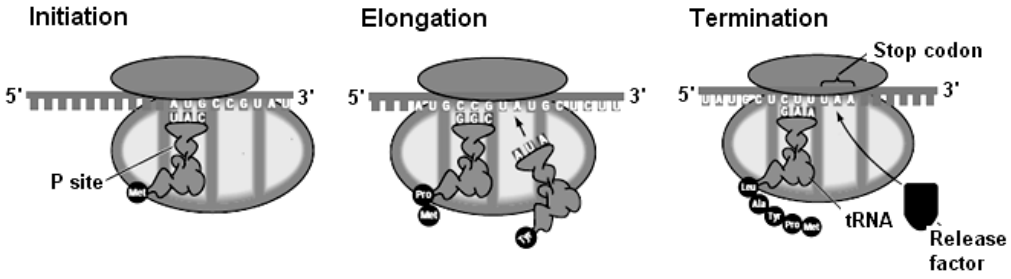

Figure 18 — Scheme of translation in eukariotes

Comprehension questions

1. What are the major transfers of genetic information?
2. What is central dogma of molecular biology?
3. Who formalized the concept of information flow?
4. List and describe the possible types of replication.
5. Outline the process of replication.
6. Describe three DNA repair mechanisms.
7. Outline the process of transcription.
8. Name the basic postulates of the genetic code.
9. Outline the process of translation.

Definitions of the terms

Codon is each sequence of three nucleotide bases along the RNA chain that specifies a particular amino acid.

Genetic code is the means of relating codons to their specific amino acids.

Promoter is a special sequence of DNA to which RNA polymerase binds very tightly.

Replication is a self-duplication of DNA molecule.

Replicon is the DNA fragment from the point of replication start to the point of replication end.

Transcription is information transfer from DNA to RNA.

Translation is the RNA-directed assembly of a protein.

Topic 7. Basic principles of heredity

Johann Gregor Mendel first discovered the principles of heredity by crossing different varieties of pea plants (*Pisum sativum*) and analyzing the pattern of transmission of traits in subsequent generations. He examined seven characteristics that appeared in the seeds and in plants grown from the seeds such as color of seed and seed coats, shape of seeds and pods and so on.

Mendel's approach to the study of heredity was effective for several reasons:

1. Selection of parent pairs (pure lines) for crossing.

2. In each generation it is necessary to keep count of organisms at each pair of alternative characteristics, without taking into account other differences between crossed organisms.

3. Use the quantitative analysis of hybrid organisms.

4. Individual analysis of the offspring from each hybrid organism.

Before examination of Mendel's crosses and the conclusions that he made from them, it will be helpful to review some terms commonly used in genetics. In the context of genetic crosses, a **gene** can be defined as an inherited factor that determines a characteristic. More specifically, a **gene** is a portion of the DNA that resides at a particular site on a chromosome and encodes a particular character. Genes frequently come in different versions called **alleles** (figure 19).

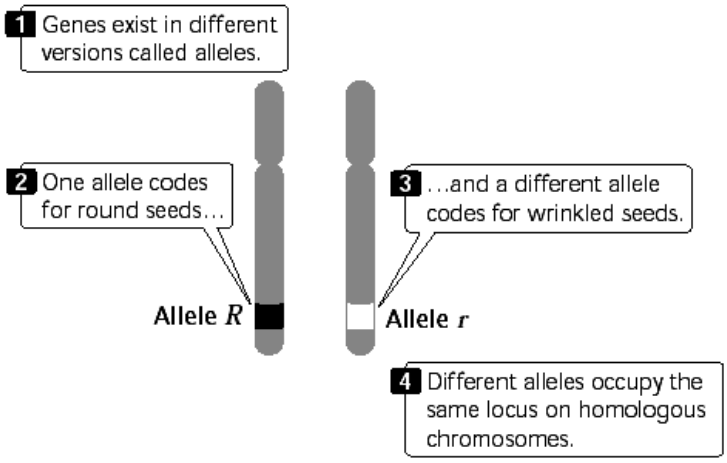

Figure 19 — At each locus, a diploid organism possesses two alleles located on different homologous chromosomes

In Mendel's crosses, seed shape was determined by a gene that exists as two different alleles: one allele codes for round seeds and the other codes for wrinkled seeds. All alleles for any particular gene will be found at a specific place on a chromosome called the **locus** for that gene. Thus, there is a specific place — a locus — on a chromosome in pea plants where the shape of seeds is determined. This locus might be occupied by an allele for round seeds or one for wrinkled seeds. The **genotype** is the set of alleles that an individual organism possesses. A diploid organism that possesses two identical alleles is **homozygous** for that locus. One that possesses two different alleles is **heterozygous** for the locus. **Phenotype** is the manifestation or appearance of a characteristic.

Mendel began by studying **monohybrid crosses** — those between parents that differed in a single characteristic. In one experiment, Mendel crossed a pea plant homozygous for round seeds with one that was homozygous for wrinkled. This first generation of a cross is the **P** (parental) **generation**. The offspring from the parents in the P generation are the **F_1** (first filial) **generation**. When Mendel examined the F_1 of this cross, he found that they expressed only one of the phenotypes present in the parental generation: all the F_1 seeds were round. Those traits that appeared unchanged in the F_1 heterozygous offspring Mendel called **dominant,** and those traits that disappeared in the F_1 heterozygous offspring he called **recessive.** When dominant and recessive alleles are present together, the recessive allele is masked, or suppressed. The concept of dominance was an important conclusion that Mendel derived from his monohybrid crosses (Mendel's first law). The **concept of dominance** states that, when two different alleles are present in a genotype, only the trait of the dominant allele is observed in the phenotype.

The scheme of crossing.
P: ♀AA x ♂aa
G: (A) (a)
F_1: Aa

For the next crossing, Mendel allowed the plants of F_1 generation to self-fertilize, to produce a second generation (the **F_2 generation**). Both of the traits from the P generation emerged in the F_2; Mendel counted round seeds and wrinkled seeds in the F_2. He noticed that the number of the round and wrinkled seeds constituted approximately a 3 to 1 ratio. Mendel conducted monohybrid crosses for all seven of the characteristics that he studied in pea plants, and in all of the crosses he obtained the same result. Mendel's conclusion was that the two alleles of an individual plant separate with equal probability into the gametes. The gametes then paired randomly to produce the following genotypes in equal proportions among the F_2: AA, Aa, Aa, aa. Because round (A) is dominant over wrinkled (a), there were three round progeny in the F_2 (AA, Aa, Aa) for every one wrinkled progeny (aa) in the F_2. The conclusions that Mendel developed have been further developed and formalized into the principle of segregation (Mendel's second law). The **principle of segregation** states that alleles of each individual diploid organism segregate (separate) when gametes are formed, and one allele goes into each gamete. Furthermore, the two alleles segregate into gametes in equal proportions.

The scheme of crossing.
P: ♀ Aa x ♂ Aa
G: (A) (a) (A) (a)
F_1: AA, Aa, Aa, aa

In 1900, when Mendel's work was rediscovered and biologists began to apply his principles of heredity, the relation between genes and chromosomes was still unclear. The theory that genes are located on chromosomes (the **chromosome theory of heredity**) was developed in the early 1900s by Walter Sutton. The chromosome theory of inheritance states that genes are located on chromosomes. The two alleles of a genotype segregate during anaphase I of meiosis, when homologous chromosomes separate. The alleles may also segregate during anaphase II of meiosis if crossing over has taken place in prophase I.

To predict the types of offspring that result from crossing can be used a shorthand method well known as a **Punnett square**. A Punnett square is constructed by drawing a grid, putting the gametes produced by one parent along the upper edge and the gametes produced by the other parent down the left side. Each cell (a block within the Punnett square) contains an allele from each of the corresponding gametes, generating the genotype of the progeny produced by fusion of those gametes (table 3).

Table 3 — The Punnett square can be used for determining the results of a genetic cross. Here, the alleles of parent's gametes and genotypes of progeny from previous scheme of crossing are illustrated

	A	a
A	AA	Aa
a	Aa	aa

In addition to his work on monohybrid crosses, Mendel also crossed varieties of peas that differed in *two* characteristics **(dihybrid crosses).** For example, he had one homozygous variety of pea that produced round seeds and yellow endosperm; another homozygous variety produced wrinkled seeds and green endosperm. When he crossed the two, all the F_1 progeny had round seeds and yellow endosperm. He then self-fertilized the F_1 and obtained the following progeny in the F_2: 9/16 round, yellow seeds; 3/16 wrinkled, yellow seeds; 3/16 round, green seeds; and 1/16 wrinkled, green seeds or in a 9:3:3:1 ratio. The conclusions that Mendel developed have been further developed and formalized into the **principle of independent assortment** (Mendel's third law). This principle states that alleles at different loci separate independently of one another.

The scheme of crossing.
P: ♀ AABB x ♂ aabb
G: (AB) (ab)
F_1: AaBb

self-fertilization of F_1:
F_1: ♀ AaBb x ♂ AaBb
G: (AB) (Ab) (aB) (ab)
F_2: 9A_B_ : 3A_bb : 3aaB_ : 1aabb,

where A_ and B_ are phenotypic radicals. **Phenotypic radical** is a dominant gene of organism, which determines it phenotype.

An important qualification of the principle of independent assortment is that it applies to characters encoded by loci located on different chromosomes because, like the principle of segregation, it is based wholly on the behavior of chromosomes during meiosis. Each pair of homologous chromosomes separates independently of all other pairs in anaphase I of meiosis; so genes located on different pairs of homologs will assort independently. Genes that happen to be located on the same chromosome will travel together during anaphase I of meiosis and will arrive at the same gamete.

More than 2000 hereditary diseases and anomalies of development which inheritances submit to Mendel's laws are known in human (table 4).

Table 4 — Examples of hereditary diseases and anomalies of development of human which inheritances submit to Mendel's laws

Dominant	Recessive
chondrodystrophia	normal development of skeleton
polydactylia (extra digits)	norm No of digits
brachydactylia	norm
normal fibrillation	hemophilia
normal color sense	daltonism
normal pigmentation	albinism (absence of skin pigment)
normal digestion of phenylalanine	phenylketonuria

Comprehension questions

1. Why was Mendel's approach to the study of heredity so successful?
2. What is the relation between the terms allele, locus, gene, and genotype?
3. What is the principle of segregation?
4. What is the concept of dominance?
5. What is the principle of segregation?
6. Give the phenotypic ratios that may appear among the progeny of simple crosses and the genotypes of the parents that may give rise to each ratio.
7. Give the genotypic ratios that may appear among the progeny of simple crosses and the genotypes of the parents that may give rise to each ratio.
8. What is the chromosome theory of inheritance?
9. What is the principle of independent assortment?
10. What is the phenotypic radical?
11. How is the principle of independent assortment related to meiosis?
12. Give examples of hereditary diseases and anomalies of development of human which inheritances submit to Mendel's laws.

Give solution of tasks

1. What gamete types give organisms with following genotypes: *BB, Dd, aabb, AaDD, Bbcc, BbCc*?

2. Short hair in rabbits (*S*) is dominant over long hair (*s*). The following crosses are carried out, producing the progeny shown. Give all possible genotypes of the parents in each cross.

Parents	**Progeny**
(a) short x short	4 short and 2 long
(b) short x short	8 short
(c) short x long	12 short
(d) short x long	3 short and 1 long
(e) long x long	2 long

Solution

For this problem, it is useful to first gather as much information about the genotypes of the parents as possible on the basis of their phenotypes. We can then look at the types of progeny produced to provide the missing information. Notice that the problem asks for *all* possible genotypes of the parents. After this we needs in correct writing of solution.

(a) short x short 4 short and 2 long

Because short hair is dominant over long hair, a rabbit having short hair could be either *SS* or *Ss*. The two long-haired offspring must be homozygous (*ss*) because long hair is recessive and will appear in the phenotype only when both alleles for long hair are present. Because each parent contributes one of the two alleles found in the progeny, each parent must be carrying the *s* allele and must therefore be *Ss*.

Writing of solution:

1. All data on phenotypes and genotypes must be included in the table:

Attribute	Gene	Genotype
Short hair	*S*	*SS, Ss*
Long hair	*s*	*ss*

2. Writing scheme of crossing:

P: ♀ Ss x ♂ Ss

G: 

F_1: SS, Ss, Ss, ss

Give solution for other variants yourself.

3. In watermelons, bitter fruit (*B*) is dominant over sweet fruit (*b*), and yellow spots (*S*) are dominant over no spots (*s*). The genes for these two characteristics assort independently. A homozygous plant that has bitter fruit and yellow spots is crossed with a homozygous plant that has sweet fruit and no spots. The F1 are intercrossed to produce the F2.

(a) What will be the phenotypic ratios in the F2?

(b) If an F1 plant is backcrossed with the bitter, yellow spotted parent, what phenotypes and proportions are expected in the offspring?

(c) If an F1 plant is backcrossed with the sweet, nonspotted parent, what phenotypes and proportions are expected in the offspring?

Definitions of the terms

Alleles are the different versions of gene.

Dominant traits are those traits that appeared unchanged in the F_1 heterozygous offspring.

Gene is a portion of the DNA that resides at a particular site on a chromosome and encodes a particular character.

Genotype is the set of alleles that an individual organism possesses.

Homozygote is a diploid organism that possesses two identical alleles for any particular locus.

Heterozygote is a diploid organism that possesses two different alleles for any particular locus.

Locus is the specific place for any particular gene on a chromosome.

Phenotype is the manifestation or appearance of a characteristic.

Phenotypic radical is a dominant gene of organism, which determines it phenotype.

Recessive traits are those traits that disappeared in the F_1 heterozygous offspring.

Topic 8. Allele and gene interaction. Interactions of genotype and environment. Gene linkage

In many cases, alleles do not show the simple relationships between dominance and recessiveness. In others, a single allele may have multiple phenotypic effects. Existing alleles can give rise to new alleles by mutation, so there can be many alleles for a single character.

Types of allele interaction (table 5).

- Dominance or complete dominance.
- Incomplete dominance.
- Codominance.

Table 5 — Types of allele interaction and their definition

Type of Dominance	Definition
Dominance	Phenotype of the heterozygote is the same as the phenotype of one of the homozygotes.
Incomplete dominance	Phenotype of the heterozygote is intermediate (falls within the range) between the phenotypes of the two homozygotes.
Codominance	Phenotype of the heterozygote includes the phenotypes of both homozygotes.

Example of incomplete dominance, if a true-breeding red snapdragon is crossed with a true-breeding white one, all the F1 flowers are pink. Example of incomplete dominance of human can be brachidactily (short digits).

Example of codominance is the IV blood group in human (I^AI^B).

For some loci, more than two alleles are present within a group of individuals—the locus has **multiple alleles**. (Multiple alleles may also be referred to as an **allelic series**.) Although there may be more than two alleles present within a group, the genotype of each diploid individual still consists of only two alleles. The inheritance of characteristics encoded by multiple alleles is no different from the inheritance of characteristics encoded by two alleles, except that a greater variety of genotypes and phenotypes are possible.

In human an allelic series I^0, I^A, I^B which determines polymorphism of blood groups is known.

Frequently, the effects of genes at one locus depend on the presence of genes at other loci. This type of interaction between the effects of genes at different loci (genes that are not allelic) is termed gene interaction.

Types of gene interactions

- Epistasis.
- Complementation.
- Polymery.

Sometimes the effect of gene interaction is that one gene masks (hides) the effect of another gene at a different locus, a phenomenon known as **epistasis**.

In epistasis, the gene that does the masking is called the **epistatic gene**; the gene whose effect is masked is a **hypostatic gene**. Epistatic genes may be recessive (recessive epistasis) or dominant (dominant epistasis) in their effects.

Complementary, or supplementing genes be named dominant genes which at a combined presence (in genotype A-B-) cause development of a new attribute in comparison with separate action of each gene (A-вв or aaB-). The normal hearing is inherited in human on the base on a principle of complementation.

Organism traits like rate of growth, weight of body, length of body, arterial pressure, and degree of pigmentation is impossible to decompose on phenotype classes.

These traits has name *quantitative*. Each of such traits is formed usually under influence of several equivalent genes. The phenomenon of trait formation as a result of action of several genes refers to — **polymery** (-ia), and genes has name polymeric.

Interactions of genotype and environment

The phenotype of an individual does not result from its genotype alone. Genotype and environment interact to determine the phenotype of an organism. Environmental variables such as light, temperature, and nutrition can affect the translation of a genotype into a phenotype.

Incomplete penetrance and variable expressivity result from the influence of other genes and environmental factors on the phenotype.

Expressivity is the degree to which a trait is expressed. Examples are a brachydactyly and polydactyly in a different degree of their expressivity.

Penetrance is the phenomenon under which the trait encoded by same gene, is expressed in one and it is not expressed in other individuals of related group. Penetrance is defined as the percentage of individuals having a particular genotype that express the expected phenotype. For example, if we examined 42 people having an allele of polydactyly and found that only 38 of them were polydactylous, the penetrance would be 38/42 = 0.90 (90 %).

Full penetration — 100 %, the gene is expressed in each individual.

Gene linkage

The number of an organism traits and properties that encoded by genes is large but the number of chromosomes in each species is relatively not many and constant. Consequently, there is not one gene in each chromosome but it amount is a large. The joint inheritance of genes that limiting their free combination was named **linkage of genes** or **linked inheritance**. Linked genes are often inherited together. Linked genes can recombine by crossing over in prophase I of meiosis. The result is recombinant gametes, which have new combinations of linked genes because of the exchange. Recombination frequency is calculated by summing the number of recombinant progeny, dividing by the total number of progeny produced in the cross, and multiplying by 100 %. Recombination rates can be used to determine the relative order of genes and distances between them on a chromosome. Maps based on recombination rates are called genetic maps; maps based on physical distances are called physical maps. One percent recombination equals one **map unit**, which is also a **centiMorgan**.

Comprehension questions

1. List the different types of allele interaction.
2. Define term dominance.
3. Define term codominance and give examples of codominant inheritance.
4. Define term incomplete dominance and give examples of incomplete dominant inheritance.
5. What is multiple alleles? Give example of traits encoded by multiple alleles.
6. List the different types of gene interaction.

7. Define term epistasis.
8. What gene named epistatic?
9. What gene named hypostatic?
10. What gene named complementary or supplementing genes?
11. What traits has name quantitative?
12. What genes has name polymeric?
13. What is expressivity and penetrance and what causes it?
14. What is linkage of genes?
15. How do calculation of recombination frequencies?
16. What is the difference between a genetic map and a physical map?
17. What is the percent recombination equals?

Definitions of the terms

Complementary genes are dominant genes which at a combined presence cause development of a new attribute in comparison with separate action of each gene.

Codominance is a type of interaction between alleles in which the phenotype of the heterozygote simultaneously expresses the phenotypes of both homozygotes.

Dominance is a type of interaction between alleles in which the heterozygote possesses the same phenotype as one of the homozygotes.

Epistasis is the effect of gene interaction under which one gene masks (hides) the effect of another gene at a different locus.

Epistatic gene is the gene that does the masking of another gene.

Expressivity is the degree to which a trait is expressed.

Hypostatic gene is the gene whose effect is masked by another gene.

Incomplete dominance is a type of interaction between alleles in which the heterozygote is intermediate in phenotype between the two homozygotes.

Linkage of genes is the joint inheritance of genes that limiting their free combination.

Multiple alleles are the presence at the locus more than two alleles within a group of individuals.

Penetrance is the phenomenon under which the trait encoded by same gene, is expressed in one and it is not expressed in other individuals of related group.

Polymery is the phenomenon of trait formation as a result of action of several genes.

Topic 9. Diversity

Diversity is the ability of organism to change their traits, getting new ones or loosing old ones.

Diversity can be divided on:

(a) Phenotypic:	(b) Genotypic:
1. Ontogenetic	1. Combinative
2. Modificational	2. Mutational

(a) **Phenotypic diversity** shows phenotype changes under environmental conditions, which not affect on genotype, but level of it expression is determined

by genotype. The range of phenotypes produced by a genotype in different environments is called the **norm of reaction**.

1. **Modificational diversity** describes the individual's changes caused by factors of an environment. Modifications can be seasonal and ecological.

2. **Ontogenetic diversity** is a continuous change of traits in ontogenetic development of an individual.

(b) The diversity, which involves changes in genotype due to mutations or gene recombination, is called **genotypic diversity**.

1. The **combinative diversity** is the formation of new allele combinations due to:

- Independent assortment of chromosomes in meiosis.
- Crossing over between homologous chromosomes in meiosis.
- Random combination of gametes at fertilization. Thus, a mating system has influence on combinative diversity of human. It may be:
  - Crosses between related individuals – *inbreeding*.
  - Crosses between unrelated individuals – *outbreeding*.

2. The diversity with rapid, strong changes of traits is called **mutational**.

Mutations are permanent, heritable changes of genetic information. **Gene mutations** affect only the genetic information of a single gene; **chromosome and genome mutations** alter the structure and the number of chromosomes and therefore usually affect many genes.

On the one hand, mutation is the source of all genetic variation, the raw material of evolution. Without mutations and the variation that they generate, organisms could not adapt to changing environments and would risk extinction. On the other hand, most mutations have detrimental effects, and mutation is the source of many human diseases and disorders. All mutations are classified on certain types (table 6).

Generative mutations are those that occur in the cells of the *germ line*— the specialized cells that give rise to gametes. A gamete with the mutation passes it on to a new organism at fertilization.

Somatic mutations are those that occur in somatic (body) cells. These mutations are passed on to the daughter cells after mitosis, and to the offspring of those cells in turn, but are not passed on to sexually produced offspring.

Table 6 — Classification of mutations

Classifying factor	Mutations' names
According to mutated cells	1. Generative 2. Somatic
According to genotype change	1. Gene (point) mutations 2. Chromosomal mutations 3. Genome mutations
According to adaptive significance	1. Useful 2. Harmful 3. Neutral
According to reason of mutation	1. Spontaneous 2. Induced

Point mutations are mutations of single base pairs and so are limited to single genes: One allele becomes another allele because of an alteration of a single nucleotide (which, after DNA replication, becomes a mutant base pair).

In general all point mutation have one of the following mechanisms (figure 20):

1. **Substitution** of base pair in DNA molecule.
2. **Deletion** (loss) of nucleotide pair (or group of the bases) in DNA molecule.
3. **Insertion** of nucleotide pair (or group of pairs the bases) in molecule DNA.

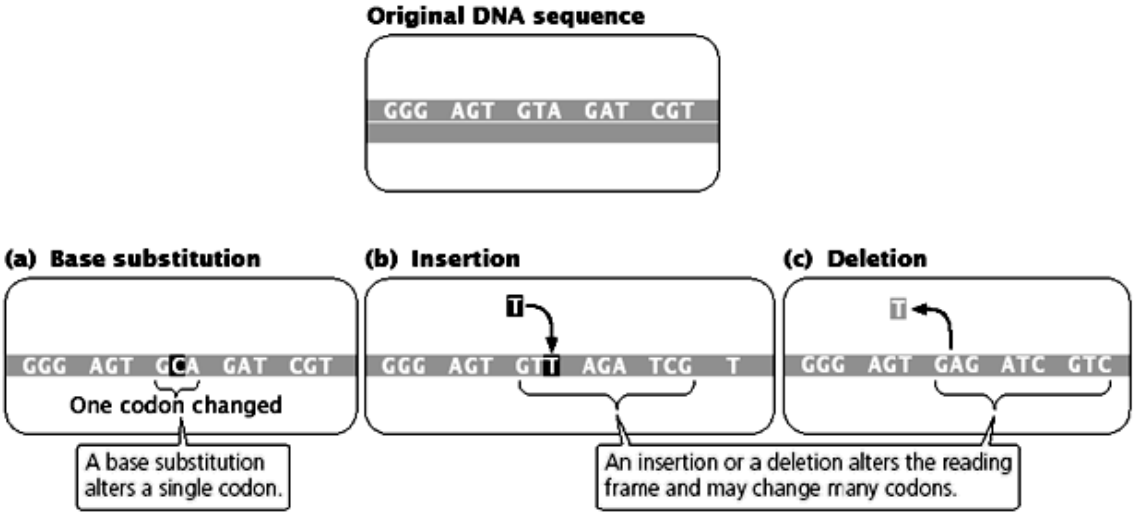

Figure 20 — Three basic types of gene mutations are base substitutions (a), insertions (b), and deletions (c)

Chromosomal mutations are more extensive alterations than point mutations. They may change the position or orientation of a DNA segment without actually removing any genetic information, or they may cause a segment of DNA to be lost.

Distinguish the following chromosome aberrations:

(a) Intrachromosomal:

1. **Deletion (**loss) of a chromosome segment.
2. **Duplication** is a mutation in which part of the chromosome has been doubled.
3. **Inversion** of a chromosome segment on 180 degrees.

(b) Interchromosomal:

1. **Translocation** entails the movement of genetic material between non-homologous chromosomes.

Genome mutations can be classified into two basic types: changes in the number of individual chromosomes (*aneuploidy*) and changes in the number of chromosome sets (*polyploidy*).

Four types of relatively common aneuploid conditions is well-known in diploid individuals: nullisomy (2n-2), monosomy (2n-1), trisomy (2n+1), and tetrasomy (2n+2).

Polyploids include triploids (3n) tetraploids (4n), pentaploids (5n), and even higher numbers of chromosome sets.

Spontaneous mutations are permanent changes in the genome that occur without any outside influence.

Induced mutations occur when some agent outside the cell—**a mutagen**—causes a permanent change in DNA. Ionizing radiation is the most important mutagen. Ionizing radiation may be electromagnetic or wavelike (gamma-rays, X-rays, cosmic rays and etc.) and corpuscular (electrons, positrons, protons, and etc.).

Comprehension questions

1. What is diversity?
2. Name the kinds of diversity.
3. What is phenotypic diversity? Name the kinds of phenotypic diversity and define each one.
4. What is norm of reaction?
5. What is genotypic diversity? Name the kinds of genotypic diversity and define each one.
6. What is mutation?
7. What differences between effect of gene mutations and chromosome or genome mutations on the genetic information?
8. What classifying factors of mutation you know?
9. Give classification of mutation according the classifying factors.
10. What is a somatic and generative mutation?
11. List the different types of point mutations.
12. List the different types of chromosome mutations.
13. List the different types of genome mutations.
14. What is spontaneous and induced mutation?

Definitions of the terms

Chromosomal mutations are changes of the position, orientation, or lost of a DNA segment.

Combinative diversity is the formation of new allele combinations.

Diversity is the ability of organism to change their traits, getting new ones or loosing old ones.

Generative mutations are those that occur in the cells of the germ line — the specialized cells that give rise to gametes.

Genome mutations are changes in the number of individual chromosomes or changes in the number of chromosome sets.

Genotypic diversity is a changes in genotype caused by mutations or gene recombination.

Induced mutations are permanent changes in the genome that occur with influence of a mutagen.

Modificational diversity is an individual's changes caused by factors of an environment.

Mutagen is some agent outside the cell.

Mutational diversity is the diversity with rapid, strong changes of traits.

Mutations are permanent, heritable changes of genetic information.

Norm of reaction is the range of phenotypes produced by a genotype in different environments.

Ontogenetic diversity is a continuous change of traits in ontogenetic development of an individual.

Phenotypic diversity is a phenotype changes under environmental conditions, which not affect on genotype, but level of it expression is determined by genotype.

Point mutations are mutations of single base pairs.

Somatic mutations are those that occur in somatic (body) cells.

Spontaneous mutations are permanent changes in the genome that occur without any outside influence.

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