Lecture complex

Discipline: "Molecular Biology and medical genetics"

Discipline code: MBMG 1203

Name and code: Medicine "6B10115"

Amount of study hours/credits: 180 hours/6 credit

Course and semester of study: 1-I

Lecture volume:12

Shymkent 2025

The lecture complex was developed in accordance with the syllabus "Molecular Biology and Medical Genetics" and discussed at the meeting of the department.

Head of the Department, Acting Professor _____ Daurenbekov K.N.

Protocol no. 1 from "27" 08 2025 y.

Molecular Biology

Lecture No. 1

1. Topic: Introduction to molecular biology. Structure and functions of proteins and nucleic acids. Ways of transmitting genetic information.

2. Purpose: To give an idea: 1) about the subject, tasks and significance of medical biology and genetics; 2) structure and functions of information molecules: proteins and NK; 3) about the types of transfer of hereditary information.

3. Lecture abstracts: Molecular biology is a branch of biology that studies the structure and interaction of molecules and macromolecular systems involved in the biological processes of living organisms, the molecular foundations of heredity and protein synthesis, the mechanisms of storage, transmission and realization of genetic information, the structure and functions of complex high-molecular compounds that make up the cell: irregular biopolymers (proteins and nucleic acids acids).

Genetics (from the Greek.γενητως — originating from someone) is the science of the laws of heredity and variability. Depending on the object of research, the genetics of plants, animals, microorganisms, humans, and others are classified; depending on the methods used in other disciplines, molecular genetics and others. The ideas and methods of genetics play an important role in medicine, agriculture, the microbiological industry, as well as in genetic engineering.

Proteins (proteins, polypeptides) are high—molecular organic substances consisting of alpha-amino acids connected in a chain by a peptide bond. In living organisms, the amino acid composition of proteins is determined by the genetic code; in most cases, 20 standard amino acids are used in synthesis. Many combinations of them give a wide variety of properties of protein molecules. In addition, amino acids in a protein often undergo posttranslational modifications, which can occur both before the protein begins to perform its function and during its "work" in the cell. Often in living organisms, several protein molecules form complex complexes, for example, a photosynthetic complex.

Highly purified proteins form crystals at low temperature, which are used to produce a model of this protein.

The functions of proteins in the cells of living organisms are more diverse than those of other biopolymers, such as polysaccharides and DNA. Thus, enzyme proteins catalyze the course of biochemical reactions and play an important role in metabolism. Some proteins perform a structural or mechanical function, forming a cytoskeleton that supports the shape of cells.

Types of transfer of genetic information. There are 3 types of information transfer processes: general transfer, specialized transfer, and prohibited transfer.

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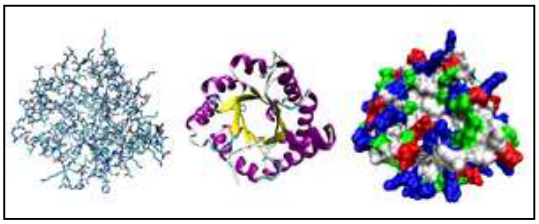
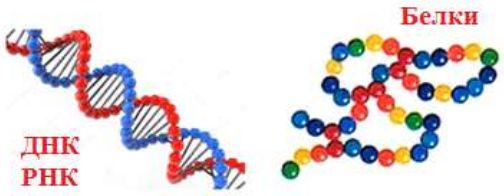
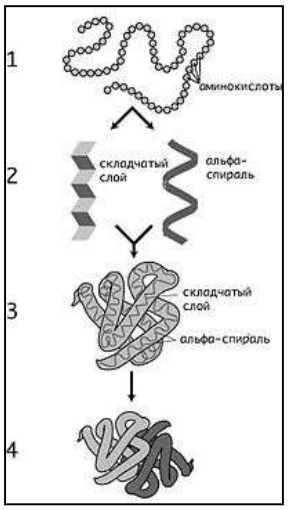
Department of Chemical Disciplines, Biology and Biochemistry | 46/ 1 p out of 12
Lecture complex

4) Illustrative material:





Different ways of depicting the three-dimensional structure of a protein using the example of the enzyme triose phosphatizomerase.



Levels of protein structure: 1 — primary, 2 — secondary, 3 — tertiary, 4 — quaternary

5.References: see appendix 1

6. Control questions: (feedback)

1. The subject of the study of molecular biology.

2. Structure and functions of proteins..

3. Structure and functions of DNA and RNA. Types of RNA and forms of DNA.

4. Primary, secondary and tertiary structures of DNA and RNA.

5. Types of transfer of genetic information.

6. The nucleosome thread.

7. Supranucleosomal DNA folding.

8. Physico-chemical properties and functions of DNA.

Lecture No. 2

1. The topic is matrix synthesis of nucleic acids. Mechanisms of replication, transcription, and translation.

2**. Purpose:** to provide an idea of the principles of recording genetic information and its further implementation.

3. Lecture abstracts: DNA replication is the process of synthesizing a daughter DNA molecule that occurs during the synthetic (S) phase of the cell's life cycle on the matrix of the

parent DNA molecule. In this case, the genetic material encoded in DNA is doubled and, in the process of subsequent division, is divided between daughter cells. DNA replication is carried out by a complex enzyme complex consisting of 15-20 different proteins.

DNA replication is carried out semi-conservatively. Replication begins with the separation of a double helix at a certain point (the ori locus or origin) and the formation of single-stranded DNA regions that serve as a template for the synthesis of new strands. The region of DNA in which replication begins and ends in eukaryotes is called a replicon. The DNA of prokaryotes doubles entirely in one replication cycle, that is, the bacterial chromosome and plasmids are one replicon. In eukaryotes, the DNA length is one million pairs of nucleotides (in humans, about 150 million pairs of nucleotides). Replication of such molecules, at a replication rate of 50 thousand bp per minute in E.coli, is 800 hours. Therefore, DNA replication occurs simultaneously at several sites (a site is any part of DNA), therefore, eukaryotic DNA has many replicons.

The following basic principles apply to all replication methods:

1. Synthesis of daughter DNA is a matrix process; the parent DNA chains are the matrix.

2. Replication is based on the principle of complementarity: the nucleotides of the daughter DNA are complementary to the nucleotides of the parent DNA matrix.

3. The transfer process is symmetrical, with both DNA strands serving as templates.

Replication factors are proteins: topoisomerases, SSB protein, helicase, DNA polymerase.

Transcription is the process of synthesizing RNA using DNA as a template that occurs in all living cells. In other words, it is the transfer of genetic information from DNA to RNA.

Transcription is catalyzed by the enzyme DNA-dependent RNA polymerase. The process of RNA synthesis proceeds in the direction from the 5' to the 3' end.

Transcription consists of the stages of initiation, elongation, and termination.

mRNA translation is the process of transferring information from a sequence of mRNA nucleotides to a specific sequence of amino acids of the corresponding protein. In the process of such information transfer, amino acids are incorporated (polymerized) into growing peptide chains in accordance with the sequence of mRNA codons, in other words, a molecular peptide is synthesized on the mRNA matrix. The following people take part in the broadcast process::

1. mRNA synthesized in the nucleus during transcription, matured and transported in combination with special proteins into the cytoplasm;

2. tRNA (several dozen species) synthesized in the nucleus, matured and modified, and transported to the cytoplasm;

3. 4 types of rRNA synthesized in the nucleus and, in the same place, forming ribosome subunits in complex with ribosomal proteins. These subunits enter the cytoplasm through the pores of the nuclear membrane.

4. 20 types of amino acids found in the cytoplasm, synthesized from carbohydrates in it, which came from outside with food or from proteins of their own tissues. The main source of amino acids used in protein synthesis are food proteins. The total weight of free amino acids in the body is 30g.

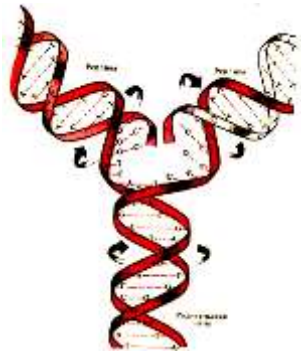
5. 20 types of enzymes - aminoacyl-tRNA synthetases.

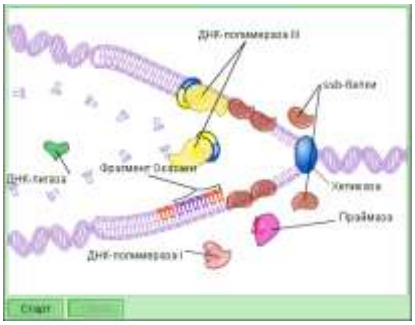
6. Additional protein factors: factors of initiation, elon-gation, and termination of translation.

After DNA transcription is completed, information from the mRNA molecule is translated into the protein molecule in accordance with the rules of the genetic code.

The genetic code is a system for recording genetic information that transfers information from the alphabet of nucleic acids to the alphabet of amino acids in proteins. The genetic code has a number of properties: the code is triplet, continuous, degenerate, specific, linear, universal.

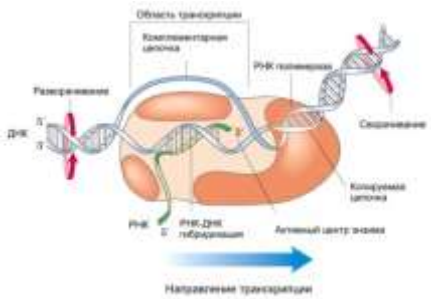
4. Illustrative material: Overview





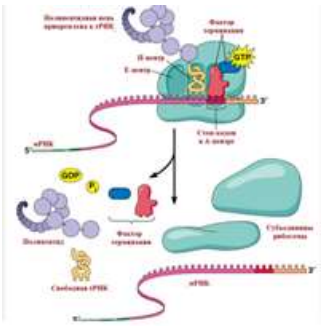
Semi-conservative DNA replication.

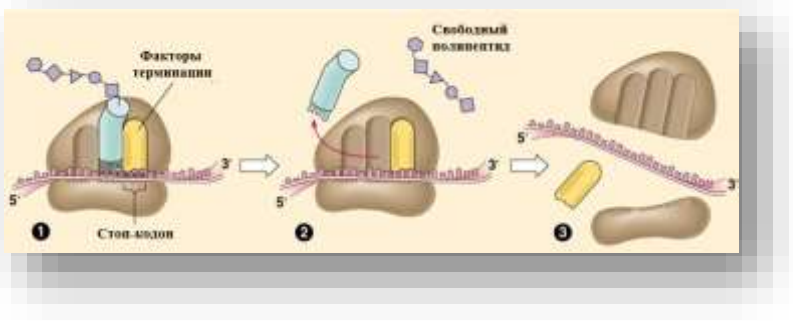
DNA transcription



Первое положение кодона (с 5'-конца)	Второе положение кодона				Третье положение кодона (с 3'-конца)
	У	Ц	А	Г	
У	УУУ – Фен	УЦУ – Сер	УАУ – Тир	УГУ – Цис	У
	УУЦ – Фен	УЦЦ – Сер	УАЦ – Тир	УГЦ – Цис	Ц
	УУА – Лей	УЦА – Сер	УАА – Стоп	УГА – Стоп	А
	УУГ – Лей	УЦГ – Сер	УАГ – Стоп	УГГ – Три	Г
Ц	ЦУУ – Лей	ЦЦУ – Про	ЦАУ – Гис	ЦГУ – Арг	У
	ЦУЦ – Лей	ЦЦЦ – Про	ЦАЦ – Гис	ЦГЦ – Арг	Ц
	ЦУА – Лей	ЦЦА – Про	ЦАА – Глн	ЦГА – Арг	А
	ЦУГ – Лей	ЦЦГ – Про	ЦАГ – Глн	ЦГГ – Арг	Г
А	АУУ – Иле	АЦУ – Тре	ААУ – Асн	АГУ – Сер	У
	АУЦ – Иле	АЦЦ – Тре	ААЦ – Асн	АГЦ – Сер	Ц
	АУА – Иле	АЦА – Тре	ААА – Лиз	АГА – Арг	А
	АУГ – Мет	АЦГ – Тре	ААГ – Лиз	АГГ – Арг	Г
Г	ГУУ – Вал	ГЦУ – Ала	ГАУ – Асп	ГГУ – Гли	У
	ГУЦ – Вал	ГЦЦ – Ала	ГАЦ – Асп	ГГЦ – Гли	Ц
	ГУА – Вал	ГЦА – Ала	ГАА – Глу	ГГА – Гли	А
	ГУГ – Вал	ГЦГ – Ала	ГАГ – Глу	ГГГ – Гли	Г

Genetic code





Translation of RNA

5.References: see appendix 1

6. Control questions: (feedback)

1. Stages of semi-conservative replication:

a. initiation,

b. elongation,

c. termination.

2. Factors of initiation, elongation, and termination of replication

3. DNA polymerases and their types.

4. PCNA protein, structure and functions.

5. DNA transcription is the first stage of expressing information about the protein structure. The mechanism of transcription.

6. Transcription factors:

- common transcription factors;

- DNA-binding proteins and their types;

- protein P-53 as a transcription factor.

7. The stages of transcription. Initiation, elongation, termination.

8. Principles of coding of genetic information.

9. The genetic code and its properties.

10. mRNA translation is the second stage of the realization of genetic information. The main components involved in protein synthesis.

Lecture No. 3

1. Topic: Molecular biology of the cell. Plasmolemma and its functions. Transport of substances through biomembranes. The adhesive function of membranes.

2. Purpose: To give an idea of the main cellular elements involved in the vital activity of the cell, as well as the mechanisms of formation of intercellular contacts, adhesion, extracellular matrix.

3. Lecture abstracts: Molecular biology is a complex of biological sciences that study the mechanisms of storage, transmission and realization of genetic information, the structure and functions of irregular biopolymers (proteins and nucleic acids). The three main components of a cell are the nucleus, cytoplasm, and the surrounding cell membrane, the plasmolemma. The cytoplasm of a cell includes the hyaloplasm, its mandatory cellular components, organelles, as well as various non—permanent structures, inclusions. The hyaloplasm is a complex colloidal system that includes various biopolymers such as proteins, nucleic acids, and polysaccharides. The hyaloplasm consists mainly of various globular proteins. The most important enzymes of the hyaloplasm include enzymes for the metabolism of sugars, nitrogenous bases, amino acids, lipids, and other important compounds. The hyaloplasm contains amino acid activation enzymes during protein synthesis, transport (transfer) RNA (tRNA).

Organelles are the most important component of a cell, cell structures that have a strictly defined structure and function.

Organelles are functionally divided into:

1 - organelles of general importance;

2 - organelles of special significance;

According to the structural principle, organelles are divided into:

1 - membrane (mitochondria, EPS, KG, lysosomes, peroxisomes);

2 – non-membrane (fibrillar) organelles (microtubules, microfilaments, cilia, flagella, centrioles) and granular organelles (ribosomes, polysomes).

Organelles are dynamic structures; they can change size, but they do not form. For the formation of new organelles, information is needed, in the form of a vestige or matrix from an existing organelle. Each organelle occupies a place in the hyaloplasm that is optimal for performing its specialized function.

Biomembranes are lipoprotein formations that restrict the cell from the outside and form some organelles, as well as the nuclear envelope - the karyolemma.

There are several types of membranes that differ in chemical composition, size, and function, but have a single structural plan.

A common feature of all cell membranes is that they are thin (6-10 nm) layers of lipoprotein nature (i.e. lipids in complex with proteins). The main chemical components of cell membranes are lipids (40%), proteins (60%) and carbohydrates (5-10%).

Lipids (Greek lipos - fat) are a group of natural substances that are insoluble in water, but soluble in nonpolar solvents (chloroform, ether, etc.). Lipid molecules are amphiphilic, that is, each lipid molecule has a hydrophilic (water–soluble) "head" and two hydrophobic (water-insoluble) of the "tail" (Fig.6, A). The molecules of the "tail" are a long hydrocarbon chain.

Membrane proteins make up 50% of the mass of cell membranes. Their role is that they ensure the functional activity of the membranes, namely:

1 - participate in the transport of substances;

2 - they are part of transport pumps and ion channels;

3 - they are enzymes and receptors involved in carrying signals into the cell;

4 - bind the cytoskeleton to the extracellular matrix;

5 - convert the energy of food substances into the chemical energy of the macro-energetic bonds of the ATP molecule.

According to their location in the membrane, proteins are divided into integral and superficial (peripheral) ones.

According to their functions, membrane proteins are divided into:

1 - structural;

2 - transport;

3 - adhesive (providing intercellular interactions);

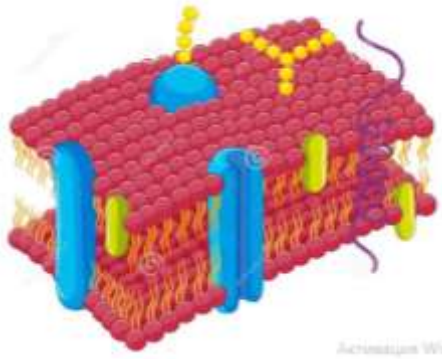
4 - involved in the transmission of signals from one cell to another;

5 - catalytic.

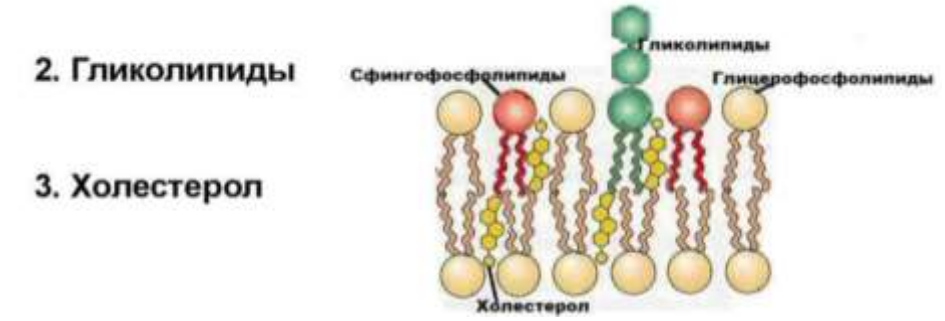
Molecular biology has historically emerged as a branch of biochemistry. By the beginning of the 21st century, data on the primary structure of all human DNA and a number of other organisms, most important for medicine, agriculture and scientific research, had been obtained, which led to the emergence of several new directions in biology: genomics, bioinformatics, etc.

Membrane transport is the transport of substances through the cell membrane into or out of the cell, carried out using various mechanisms — simple diffusion, facilitated diffusion and active transport. The most important property of a biological membrane is its ability to pass various substances into and out of the cell. Membrane transport (transport of substances through the lipid bilayer): passive and active. Active membrane transport is against an (electro)chemical gradient, i.e. energy costs are required (coupled with an energy–efficient process): primary and secondary.Passive membrane transport – along an (electro)chemical gradient, does not require energy costs: diffusion or facilitated diffusion. Energy sources for active membrane transport: ATP hydrolysis, light, redox reactions, (electro)chemical gradient. The energy for primary active transport comes from a source other than the existing (electro) chemical gradient. Channel proteins (protein channels) are a type of transport protein that acts as a pore in a membrane that quickly passes water molecules or small ions. Water channel proteins (aquaporins) allow water to diffuse through the membrane very quickly. Ion channel proteins allow ions to diffuse through the membrane. In most cases, signal transmission inside the cell is a chain of sequential biochemical reactions carried out by enzymes, some of which are activated by secondary mediators. Such processes are usually fast: their duration is on the order of milliseconds in the case of ion channels and minutes in the case of activation of protein kinases and lipid—mediated kinases. However, in some cases, it may take hours or even days from a cell receiving a signal to responding to it (in the case of gene expression). Signal transmission paths, or signal paths, are often organized as signal cascades. signal cascade): The number of protein molecules and other substances involved in signal transmission increases at each subsequent stage as it moves away from the initial stimulus. Thus, even a relatively weak stimulus can elicit a significant response. This phenomenon is called signal amplification. The original English term signal transmission first appeared in refereed journals in 1974, and appeared in the title of the article in 1979.

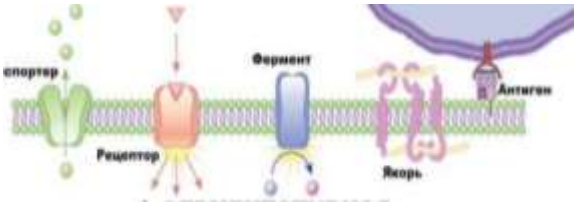
4. Illustrative material: Overview



Cell membrane: structure, properties, functions



LIPIDS OF BIOLOGICAL MEMBRANES



Functions of proteins in the composition of membranes

5. References: see appendix 1

6. Control questions: (feedback)

1. The structure of biomembranes
2. Biomembrane function
3. Adhesive function of biomembranes
4. Active transport.
5. Passive transport.
6. Structure and functions of cell organoids
7. The structure and functions of the nucleus
8. Structure and functions of the cell membrane
9. Structure and functions of the Golgi apparatus
10. Structure and functions of mitochondria
11. Structure and functions of the lysosome

Lecture No. 4

1. Topic: The molecular structure of cells and diseases that occur when their functioning is disrupted.

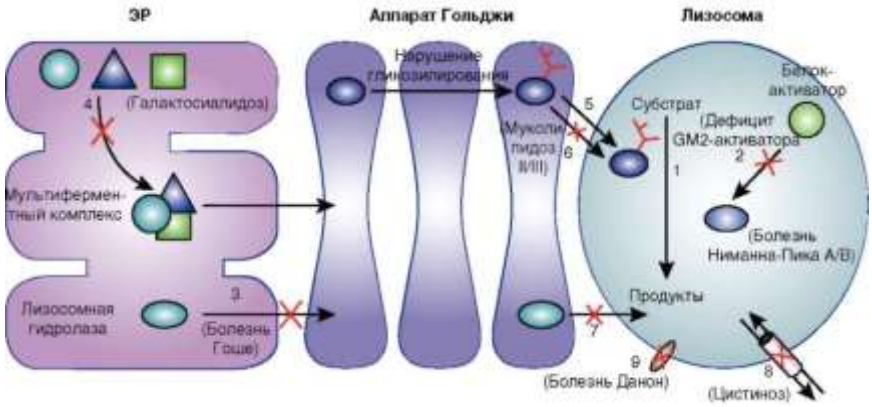
2. Purpose: To study the molecular structure of cells and diseases that occur when their functioning is disrupted

3. Lecture abstracts: Lysosomal Storage Diseases is the common name of a group of very rare hereditary diseases caused by impaired function of intracellular organelles of lysosomes. These single-membrane organoids are part of the cell's endomembrane system and specialize in the intracellular breakdown of substances: glycogen, glycosaminoglycans, glycoproteins, and others. Lysosomal storage diseases are caused by a genetically determined deficiency of lysosomal enzymes, which leads to the accumulation of macromolecules, which are the substrate of these enzymes, in various organs and tissues of the body. The clinical picture of the first hereditary disease from the group of lysosomal storage diseases (Tay—Sachs disease) was described in 1881. Then, in 1882, the disease was described, named after the French physician Philippe Gaucher, who first described it. In 1932, the Dutch physician John Pompe described glycogenosis of the second type, which was later named after him Pompe disease. In the late 1950s and early 1960s, the Belgian biochemist Christian de Duve and his co-authors, using the cell fractionation technique, discovered lysosomes as cellular organelles responsible for the cleavage and utilization of macromolecules. This scientific discovery soon made it possible to identify the pathophysiological basis of lysosomal storage diseases. Pompe disease was the first hereditary disease identified as lysosomal storage disease. In 1963, the Belgian physiologist and biochemist Henry Hers (eng. Henri G. Hers) published a paper in which he linked the cause of the development of this symptom complex with a deficiency of α-glucosidase and suggested the connection of other genetic diseases, including mucopolysaccharidoses, with a deficiency of an enzyme. Mitochondrial diseases are caused by genetic, structural, and biochemical defects in the mitochondria, leading to impaired tissue respiration. They are transmitted only through the female line to children of both sexes, since spermatozoa transfer half of the nuclear genome to the zygote, and the egg cell supplies both the second half of the genome and mitochondria. Pathological disorders of cellular energy metabolism can manifest themselves as defects in various links in the Krebs cycle, in the respiratory chain, beta oxidation processes, and so on. Not all enzymes and other regulators necessary for the efficient functioning of mitochondria are encoded by mitochondrial DNA. Most of the mitochondrial functions are controlled by nuclear DNA. Two groups of mitochondrial diseases can be distinguished:

Pronounced hereditary syndromes caused by mutations of genes responsible for mitochondrial proteins (Barth syndrome, Kearns—Sayre syndrome, Pearson syndrome, MELAS syndrome, MERRF syndrome, and others). Secondary mitochondrial diseases, including impaired cellular energy exchange as an important link in the formation of pathogenesis (connective tissue diseases, chronic fatigue syndrome, glycogenosis, cardiomyopathy, migraine, liver failure, pancytopenia, as well as hypoparathyroidism, diabetes, rickets, and others).

Mitochondria are inherited differently from nuclear genes. Nuclear genes in each somatic cell are usually represented by two alleles (with the exception of most sex-linked genes in the heterogeneous sex). One allele is inherited from the father, the other from the mother. However, mitochondria contain their own DNA, and each human mitochondria usually contains 5-10 copies of a circular DNA molecule (see Heteroplasmy), and all mitochondria are inherited from the mother. When a mitochondria divides, copies of DNA are randomly distributed among its descendants. If only one of the original DNA molecules contains a mutation, as a result of random distribution, such mutant molecules can accumulate in some mitochondria. Mitochondrial disease begins to manifest itself at the moment when a significant number of mitochondria in many cells of a given tissue acquire mutant copies of DNA (threshold expression). Mutations in mitochondrial DNA occur, for various reasons, much more often than in nuclear DNA. This means that mitochondrial diseases often manifest themselves due to spontaneous newly emerging mutations. Sometimes the rate of mutation increases due to mutations in nuclear genes encoding enzymes that control mitochondrial DNA replication.

4. Illustrative material: Overview



Biochemical and cellular bases of the pathogenesis of lysosomal storage diseases (according to Futerman A.H., G.v. Meer G., 2004): 1 - defect of the lysosomal enzyme, leading to a decrease in its activity; 2 - damage to the activator protein; 3 - disruption of enzyme transport from the endoplasmic reticulum (mutations causing a violation of protein conformation); 4 - violation of the formation of a multi-enzyme complex necessary for the transport of the enzyme from the endoplasmic reticulum; 5 - violation of the glycosylation of the enzyme in the Golgi apparatus; 6 - violation of enzyme glycosylation in the Golgi apparatus leads to the inability of the enzyme to bind to mannose-6-phosphate receptors and enter the lysosome; 7 - defect in enzyme transport from the Golgi apparatus; 8 - defects in lysosomal membrane carrier proteins; 9 - defects in lysosomal membrane proteins that play an important regulatory role in the functioning of lysosomes

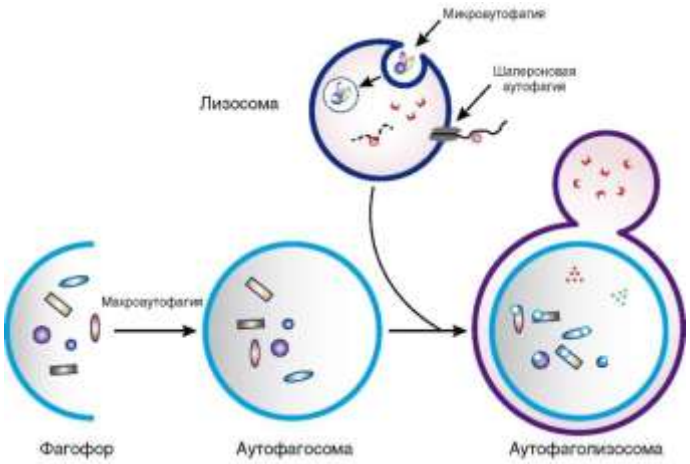


Fig. Types of autophagy

5. Literature:

Appendix 1

Main:

1. Esirkepov, M. M. Molecular biology of the cell: textbook. manual / M. M. Yessirkepov; Ministry of Healthcare of the Republic of Kazakhstan; Educational and methodological society of medical universities of the Republic of Kazakhstan. - Karaganda: IP "Publishing house of AKNUR", 2013. - 146 p.

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10. Internal diseases. Military field therapy / Edited by prof. A.L. Rakov and prof. A.E. Sosyukin. – C-Pb. - 2003*

11. Fundamentals of medical radiobiology / Edited by I.B. Ushakov. – St. Petersburg: Foliant Publishing House, LLC, 2004*.

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Additional information:

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2. Fundamentals of cell molecular Biology. Textbook. 3tomach. B.Alberts et al., OZON Publishing House.RU, 2018.

3. Fundamentals of molecular biology: a course of lectures/edited by T.A.Muminov;T.A.Muminov [et al.]. - 2nd ed., ispr. and add. - Almaty: Lit. Print. Kazakhstan, 2017. - 556 p.

6. Control questions: (feedback)

1.Diseases related to the pathology of the nucleus: reduction of genetic material atypical mitoses pathology of synthesis of ribosome and tRNA subunits in the nucleolus

2. Diseases associated with impaired functioning and structure of EPS: expansion of EPS cisterns, EPS fragmentation, EPS hyper and hypotrophy, blockade of synthetic and/or transport processes in the cell.

3. Diseases associated with impaired functioning and structure of the Golgi apparatus: diseases associated with impaired intracellular transport signals

4. Diseases associated with impaired functioning and structure of mitochondria: mitochondrial diseases associated with defects in nuclear DNA mitochondrial diseases caused by defects in mtDNA

5. Diseases associated with impaired functioning and structure of lysosomes: diseases of accumulation of mucopolysaccharides or genetic accumulation diseases; diseases associated with disorders of sorting and transport of lysosomal enzymes - hydrolases. diseases associated with damage to lysosomal membranes. diseases associated with extracellular release of lysosomes in the development of inflammatory processes

6. Diseases associated with impaired functioning and structure of peroxisomes: diseases caused by an almost complete loss of peroxisome function; diseases caused by an excess of peroxisome enzymes; diseases caused by a malfunction of only one peroxisome enzyme.

7. Diseases associated with impaired membrane functioning. Diseases associated with changes in the structure and number of elements of the cytoskeleton.

8. What diseases are caused by cell malfunction? - Cancer, ciliopathies, Alzheimer's and Parkinson's diseases are just some of the numerous diseases associated with impaired cell transport

9. What is the cause of the disease at the molecular and cellular level? - Small changes in cells at the molecular level lead to disruption of cellular function, which leads to disease. For example, mutations in the p53 gene alter the ability of cells to cause cell cycle arrest to repair DNA damage, leading to faulty cell division, potentially leading to cancer.

Medical genetics

Lecture No. 1

1. Topic: Introduction to genetics. Fundamentals of general genetics.

2. Purpose: To provide a presentation on the subject and objectives of medical genetics, its role in medicine;

3. Lecture theses: Genetics is the science of the laws of heredity and variability. Depending on the object of research, plant, animal, and other genetics are classified; depending on the methods used in other disciplines, molecular genetics, environmental, and others. The ideas and methods of genetics play an important role in medicine, agriculture, the microbiological industry, as well as in genetic engineering.

Medical genetics is a field of genetics, a science that studies:

- phenomena of heredity and variability at all levels of its organization and existence: molecular, cellular, organismic, population-specific features of the manifestation and development of normal and pathological signs,

- the role of heredity in human pathology, patterns of transmission from generation to generation of hereditary diseases,

- hereditary human diseases,

- dependence of diseases on genetic predisposition and environmental conditions, methods of diagnosis, treatment and prevention of hereditary pathology, including diseases with hereditary predisposition.

Tasks of medical genetics:

diagnosis of hereditary diseases

• analysis of their prevalence in different populations and ethnic groups

• prevention of hereditary diseases based on prenatal (prenatal) diagnostics

• study of the molecular and genetic foundations of the etiology and pathogenesis of hereditary diseases

• identification of sick children

• development of recommendations for their treatment.

• It is impossible to study human inheritance using hybridological analysis (the method of crosses).

• Specific methods are used for human genetic analysis:

• genealogical (a method of analyzing pedigrees),

• gemini,

• Cytogenetic,

• biochemical,

• dermatoglyphics and palmoscopy

• molecular genetic (DNA diagnostics)

• population statistical,

• genetics of somatic cells

The cytogenetic method is based on microscopic examination of chromosomes, normal human karyotype and pathology.

This method allows us to establish the presence of hereditary human diseases, study the structures of chromosomes, detect translocations, build genetic maps, analyze chromosomal and genomic mutations, conduct cytochemical studies of gene activity, etc

. The clinical and genealogical method was proposed at the end of the 19th century by F. Galton. It is based on the construction of pedigrees and tracing the transmission of a hereditary trait in a number of generations.

The biochemical method makes it possible to determine the contribution of genetic (hereditary) and environmental factors (climate, nutrition, education, upbringing, etc.) in the development of specific signs or diseases in humans.

Hereditary diseases are numerous (over 6,000 are known) and diverse in their manifestations.

Such diseases can occur quite rarely, but due to the fact that there are many of them, their total frequency is quite high.

They differ from other diseases in that, as a rule, it is possible to find the exact cause of the disease, which is associated with damage to the hereditary apparatus.

Classification of hereditary human diseases, the most commonly used:

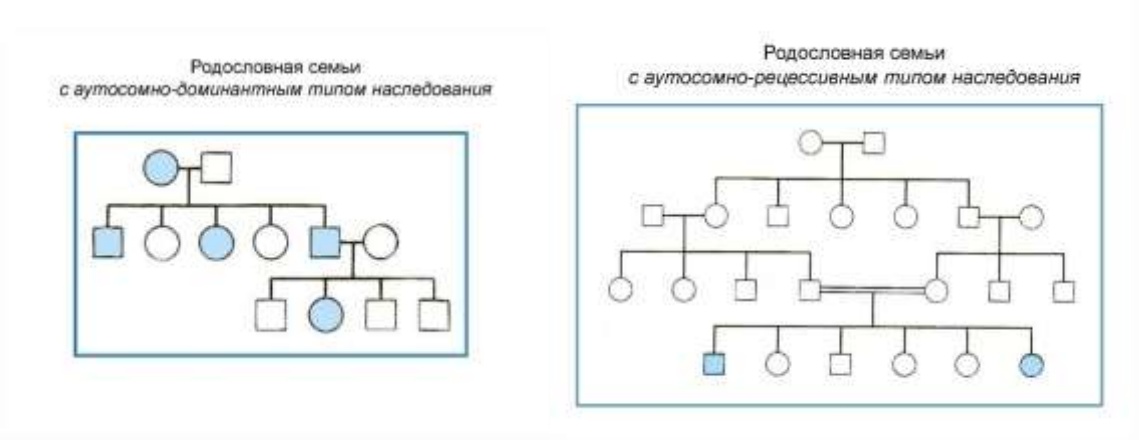
1) monogenic mendelian diseases);

2) chromosomal syndromes resulting from structural or quantitative rearrangements of chromosomes;

3) multifactorial diseases,

4) monogenic diseases with an unconventional, different from Mendelian, type of inheritance — this group has been identified in the last decade.

4. Illustrative material: Overview



The genealogical method

5.References: see appendix 1

6. Control questions: (feedback)

1. Who is the father of genetics?

2. What is the significance of genetics for medicine?

3. What is the characteristic of independent inheritance?

4. What is the essence of linked inheritance?

5. What are the causes of hereditary human diseases?

Lecture No. 2

1. Topic: Introduction to medical genetics. Methods of human genetics research

2. Purpose: To provide an introduction to the methods of studying human genetics: twin, dermatoglyphics and palmoscopy, genetics of somatic cells, population –static, biochemical, cytogenetic, clinical and genealogical. The principle of pedigree analysis

3. Lecture abstracts: Medical genetics is a field of genetics, a science that studies:

the phenomena of heredity and variability at all levels of its organization and existence: molecular, cellular, organizational, population features of the manifestation and development of normal and pathological signs, the role of heredity in human pathology, patterns of transmission from generation to generation of hereditary diseases, hereditary human diseases, dependence of diseases on genetic predisposition and environmental conditions, diagnostic methods, treatment and prevention of hereditary pathology, including diseases with hereditary predisposition.

Tasks of medical genetics: diagnosis of hereditary diseases

• analysis of their prevalence in different populations and ethnic groups

• prevention of hereditary diseases based on prenatal (prenatal) diagnostics

• study of the molecular and genetic foundations of the etiology and pathogenesis of hereditary diseases

• identification of sick children

• development of recommendations for their treatment.

• It is impossible to study human inheritance using hybridological analysis (the method of crosses).

• Specific methods are used for human genetic analysis:

• genealogical (a method of analyzing pedigrees),

• gemini,

• Cytogenetic,

• biochemical,

• dermatoglyphics and palmoscopy

• molecular genetic (DNA diagnostics)

• population statistical,

• genetics of somatic cells

The cytogenetic method is based on microscopic examination of chromosomes, normal human karyotype and pathology.

This method allows us to establish the presence of hereditary human diseases, study the structures of chromosomes, detect translocations, build genetic maps, analyze chromosomal and genomic mutations, conduct cytochemical studies of gene activity, etc

. The clinical and genealogical method was proposed at the end of the 19th century by F. Galton. It is based on the construction of pedigrees and tracing the transmission of a hereditary trait in a number of generations.

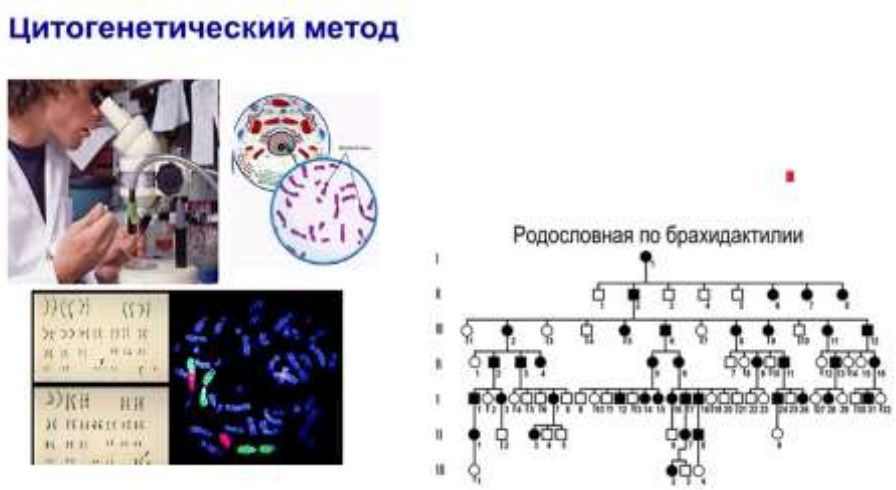
The biochemical method makes it possible to determine the contribution of genetic (hereditary) and environmental factors (climate, nutrition, education, upbringing, etc.) in the development of specific signs or diseases in humans.

Hereditary diseases are numerous (over 6,000 are known) and diverse in their manifestations.

Such diseases can occur quite rarely, but due to the fact that there are many of them, their total frequency is quite high.

They differ from other diseases in that, as a rule, it is possible to find the exact cause of the disease, which is associated with damage to the hereditary apparatus.

4. Illustrative material: Overview



The genealogical method

5.References: see appendix 1

6. Control questions: (feedback)

1. What is the significance of genetics for medicine?

2. What characterizes independent inheritance?

3. What are the causes of human hereditary diseases?

4. Is it possible to cure human hereditary diseases?

5. Definition of hereditary diseases

6. Mechanisms of occurrence of hereditary diseases

Lecture No. 3

1. Topic: Monogenic, polygenic or multifactorial diseases. Chromosomal diseases and diseases with an unconventional type of inheritance

2. Purpose: To study hereditary human diseases. The study of the etiology, pathogenesis and epidemiology of hereditary diseases. Hereditary diseases. Genetic mechanisms of occurrence.

3. Lecture notes: Hereditary diseases are numerous (over 6,000 are known) and diverse in their manifestations.

Such diseases can occur quite rarely, but due to the fact that there are many of them, their total frequency is quite high.

They differ from other diseases in that, as a rule, it is possible to find the exact cause of the disease, which is associated with damage to the hereditary apparatus.

Classification of hereditary human diseases, the most commonly used:

-monogenic mendelian diseases);

-chromosomal syndromes resulting from structural or quantitative rearrangements of chromosomes;

- multifactorial diseases,

-monogenic diseases with an unconventional, different from Mendelian, type of inheritance — this group has been identified in the last decade.

Hereditary diseases are diseases that are transmitted from parents to offspring. Hereditary diseases are formed due to changes in the genetic material caused by gene, chromosomal and genogenomic mutations.

Hereditary diseases according to genetic classification:

* monogenic;

* Chromosomal;

* multifactorial (polygenic).

Monogenic diseases are caused by mutations in the structural genes in which genetic information is recorded. The transmission of these diseases to offspring is called Mendelian hereditary disease, as it occurs according to the laws of inheritance of G. Mendel. Monogenic type of autosomes.- dominant (arachnodactyly, brachydactyly, polydactyly, etc.), autosomal.- recessive (more common in people married to two and sometimes three cousins; agammaglobulinemia, alkaptonuria, etc. d.) and combined with sexual X and Y chromosomes (depending on the gene, the male gets sick, and the disease is transmitted by the female; hemophilia, etc. Diseases) are divided into hereditary diseases.

Chromosomal diseases are formed due to genomic (changes in the number of chromosomes) and chromosomal (changes in the structure of chromosomes) mutations. Among the most common chromosomal diseases are trisomies. At this point, an additional 3 chromosome is formed in one of the pairs of chromosomes. For example, autosomes in Down's disease. Trisomy in pairs of 21 is present in pairs of 13 in Patau syndrome and in pairs of 18 in Edwards syndrome. Due to a violation of meiotic division in gametogenesis in

women, if one of the sex X chromosomes is missing, then Shereshevsky-Turner syndrome, on the contrary, with an excess of one chromosome, leads to the formation of triplo-X syndrome (Klinefelter syndrome in men). The chromosomes of infants during pregnancy in women over 35 years of age. childbirth with this disease is associated with a high risk.

Multifactorial diseases occur as a result of mutations and interactions of several genes, when adaptation to the disease increases, and due to environmental factors.

Such diseases include

• gout;

* diabetes mellitus;

* hypertension;

* stomach and intestinal ulcers;

* atherosclerosis;

* refers to coronary heart disease, etc.

The cause of this type of hereditary diseases is still not fully understood. The clinical classification of hereditary diseases is carried out according to organs and systems that have undergone pathological changes. For example, hereditary diseases of the nervous and endocrine systems, circulatory system, liver, kidneys, skin, etc. are classified as hereditary diseases of organs. Clinics and hospitals of neurology, therapy, and surgery are engaged in the diagnosis and treatment of hereditary diseases in the republic.

Diseases with an unconventional type of inheritance include: mitochondrial diseases resulting from mutations of mitochondrial genes. Diseases of genomic imprinting. Diseases of the expansion of trinucleotide repeats, in the regulatory or transcribed parts of genes. Diseases caused by impaired epigenetic regulation of gene expression.

4. Illustrative material: Overview

5.References: see appendix 1

6. Control questions: (feedback)

1. What are the causes of hereditary human diseases?
2. Is it possible to cure hereditary human diseases?
3. Definition of hereditary diseases
4. Mechanisms of occurrence of hereditary diseases
5. Monogenic diseases
6. Polygenic diseases
7. Chromosomal diseases and their place in general human pathology.
8. Classification of chromosomal diseases:

a. Etiological (based on the nature of the mutation):

b. Chromosomal diseases associated with numerical abnormalities of chromosomes while maintaining their structure;

9. Chromosomal diseases caused by structural rearrangements of chromosomes: deletions, duplications, inversions, translocations.

10. Definition of diseases by the Lemendel type of inheritance.

Lecture No. 4

1. Topic: Fundamentals of molecular genetic diagnostics, prenatal diagnosis and prevention of hereditary diseases

2. Purpose: The purpose of combining molecular genetic diagnostics, prenatal diagnosis and prevention of hereditary diseases is to prevent the birth of children with severe hereditary diseases by early detection of fetal pathology and providing parents with information to make informed decisions about further pregnancy

3. Lecture notes: Molecular genetic diagnostics uses DNA analysis to detect hereditary diseases, whereas prenatal diagnostics includes ultrasound and biochemical markers to detect pathologies in the fetus before birth. The prevention of hereditary diseases is aimed at reducing the risk of their occurrence and is carried out through medical and genetic counseling and prenatal research.

The basics of molecular genetic diagnostics are research methods that identify changes in the genetic material (DNA) that can lead to hereditary diseases. They are used to diagnose specific hereditary syndromes and diseases.

Prenatal diagnosis is the conduct of research during pregnancy to identify pathologies, which allows you to decide on further actions or prepare for the birth of a child with certain health characteristics. This is a set of measures aimed at detecting congenital and hereditary fetal diseases at the stage of intrauterine development.

Methods include:

• Non-invasive methods:

- Ultrasound: Visualization of the fetus to identify developmental abnormalities.

-Biochemical markers: Analysis of HCG (human chorionic gonadotropin) and RARP-A (pregnancy-associated plasma protein A) levels as indicators of chromosomal abnormalities.

• Invasive methods:

-Placentocentesis and cordocentesis: Sampling of placental or umbilical cord blood for a more accurate analysis of the genetic material of the fetus.

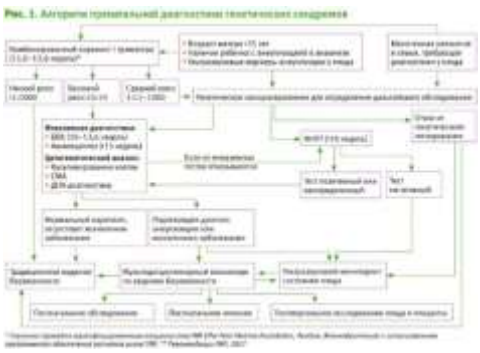
-Application: Diagnosis of Down syndrome, Klinefelter syndrome, Turner syndrome and other genetic abnormalities.

Prevention of hereditary diseases - preventive measures are aimed at reducing the risk of having a child with a hereditary disease. Main approaches:

-Medical and genetic counseling:

Consultation with a geneticist to assess the risks of hereditary diseases in the family and determine further actions.

- The cytogenetic method is used to study chromosomes, as well as in the diagnosis of hereditary diseases associated with genomic and chromosomal mutations. In addition, this method is used to study the mutagenic effects of various chemicals, pesticides, insecticides, medicines, etc.



4. Illustrative material: Overview

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Appendice1

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6. Control questions: (feedback)

1. What is molecular genetic diagnostics? - a relatively new method of examination of the body, which makes it possible to accurately and quickly identify viruses and infections, gene mutations that cause pathology, and assess the risks of hereditary and other diseases. And this is far from the full range of possibilities for DNA research.

2. When is the genetic testing performed? - the use of genetic research is relevant in cases where the patient seeks to obtain information about the state of his body. This is usually necessary in the following situations:

• to make an accurate diagnosis. For example, it is very common to misidentify an allergen or untimely diagnosis of a viral disease. Meanwhile, the success of treatment depends on it.;

• for the prevention of possible pathologies. If a patient is aware of an increased risk of cardiovascular disease or cancer, they can take appropriate measures, for example, to give up bad habits.;

• to increase the effectiveness of treatment. For example, oncological diseases have many treatment options. The choice of treatment tactics by trial and error leads to the loss of precious time and health, and sometimes to death.;

3. Methods of molecular genetic diagnostics? - Methods of molecular cytogenetics, Molecular diagnostics by PCR, The method of fluorescent hybridization (FISH), Microchipping

4. Prenatal diagnostics (ultrasound, biochemical screenings, invasive methods such as chorionic biopsy and amniocentesis),

5. As well as methods of preventing hereditary diseases

6. Such as genetic counseling and preimplantation diagnosis.

Embryology

Lecture No. 1

1. The theme: Introduction to embryology

2. Purpose: To give an idea of the subject and objectives of embryology, its role in medicine.

3. Lecture notes: Embryology is a science that studies the development of the embryo from conception to birth, as well as the patterns of individual development of the body as a whole. She considers the proembryonic period (gametogenesis — the formation of germ cells), the prenatal period (development inside the mother's womb) and subsequent stages, including zygote formation, fragmentation, gastrulation, organogenesis and systemogenesis. Knowledge of embryology is important for medicine, especially in obstetrics, pediatrics and reproductive medicine, including IVF and the diagnosis of congenital pathologies.

Basic concepts:

Embryogenesis is the process of individual development of an organism, beginning with the formation of a zygote and ending with birth.

-The proembryonic period (progenesis, gametogenesis) — precedes the formation of the zygote and includes the formation of male (spermatogenesis) and female (oogenesis) germ cells.

- The prenatal period is the intrauterine development of the body.

- An embryo is an embryo in the early stages of development, from conception to the end of the 8th week of pregnancy.

- The fetus is an organism from the 9th week of development to birth.

The main stages of embryogenesis

1. Fertilization: The formation of a zygote by the fusion of male and female germ cells.

2. Fragmentation: Rapid division of the zygote to form a blastocyst.

3. Gastrulation: The formation of germ layers, from which all tissues and organs then develop.

4. Organogenesis and histogenesis: Development and formation of organs and tissues.

5. Systemogenesis: The formation of the main organ systems of the body.

Application and meaning

- Medicine: Diagnosis of fetal developmental disorders, identification of the causes of birth defects, infertility treatment.

- Assisted reproductive technologies (ART): Development of contraceptive drugs, in vitro fertilization (IVF), cryopreservation of embryos.

Understanding evolution: Human embryological development partially reflects the evolution of vertebrates.

Human embryology is a branch of science that studies the development of the embryo, that is, the body in the early stages of development before birth. Knowledge in the field of human embryology is essential for all doctors, especially those working in pediatrics and obstetrics.

Knowledge of embryology helps in the diagnosis of disorders in the mother-fetus system, the detection of diseases of children after birth, as well as the identification of the causes of deformities.

Today, knowledge in the field of embryology is used to identify and eliminate the causes of infertility, develop contraceptive drugs, and transplant fetal organs. The problems of embryo transplantation into the uterus, in vitro fertilization and egg culture have become urgent.

Embryology studies several stages of embryo development:

• fertilization followed by the formation of a zygote;

• crushing and formation of blastocyst;

• gastrulation is the process of formation of germ sheets and axial organs;

• organogenesis and histogenesis of extra-germinal and germinal organs;

• systemogenesis.

Intrauterine development is divided into three main periods:

• initial – the first week;

• germinal – second to eighth weeks;

• fetal – begins at the ninth week and ends with the birth of a child.

On average, a person's intrauterine development lasts 280 days.

Embryology: the stage of fertilization and zygote formation

Fertilization is the process of fusion of male and female germ cells, as a result of which a diploid set of chromosomes is restored and a new cell is created – a fertilized egg (zygote). For the possibility of fertilization, the concentration of spermatozoa in the ejaculate should correspond to 20-200 million/ml, and their total number should be 150 million/ml.

The fertilization process consists of three phases:

• distant interaction and convergence of gametes;

• contact interaction with egg activation;

• penetration of the sperm into the egg, followed by syngamy (fusion). Distant interaction provides chemotaxis, a set of specific factors responsible for increasing the likelihood of male and female germ cells meeting. Chemicals produced by germ cells play an important role in this process. Immediately after ejaculation, the process of capacitation occurs – spermatozoa acquire fertilizing ability under the influence of the secretion of the female genital tract. Hormonal factors (for example, progesterone) that activate the secretion of the fallopian tubes have a great influence on the mechanism of capacitation. Fertilization occurs in the fallopian tubes, preceded by insemination due to chemotaxis. During contact, spermatozoa approach the egg, and then come into contact with its shell. Next, the process of penetration of the head

and tail of sperm into the ovoplasm occurs. A fertilization shell forms on the periphery of the ovoplasm. Within 12 hours after the convergence of the male and female pronuclei, a single–celled embryo, a zygote, is formed in a woman's body.

Embryology: the stage of fragmentation and blastocyst formation

Fragmentation is the sequential process of dividing a zygote without the growth of blastomeres. In humans, fragmentation is complete, asynchronous, and uneven. After the first crushing, two blastomeres are formed in a woman's body. One of the blastomeres has a larger size and a dark color, the second is light and smaller. A large blastomer forms the embryo and most of the accessory organs: the fetal part of the placenta and connective tissue of the chorion, yolk sac, amnion, and allantois. A trophoblast develops from the second blastomer.

Formation of a blastula: Small cells divide faster than large ones during crushing and grow on their outside. Thus, a morula is formed – a cluster of cells. Inside it are large cells called embryoblasts, and outside are small cells called trophoblasts. During cell division, the morula increases in size, and fluid begins to be secreted by the cells of the embryo and accumulate under the trophoblast. Subsequently, the volume of fluid increases, a cavity inside the embryo is formed, filled with such a liquid, the embryoblast is pushed to the periphery and adheres to the trophoblast. A blastocyst is formed. The trophoblast forms outgrowths, villi, as a result of which the surface of the blastula is uneven. The trophoblast is the first provisory organ formed in the embryo. In the future, the trophoblast will become part of the placenta. Through the trophoblast, the embryo is implanted into the uterine mucosa.

Embryology: gastrulation stage: As a result of the movement of cells after the formation of a blastula, a gastrula is formed – a two-layered embryo. The process of gastrula formation is

called gastrulation. During gastrulation, intensive cell movement occurs – the future beginnings of tissues move in accordance with the plan of the structural organization of the future full-fledged organism. At the gastrulation stage, the embryo consists of germ sheets, divided layers of cells. The outer layer is the ectoderm, the inner layer is the endoderm. In vertebrates, a third layer (the middle one) is formed – the mesoderm.

The following develop from the ectoderm:

• Skin epithelium;

• nervous system;

• tooth enamel;

• Sensory organs.

Endoderm develops into:

• lung epithelium;

• digestive glands;

• the epithelium of the midgut.

They develop from the mesoderm:

• the circulatory system;

• connective and muscular tissues;

• sex glands;

• kidneys , etc .

There are several ways of gastrulation:

• Invagination is carried out by retracting the blastula wall into the blastocele;

• delamination – cells located on the outside are transformed into the epithelial layer of the ectoderm, and the remaining cells form the endoderm. Delamination is typical for intestinal cavities;

• epibolia – overgrowth of cells with incomplete fragmentation of the internal mass of the yolk or overgrowth of cells by other rapidly dividing cells;

• immigration – migration into the blastocele of a part of the cells of the blastula wall;

• involution – the insertion of the outer layer of cells, increasing in size, into the embryo.

Embryology: the stage of histogenesis and organogenesis of extra-embryonic and germinal organs

Organogenesis is a set of processes leading to the formation of rudiments of organs and their subsequent differentiation during embryonic development.

In organogenesis, they are distinguished:

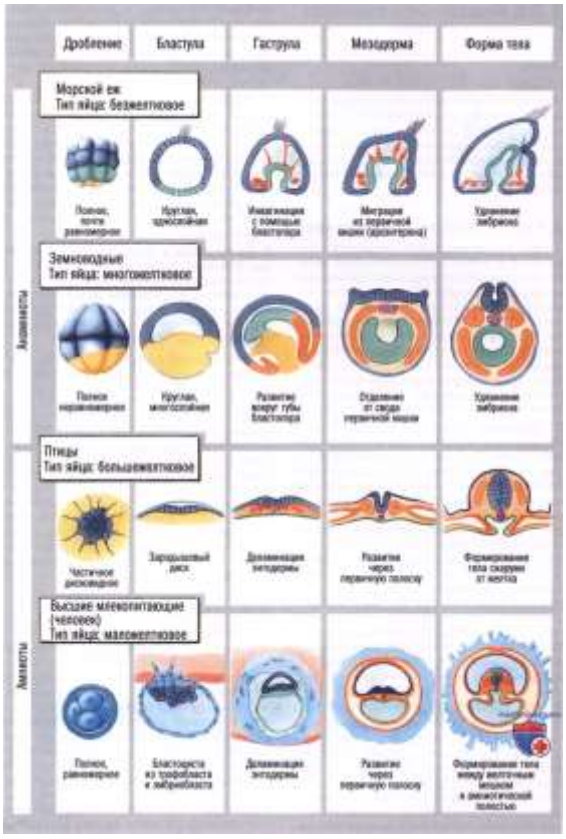
• Neurulation is the process of neurula formation. The mesoderm is laid in the neurule, which, in turn, consists of germ layers and an axial organ complex – the chord, neural tube and intestine. The cells of the organ complex influence each other. This effect is called embryonic induction.

• Histogenesis is a series of processes that ensure the formation and repair of tissues during ontogenesis.

Today, embryology has become one of the most important areas of science. In medicine, its use is not limited to the field of histology and anatomy. Embryology is important in the development of preventive medicine aimed at the development and testing of new medicines

and the fight against hereditary diseases. Embryology has great prospects related to the development of genetics and a number of other sciences. Embryology is also closely related to IVF, as the embryological period is one of the most important stages of the in vitro fertilization program. Clinical embryology studies the causes of disorders of embryonic development, the mechanisms of development of deformities, as well as ways to influence embryogenesis. IVF developments have become possible thanks to the use of high-tech medicine and the development of clinical embryology. The outcome of in vitro fertilization largely depends on the knowledge and experience of an embryologist.

4.Illustrative material: Overview



Comparison of early embryo development in different species

5. References: see appendix 1

6. Control questions: (feedback)

1. What is embryology? Embryology is a scientific discipline that studies the development of living organisms, starting with a fertilized egg (zygote) and ending with birth.

2. The main stages of the body's development: Fragmentation: -the initial stage when a unicellular zygote divides into many cells. Gastrulation: -the process of forming three germ layers that will give rise to all tissues and organs. Neurulation: -the formation of a neural tube from which the brain and spinal cord develop. Organogenesis: -formation of all internal organs and body systems.

3. How does the fusion of male and female germ cells occur? Fertilization

4. What is the role of germ layers in organ formation? Germ leaves:

5. How do genes affect embryo development? Genetics:

6. How are the various organs and systems of the body formed and functioning? Organ development:

7. How does embryology help in the treatment of infertility and IVF? Reproductive health:

8. How can embryonic development help us understand the evolution of species? Evolution:

Lecture No. 2

1. Topic: The actual embryonic stage of human embryogenesis

2. Purpose: formation of the human body from a fertilized egg (zygote) to birth, including the stages of division, formation of germ layers (gastrulation), formation of organs and systems (organogenesis), as well as the formation of extra-germ organs such as the placenta. The main processes occurring at this stage are fertilization, crushing, implantation, gastrulation and organogenesis, which lead to the creation of the basic vital systems of the future person.

3. Lecture abstracts: Human embryogenesis - the development and formation of the human embryo. It is characterized by the process of cell division and cell differentiation in the embryo, which occurs in the early stages of development. In biological circles, human development implies growth from a unicellular zygote to an adult. Fertilization occurs when a sperm cell successfully enters and merges with an egg cell. The genetic material of the sperm and egg is then combined into a single cell called a zygote. Embryogenesis covers the first eight weeks of development; at the beginning of the ninth week, the embryo begins to be called a fetus. The 8 weeks consist of twenty-three stages.

Human embryology is the study of fetal development during the first eight weeks after fertilization. The normal gestation period is about 9 months or 40 weeks.

The embryonic stage refers to the period beginning with fertilization through the development of an early embryo before the embryo is fixed in the uterus. The germinal stage takes about 10 days[3]. At this stage, the zygote begins to divide. This process is called splitting. Then a blastocyst is formed, which is then implanted into the uterus. Embryogenesis continues with the next stage, gastrulation, when 3 germ layers are formed during

histogenesis. This is followed by the processes of neurulation and organogenesis. Compared to the embryo, the fetus has a more recognizable appearance and a more complete set of internal organs. The entire process of embryogenesis involves coordinated spatial and temporal changes in gene expression, cell growth, and differentiation.

Fragmentation is a series of successive mitotic divisions of a fertilized or development-initiated egg. Fragmentation is the first period of embryonic development, which is present in the ontogenesis of all multicellular animals. At the same time, the mass of the embryo and its volume do not change, remaining the same as at the beginning of crushing. The egg divides into smaller and smaller blastomere cells. A characteristic feature of fragmentation is the leading regulatory role of the cytoplasm in development. The type of crushing depends on the amount of yolk and its location in the egg.

Gastrulation is the separation of two primary germ layers (ectoderm and endoderm) in the embryonic development of all multicellular animals. The second stage of ontogenesis after fragmentation. During gastrulation, a gastrula is formed from the blastula. The third germ layer, the mesoderm, can form during gastrulation or (as in the lancelet) later. It is a collection of cells located between the ectoderm and the endoderm. Due to the appearance of the mesoderm, the embryo becomes three-layered. Sponges and bilayers (scallops and ctenophores) do not have mesoderms.

The concept of gastrula and gastrulation

Gastrulation is the process of formation of three germ layers.:

- Ectoderm: (the outer layer) from which the nervous system, skin epidermis, and sensory organs develop.

- Endoderm: (inner layer), from which the epithelium of the digestive tract, respiratory tract and gland is formed.

- Mesoderm: (the middle layer), from which the bone and muscle systems, circulatory system and kidneys are formed.

Organogenesis, histogenesis, and morphogenesis are the stages of embryonic development associated with the formation of organs, tissues, and the overall shape of an organism. Histogenesis is the formation of tissues from germ layers, organogenesis is the subsequent formation of organs from these tissues, and morphogenesis is a broader process of formation of an organism and its parts, including both histo- and organogenesis, as well as other stages.

Histogenesis is the process of tissue formation (a collection of cells of similar structure and functions) from germ layers (ectoderm, mesoderm and endoderm).

When it occurs: It begins after the completion of gastrulation, when these three germ layers are formed.

Example: Muscles and connective tissues are formed from the mesoderm, and skin and nervous system are formed from the ectoderm.

Organogenesis is the process of forming organs that are more complex structures consisting of various tissues.

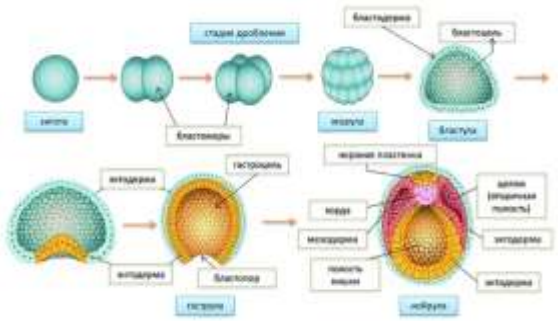
When it occurs: It follows histogenesis, at this stage the embryo already contains organs and systems.

Example: The central nervous system begins to form from already formed tissues (for example, from nervous tissue).

Morphogenesis is a general term that encompasses all the processes by which an organism acquires its specific shape and structure.

What it includes: Morphogenesis includes histogenesis, organogenesis, and other processes such as cell differentiation, growth, and formation of organs and tissues.

4.Illustrative material: Overview



Embryonic development

5.References: see appendix 1

6. Control questions: (feedback)

1. Histogenesis? The process of tissue formation (a collection of cells of similar structure and functions) from germ layers (ectoderm, mesoderm and endoderm).

2. Organogenesis? The process of forming organs, which are more complex structures consisting of various tissues.

3. Morphogenesis? A general term that covers all the processes by which an organism acquires its specific shape and structure.

4. Gastrulation? This is the process of forming three germ layers.:

5. Ectoderms? (the outer layer), from which the nervous system, the epidermis of the skin, and the sensory organs develop.

6. Endoderms? (inner layer), from which the epithelium of the digestive tract, respiratory tract and gland is formed.

7. Mesoderms? (middle layer), from which the bone and muscle systems, circulatory system and kidneys are formed.

8. Crushing? A series of successive mitotic divisions of a fertilized or development-initiated egg.

Lecture No. 3

1. Topic Genetics of embryonic development

2. Purpose: to study hereditary information at all stages of the formation of the body, to identify and prevent congenital genetic abnormalities and diseases, as well as to investigate the influence of endogenous and exogenous factors on the development of the embryo in order to ensure the birth of healthy offspring.

3. Lecture notes: Embryonic development takes 8 weeks before the embryo develops. The development of the human embryo depends on stem cells. During embryonic development, cells divide, migrate, and specialize. In the early stages of development, a group of cells is formed called the inner cell mass, which is capable of producing all body tissues. Later, during gastrulation, three germ layers form, and most cells become limited in the type of cells they produce.

The genetics of embryonic development studies how genes control the processes of cell division, migration, and specialization leading to the formation of a multicellular organism from a fertilized egg. Genes responsible for the early stages of development - fragmentation, gastrulation, neurulation, and organogenesis - are inherited and expressed, followed by the formation of organs and tissues. Modern technologies, such as PGD and NIPT, make it possible to study the genetic data of the embryo and the mother's body for the diagnosis of chromosomal abnormalities and hereditary diseases.

The main processes controlled by genes: During embryonic development, a complex sequence of events occurs that are regulated by genes:

- Fragmentation: After fertilization, the zygote begins to divide, forming blastomeres. Genes determine the synchronicity of these divisions and the beginning of cell differentiation.

Gastrulation: This stage involves the formation of germ layers (ectoderm, mesoderm and endoderm), which will give rise to all tissues and organs of the body.

- Neurulation: The formation of the neural tube, the precursor of the nervous system, is also controlled by genetic programs.

- Organogenesis: The formation and development of the main organs and systems of the body from germ layers, which is a complex process of gene regulation.

The importance of genetic control:

- Species specificity: Genes determine what a developing organism will be like, providing it with species-specific traits.

- Cell differentiation: Genes control the process when different specialized cells, tissues, and organs are formed from identical cells.

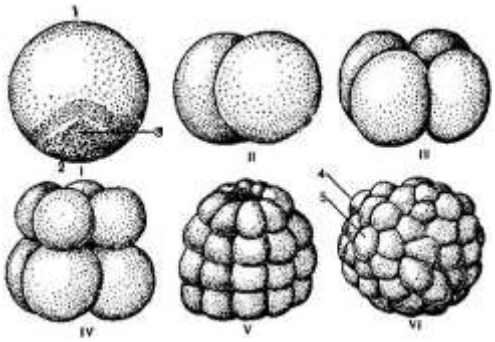
- Development regulation: Genetic programs ensure the correct sequence and timeliness of all stages of development.

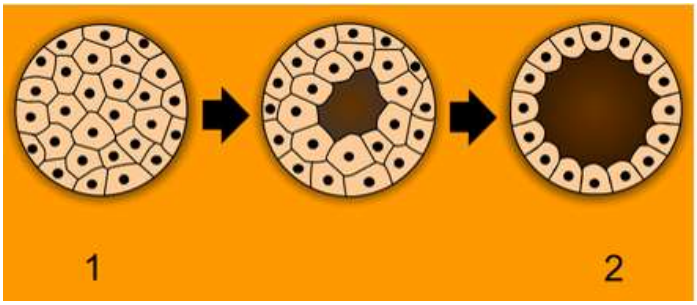
Genetics in embryology and reproductive medicine:

- Preimplantation genetic diagnosis (PGD): Examination of the genetic material of an embryo in the early stages, before it is implanted into the uterus. It helps to identify genetic pathologies and increase the chances of having a healthy child.

- Noninvasive prenatal test (NIPT): A genetic analysis performed on the mother's blood during pregnancy. It allows you to identify fetal chromosomal abnormalities, such as Down syndrome, without harming the embryo.

4.Illustrative material: Overview





Fragmentation of the zygote **1) Morula, 2) Blastula (inside the blastocele = space)**

5.References: see appendix 1

6. Control questions:

1. What is the essence of the embryonic period of development? - The embryonic period begins with the formation of a zygote and ends with the birth or exit from the egg or germ membranes of a young individual. It consists of four stages: crushing, gastrulation, neurulation and organogenesis. CRUSHING is the initial stage of development of a fertilized egg.

2. What role does genetics play in fetal development? - During embryonic development, various genes are activated that help cells grow and form various tissues and organs of the body. However, when certain genes do not function properly, it can disrupt normal development and lead to birth defects.

3. What is the main function of the embryonic period? - The second stage, called the embryonic stage, involves the early growth of the embryo, including the formation of a neural tube that will eventually turn into a brain and spinal cord. The fetal period is the longest stage of prenatal development, during which organs, tissues, and the entire body grow.

4. What is the essence of the embryonic period of development? - The embryonic period lasts from fertilization to the 56th day of development (8 weeks), during which time the developing human body is called an embryo, or embryo. The period from the 9th week of development to birth is called fetal in medical terminology, and the intrauterine developing organism is referred to by the term fetus.

5. What happens during embryonic development? - During embryogenesis, fertilization, embryo fragmentation, implantation, gastrulation (formation of germ layers), organ formation, and placentation occur.

Lecture No. 4

1. Topic Fetal stage of human embryogenesis

2. Goal: the final formation of all organs and systems of the body, achieving the size and physiological maturity of the fetus necessary for life outside the womb, and preparing for birth.

3. Lecture notes: The fetal stage of human embryogenesis begins at the 9th week of pregnancy (after the end of the 8th week) and lasts until birth. At this stage, it is not the formation of new organs that takes place, but the development of already established rudiments of organs and systems. There is active growth, an increase in size, the formation of functional body systems and the final acquisition of human features.

Main characteristics of the fetal stage:

Development of organs and systems: Organs and systems develop from the rudiments formed during the embryonic period, becoming more functional.

Active growth and differentiation: The fetus rapidly increases in size, fat deposits form, and the brain becomes more complex.

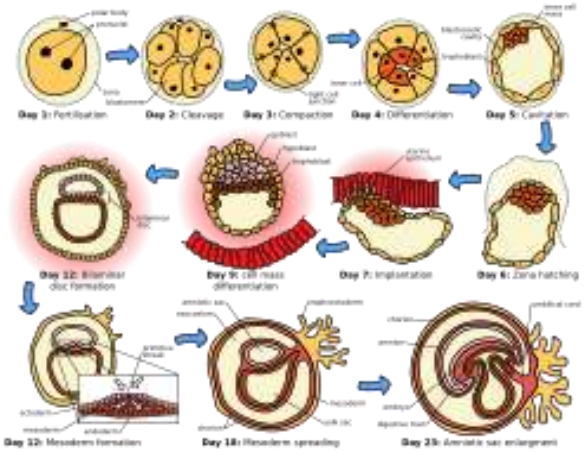
The formation of functional systems: The kidneys, liver, and pituitary gland begin to function, and a connection is established between them and the hypothalamus.

Recognizable appearance: The fetus acquires features similar to a human baby: shoulders appear, arms and legs develop, and external genitalia are formed.

The end of the embryonic period: In the last week of the embryonic period, the primary formation of the main body systems is completed.

Transition to an independent life: By the end of the fetal stage, systems are formed that ensure life outside the mother's womb, for example, the lungs are developed to a level that allows them to survive in artificial conditions.

4.Illustrative material: Overview



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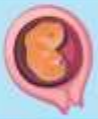
Начальный	1 - я неделя	
Зародышевый	2 - 8 - я недели	
Плодный	С 9 - й недели до рождения	

The initial stages of human embryogenesis are periods of embryonic development

5. Literature:

Appendix 1.

Main:

1. Golichenkov, V.A. Embryology [Text]: textbook / V.A. Golichenkov, E.A. Ivanov, E.N. Nikeryasova. – M.: Publishing Center "Academy", 2004. – 224 p.

2. Danilov, R.K. Histology, embryology, cytology [Text]: textbook / R.K. Danilov, T.G. Borovaya. – M.: GEOTAR-Media, 2018. – 518 p.

3. Studenikina, T.M. Embryology [Text]: textbook / T.M. Studenikina, B.A. Sluka. – 2nd ed., revised and additional – Minsk: "Harvest", 2009. – 304 p.

4. Danilov, R.K. General and medical embryology [Text]: textbook / R.K. Danilov, T.G. Borovaya. Saint Petersburg: SpetsLit, 2003– 231 p

. 5. Baranchugova, L.M. Human Embryology [Electronic resource] / L.M. Baranchugova, M.A. July, A.V. Pateyuk. Chita: CHGMA Publishing House, 2015. 117 p.: Access mode: https://www.booksup.ru

6. Kolesnikov, L.L. Terminologia Embryologica. International terms on human embryology with the official list of Russian equivalents [Electronic resource] / L.L. Kolesnikov, N.N. Shevlyuk, L.M. Yerofeyeva – M.: GEOTAR-Media, 2014. – 417 p.: Available at: http://www.studentlibrary.ru

7. Histology, embryology, cytology [Electronic resource]: textbook / Yu.I. Afanasyev, N.A. Yurina, E.F. Kotovsky et al.; edited by Yu.I. Afanasyev, N.A. Yurina. – 6th ed., revised and add. – M.: GEOTAR-Media, 2018. – 800 p.: Access mode: http://www.studentlibrary.ru

8. Histology, embryology, cytology [Electronic resource]: textbook for universities / edited by E.G. Ulumbekov, Yu.A. Chelyshev. – 3rd ed. – M.: GEOTAR-Media, 2012. – 480 p.: Access mode: http://www.studentlibrary.ru

Additional information:

1. Study Genetics. Textbook for universities/Edited by Academician of the Russian Academy of Medical Sciences V.I. Ivanov, Moscow: ICTS "Akademkniga", 2011-638s.: ill.

2. W. Klug, M. Cummings. Fundamentals of Genetics– Moscow: Technosphere, 2009.

3. Kurchanov.A. Human genetics with the basics of general genetics: textbook. manual -St. Petersburg, 2009.

6. Control questions:

1. Does human embryogenesis occur in the fetal period? - During embryogenesis, fertilization, embryo fragmentation, implantation, gastrulation (formation of germ layers), organ formation, and placentation occur. Therefore, embryogenesis and the first trimester of pregnancy in general, when the main vital systems of the future person are formed, is considered important.

2. The germ stage? – Fertilization – Cleavage - Blastocyst formation: compactification and cavitation – Implantation - Embryonic disc

3. The main stages of fetal development? - Intrauterine stage of development (from conception to birth – 40 weeks)

- Germinal (or the germinal period itself) ...

- The implantation period is 2 days long. ...

- Embryonic (embryonic) – duration of 2 weeks...

- Fetal (fetal) – lasts from the 9th to 40-42 weeks. ...

- The intranatal stage of development.