MEDICAL BIOLOGY FOR INTERNATIONAL STUDENTS 1ST YEAR

PRACTICAL BOOK

Minsk BSMU 2016

МИНИСТЕРСТВО ЗДРАВООХРАНЕНИЯ РЕСПУБЛИКИ БЕЛАРУСЬ БЕЛОРУССКИЙ ГОСУДАРСТВЕННЫЙ МЕДИЦИНСКИЙ УНИВЕРСИТЕТ КАФЕДРА БИОЛОГИИ

МЕДИЦИНСКАЯ БИОЛОГИЯ для иностранных студентов 1-го года обучения

MEDICAL BIOLOGY FOR INTERNATIONAL STUDENTS 1ST YEAR

Практикум

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Минск БГМУ 2016

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Включены основные термины и понятия, закрытые и открытые тесты для самоконтроля, тексты задач по цитологии, генетике, паразитологии и эволюции систем органов хордовых животных, схемы и контуры рисунков, оригинальные фотографии изучаемых препаратов, контрольные и экзаменационные вопросы. Первое издание вышло в 2015 году.

Предназначен для студентов 1-го курса медицинского факультета иностранных учащихся, обучающихся на английском языке.

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Current marks

Name of the student		(I semester)
		 ` /

No	Topic of practice	mark	Teacher's signature
1.	Human in the system of nature. Methods of studying cells		
2.	Biology of the cell. The flow of substance and energy in the cell		
3.	Temporal organization of the cell		
4.	Fundamentals of cytogenetic		
5.	Organization of hereditary material (I)		
6.	Organization of hereditary material (II)		
7.	Genetic engineering		
8.	Control practice in cytology and molecular biology		
9.	Inheritance regularities. Interaction of genes		
10.	Genetic linkage		
11.	Variation		
12.	Biology and genetics of sex		
13.	Fundamentals of human genetics (I)		
14.	Fundamentals of human genetics (II)		
15.	Control practice in genetics		
16.	Human genetic and chromosomal diseases		
17.	Genetic counseling		
18.	Control practice in cytology, molecular biology and genetics		

Current marks

Name of the student ______ (II semester)

No	Topic of practice	mark	Teacher's signature
1.	Reproduction of organisms		
2.	Fundamentals of ontogenesis (embryonic development)		
3.	Fundamentals of ontogenesis (post-embryonic development)		
4.	Introduction to parasitology		
5.	Phylum Sarcomastigophora. Classes Sarcodina, Zoomastigota		
6.	Phylum Infusoria. Class Ciliata. Phylum Apicomplexa. Class Sporozoa		
7.	Phylum Plathelminthes. Class Trematoda		
8.	Phylum Plathelminthes. Class Cestoda		
9.	Phylum Nemathelminthes. Class Nematoda (I)		
10.	Phylum Nemathelminthes. Class Nematoda (II)		
11.	Phylum Arthropoda. Class Arachnida		
12.	Phylum Arthropoda. Class Insecta (I)		
13.	Phylum Arthropoda. Class Insecta (II). Solving situational problems		
14.	Diagnosis of parasitic micropreparations		
15.	Control practice in parasytology		
16.	Evolution of organ systems (I)		
17.	Evolution of organ systems (II)		
18.	Toxic animals		

DEMANDS	OF THE	RIOL	$\cap GV$	DEPARTMENT	\mathbf{ON}	THE	STUDENTS:
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- 1. Observe the safety rules in the classrooms of the department (the safety instructions have been carried out), obey internal regulations of the Belarusian State Medical University.
- 2. Do not come late to the practical classes. Students who are late for practical classes are not admitted.
- **3.** Coming to the class, students must have **gowns**, **practical books**, **pencils**. Students who do not have gowns and practical books **are not admitted** to the classes.
- 4. Missed classes must be fulfilled within 2 weeks.
- 5. Students who have not fulfilled the missed classes within 2 weeks are not admitted to the further classes, colloquiums, and examination without dean's permission.
- **6.** Students with average marks for the year (except colloquiums) **lower than 4.0** who got a **poor mark** at the examination can retake the examination only **at the end of** August.
- 7. Students with average marks for the year 8.25 and higher (under condition that they pass all colloquiums with marks "8", "9" and "10") may be examined only for micropreparations and problems. If the task is done successfully, the student obtain a "ten".

I have read the demands of the department:	201	(signature
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CONTROL QUESTIONS	5. Immersion –
 Organization levels of living things. Properties and characters of livin things. Methods of studying living things (methods of biological sciences). The significance of Biology for medicine. 	6. Objective lens –
4. The position of the human in the animal world system.	7. Ocular lens –
5. Humans as biological and social beings.6. The subject, tasks and methods of cytology.7. Magnifying devices and their purpose. Arrangement of a light microscope.	8. Resolution –
8. Rules of working with the microscope.	9. Revolving nosepiece –
BASIC TERMS AND CONCEPTS	
1. Phylogenetic tree –	10. Self-regulation –
2. Condenser –	11. Self-renewal –
3. Cremaliera –	12. Self-reproduction –
4. Draw-tube –	13. Systemic position of Homo sapiens –

_201___ year

Practice 1. Topic: HUMAN IN THE SYSTEM OF NATURE. METHODS OF STUDYING CELLS

- **1. Organization levels of living matter are:** a) molecular-genetic and cellular; b) tissue and colonial; c) subcellular and siphon; d) organism, biospheric and colonial; e) population-specious and biogeocenotic.
- **2.** The substrate of life is the: a) complex of proteins and carbohydrates; b) complex of proteins and fats; c) complex of fats and carbohydrates; d) complex of fats and nucleic acids; e) complex of proteins and nucleic acids.
- **3.** Living things as the open systems are characterized by: a) substance exchange with the environment; b) absence of substance exchange with the environment; c) energy exchange with the environment; d) absence of energy exchange with the environment; e) information exchange with the environment.
- **4.** The human as the biological being is characterized by: a) heredity and variation; b) public mode of life; c) struggle for existence; d) metabolism, mentality and consciousness; e) presence of speech.
- **5.** The human as the social being is characterized by: a) heredity, variation and mentality; b) presence of speech and public mode of work; c) metabolism, growth, development and ability to work; d) growth, development and ability to work; e) public mode of life and consciousness.
- **6.** The human has following attributes of the class mammals: a) primary body cavity and teeth differentiation; b) mammary glands and diaphragm; c) hair covering and left-sides aortic arch; d) diaphragm and right-sides aortic arch; e) dextral arch of the aorta and intrauterine development.
- **7.** The human has following attributes of the order primates: a) nails; b) binocular eyesight; placenta; c) hairy integument; d) opposition of a thumb to a palm; e) arms of "catching" type; teeth differentiation.
- **8.** Species attributes of Homo sapiens (a reasonable man) are: a) high brain development; b) mentality and consciousness, straight walking; c) presence of hairy integument and nails; d) arms of "catching" type and straight walking; e) opposition of a thumb to a palm.

- 1. The ability to change the parameters of vital activity according to environmental changes is ...
- **2.** Interactions of populations of various species occur at ... the level of life organisation.
- **3.** The quadratic weight parameter of the human brain is ...
- **4.** Homo sapiens belongs to the family ...
- 5. Homo sapiens belongs to the subclass ...

Table 1. Times of taxonomic group's divergence that is generally accepted in the molecular evolution

Group	Periods (mln years)
LANCELETS	564
ACTINOPTERYGIANS	450
AMPHIBIA	360
REPTILES	330
BIRDS	310
GNAWING ANIMALS	110
SOLID-HOOFED ANIMALS	92
LAGOMORPHS	91
CHIMPANZEE / HUMAN	5,5

RULES OF WORKING WITH A SMALL MAGNIFICATION (7×8) MICROSCOPE

- 1. Put the microscope at the distance approximately a palm width from the table edge; set the column towards yourself and the mirror towards the light origin.
- 2. Set the objective lens to 2-3 cm from the surface of the stage by the macrometric knob.
- 3. Check the adjustment of the objective with small magnification (8×) until it 'clicks', it should be fixed opposite the aperture on the stage.
- 4. Put the condenser into a neutral position and open the diaphragm completely.
- 5. Looking into the ocular, direct the mirror surface to the light source for | 5. Looking into the ocular, turn slightly a macrometric screw "towards" good illumination.
- 6. Place the micropreparation on the stage, the cover glass should be directed towards the objective!
- 7. Looking on the side (!), lower the objective 0.5 cm from the surface of the cover glass with a macrometric screw (the focal distance of the objective with $8 \times$ is about 1 cm).
- 8. Looking into the ocular, rotate the macrometric screw towards "yourself" slowly (!) to get a clear image of the object.
- 9. Study the object. Move the preparation manually.

Note: If the object is too small and is not seen at small magnification, then you may set the microscope to an edge of the cover glass. Having obtained a clear image of the glass edge, move it further to a working field searching the object.

RULES OF WORKING WITH A LARGE MAGNIFICATION (7×40) MICROSCOPE

- 1. Get a clear object image at small magnification (see above).
- 2. Center the needed area of a micropreparation move it to the center of the field of vision.
- 3. Rotate the objective lens with large magnification (×40) using a revolver until it 'clicks'
- 4. Put the condenser into an upper position. Looking from the side, carefully lower the large magnification objective with the macrometric screw until it touches the surface of the cover glass (the focal distance of 40x objective is approximately 1–2 mm).
- *yourself*" (!) until the object outlines appear.
- 6. Use a micrometric screw for getting a better image turning it towards yourself or from yourself.
- 7. Study the needed area of the micropreparation.

TERMINATING THE WORK WITH THE MICROSCOPE:

- 1. Having finished studying the object, raise the draw-tube 2–3 cm with a macrometric screw and take off the preparation off the stage.
- 2. Set a small magnification objective until it 'clicks' by turning the revolver and fix it against the aperture on the stage.
- 3. Lower the objective to the stage level with a macrometric screw.

PRACTICAL WORK

Task 1. Make indications at the picture

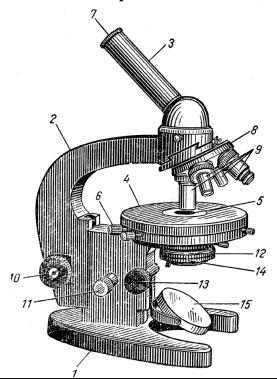


Fig. 1. Microscope
1 - 9 2 - 10 3 - 11 4 - 12 5 - 13 6 - 14 7 - 15 -

Task 2. Study preparations, color pictures and make indications



Fig. 2. Fragment of the fly's wing (7×8)

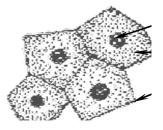


Fig. 3. Frog skin epithelial tissue:

1 — membrane, 2 — cytoplasm, 3 — nucleus

Teacher's signature

Practice 2. Topic: BIOLOGY OF THE CELL. THE FLOW OF SUBST	ANCE AND ENERGY IN THE CELL201 year
Purpose of the practice : to study distinguishing features of prokaryotic a electron-diffraction photographs	and eukaryotic cells, anabolic and catabolic systems of the cell, to analyze
CONTROL QUESTIONS	5. Enzymes of Krebs cycle –
 Present state of the Cell Theory. Distinguishing features of pro- and eukaryotic cells. Structure (models) of plasma membrane, its properties and functions. Ways of passing substances into the cell. Anabolic system of the cell. Catabolic system of the cell. Energy exchange in the cell. Enzymatic systems of mitochondria. 	6. Enzymes of oxidative phosphorylation –7. Enzymes of tissue respiration –
BASIC TERMS AND CONCEPTS 1. Concentration gradient –	8. Mesosomes –
2. Glycocalix –	9. Nucleoid –
3. Glycolysis –	10. Peroxisomes –
4. Glyoxysomes –	11. Plasmalemma –

- **1. Properties of elementary membrane are:** a) plasticity; b) impermeability and fluidity; c) semi-permeability; d) elasticity; e) self-locking.
- 2. Transport of substances into the cell that require ATP energy is:
 a) transport of ions into the cell according to the concentration gradient;
 b) phagocytosis; c) pinocytosis and diffusion; d) osmosis and endocytosis;
 e) transport of substances into the cells against the concentration gradient.
- **3.** Organelles of the cell anabolic system are: a) mitochondria and endoplasmic reticulum; b) ribosomes and Golgi complex; c) endoplasmic reticulum; d) lysosomes and peroxisomes; e) glyoxysomes and ribosomes.
- **4. Organelles of the cell catabolic system are:** a) mitochondria; b) ribosomes, glyoxysomes and endoplasmic reticulum; c) endoplasmic reticulum and mitochondria; d) Golgi complex and peroxisomes; e) peroxisomes and lysosomes.
- **5. Ribosomes are located:** a) on membranes of endoplasmic reticulum and in hyaloplasm; b) in hyaloplasm and karyoplasm; c) on internal nuclear membrane and in chloroplasts; d) on external nuclear membrane and in the mitochondria; e) in mitochondrial matrix and lysosomes.
- **6. Functions of the endoplasmic reticulum are:** a) synthesis of proteins; b) DNA synthesis and compartmentalization; c) synthesis of fats and carbohydrates; d) compartmentalization and transport of substances; e) formation of peroxisomes and RNA synthesis.
- **7. Functions of Golgi complex are:** a) sorting, packing and secretion of substances; b) formation of lysosomes and complex organic compounds; c) synthesis of ATP, proteins and glyoxysomes; d) synthesis of cell membranes; e) protein synthesis and substance secretion.
- **8. Functions of mitochondria are:** a) synthesis of specific proteins; b) splitting of proteins into amino acids; c) synthesis of monosaccharides and ATP; d) synthesis of AMP (adenylic acid); e) splitting of organic substances into H₂O and CO₂.
- **9.** Anaerobic stage of energy exchange occurs in: a) intestine; b) cytoplasm and mitochondria; c) cytoplasm and endoplasmic reticulum; d) cytoplasm; e) Golgi complex and cell nucleus.

- **1.** The ability of biological membranes to divide cytoplasm of the cell is called ...
- **2.** The receptor apparatus located on the outer surface of a plasmalemma is called ...
- 3. Microfilaments of cytoskeleton have the diameter ... nanometers.
- **4.** ER (endoplasmic reticulum) and ... form the transport system of the cell.
- **5.** Conversion of fats into carbohydrates in a plant cell occurs in ...
- **6.** Glyoxysomes are formed in ...
- 7. The destruction of cell organelles by its own lysosomes is called...
- **8.** Integral proteins in the outer mitochondrion membrane forming pores and providing permeability of membranes are called ...
- **9.** The large subunit of ribosomes contains 40–50 molecules of proteins and ... molecules of r-RNA.
- 10. The efficiency of the anaerobic stage of energy exchange is ... %.

PRACTICAL WORK

Task 1. Fill the table

Structure	ER	Ribo- somes	Golgi complex	Lysosome	Mitochon- dria
Membrane					
Cisterns					
2 membranes					
Vesicles					
Cristae					
Hydrolyzing					
enzymes					
ATP-some					
Subunits					

Task 2. Study the diagram, make indications

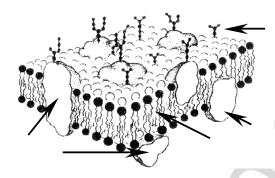


Fig. 1. Elementary membrane constitution scheme:

1 — lipids, 2 — integral protein, 3 — semi-integral protein, 4 — peripheral proteins, 5 — glycocalyx

Task III. Study electron-diffraction photographs and make indications

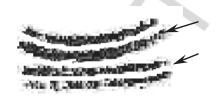


Fig. 2. Elementary membrane electron-diffraction photograph:
1 — protein layer, 2 — lipid layer

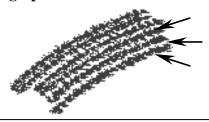


Fig. 3. Electron-diffraction photograph of a rough endoplasmic reticulum:

1 — membrane, 2 — canal, 3 — ribosomes



Fig. 4. ATP-some on the mitochondrion cristae:
1 — inner membrane,
2 — ATP-some

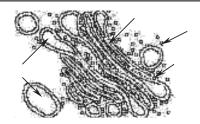


Fig. 5. Electron-diffraction photograph of a Golgi complex:

1 — membrane, 2 — canal, 3 — cistern,

4 — lysosome, 5 — vesicle

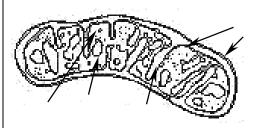


Fig. 6. Electron-diffraction photograph of a mitochondrion:

1 — outer membrane, 2 — inner membrane, 3 — matrix, 4 — cristae,

5 — ribosomes

Teacher's signature

Practice 3. Topic: TEMPORAL ORGANIZATION OF THE CELL

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Purpose of the practice: to study the microscopic and submicroscopic structure of the cell nucleus, cell cycle and principles of interphase, types of cell division, to know how to write down the content of genetic material in different interphase periods and in different stages of mitosis and meiosis.

1	
CONTROL QUESTIONS	5. Crossing-over –
1. The structure and functions of the nucleus.	
2. Types of chromosomes. The structure of a metaphase chromosome.	
3. Cell and mitotic cycles.	6. Chiasms –
4. Interphase, characteristic of periods. Reasons of mitosis.	
5. Characteristic and significance of mitosis.	
6. Characteristic and significance of meiosis.	7. Chromatin –
7. Amitosis.	
BASIC TERMS AND CONCEPTS	
1. Bivalents –	8. Meiosis –
2. Karyolymph –	9. Mitotic cycle –
3. Cell cycle –	10. Nuclear-cytoplasmic ratio –
4. Synapsis of chromosomes –	11. Telomeres of chromosomes –

- 1. Processes that take place in the cell during the pre-synthetic period of interphase are: a) synthesis of RNA, proteins and enzymes; b) synthesis of DNA, RNA, proteins and ATP; c) ATP synthesis and cell growth; d) accumulation of DNA nucleotides, synthesis of proteins of a division spindle; e) synthesis of DNA, RNA and proteins of a division spindle.
- 2. Processes that take place in the cell during the post-synthetic period of interphase are: a) synthesis of DNA and enzymes; b) synthesis of DNA, r-RNA, cell growth; c) ATP synthesis; d) accumulation of DNA nucleotides; e) synthesis of proteins of a division spindle.
- 3. The content of genetic material in the cell at the end of synthetic period of interphase is: a) 1n 1chr 1c; b) 1n 2chr 2c; c) 2n 1chr 2c; d) 2n 2chr 4c; e) 1n 4chr 4c.
- 4. **Reasons of mitosis are:** a) increase of nuclear-cytoplasmic ratio; b) decrease of nuclear-cytoplasmic ratio; c) replication of DNA and «wound hormones»; d) «wound hormones» and mitogenetic rays; e) impairment of karyolemma's integrity.
- 5. The content of genetic material in the cell during the telophase of mitosis is: a) 1n 1chr 1c; b) 1n 2chr 2c; c) 2n 1chr 2c; d) 2n 2chr 4c; e) 1n 4chr 4c.
- 6. Cells that divide by mitosis are: a) somatic cells; b) cells of gonads; c) gametogoniums; d) tumor cell; e) cells of regenerating tissues.
- 7. Cells that divide by amitosis are: a) somatic and old cells; b) cells of gonads and embryo; c) gametogoniums; d) tumor cells; e) cells of regenerating tissues.
- 8. **Cells that divide by meiosis are:** a) somatic and old; b) cells of gonads and embryo; c) gametocytes; d) tumor cells; e) cells of regenerating tissues.
- 9. Content of cell genetic material in meiosis I prophase is: a) 1n 1chr 1c; b) 1n 2chr 2c; c) 2n 1chr 2c; d) 2n 2chr 4c; e) 1n_{biv} 2chr 2c.
- **10.Processes that take place in the cell during the telophase of meiosis I are:** a) spiralization of chromatin and dissolution of nucleolus; b) despiralization of chromosomes and formation of nucleolus; c) formation of karyolemma; d) synapsis and crossing-over; e) cytokinesis.

- 1. The nuclear lamina consists mostly of proteins that are called ...
- **2.** A protein complex at the site of primary constriction of a chromosome which provide attachment of division spindle fibers is called ...
- 3. A secondary constriction site of a chromosome is also called ...
- **4.** The content of genetic material in a cell during the G_2 period is ...
- 5. The content of genetic material in a cell during the diplotene period is ...
- **6.** The content of genetic material in a cell during the diakinesis period is ...
- 7. The content of genetic material in a cell during the pachitene period is ...
- **8.** Bivalents are bound together only by sites called ... during the diplotene of the meiosis I prophase.
- **9.** ... are observed in the equatorial plate during the metaphase of meiosis I.
- **10.** The content of genetic material in a cell during the metaphase of meiosis II is ...

PRACTICAL WORK

Task I. Study the diagrams, electron-diffraction photographs, make indications.

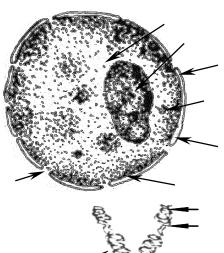


Fig. 1. Electron-diffraction photograph of the nucleus:

1 — external membrane, 2 — internal membrane, 3 — perinuclear space,

4 — pore; 5 — nucleoplasm,

6 — chromatin, 7 — nucleolus

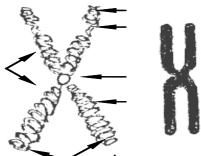






Fig. 2. Make indications in schemes of metaphase chromosome structure (A) and chromosome types (B):

1 — arm, 2 — centromere, 3 — secondary constriction, 4 — satellite, 5 — chromatid, 6 — telomeres, 7 — metacentric chromosomes, 8 — submetacentric chromosomes, 9 — acrocentric chromosomes

Task II. Solve the problem:

Genes which might become active in the G_2 period remained inactive. Will it affect the process of mitosis?

Task III. Fill the table

Fill the formulas of genetic material content during the different periods of mitotic cycle, stages of mitosis and meiosis

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Phases and periods		Interphase	Mitosis	Meiosis I	Meiosis II	
I.	Presynthetic					
II.	Synthetic					
III.	Postsynthetic					
	A. Prophase					
	• leptotene					
	 zygotene 					
	 pachytene 					
	 diplotene 					
	 diakinesis 					
- 4						
	B. Metaphase					
	C. Anaphase					
	D. Telophase					

Task IV. View the micropreparation, using the high magnification of the microscope, make a sketch and indications

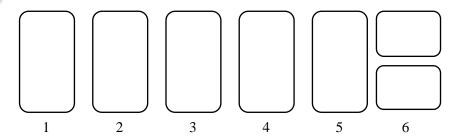


Fig. 3. Mitosis in the cells of onion roots (7×40) :

1 — interphase, 2 — prophase, 3 — metaphase, 4 — anaphase, 5 — telophase, 6 — daughter cells, 7 — chromosomes

Teacher's signature

Practice 4. Topic: FUNDAMENTALS OF CYTOGENETICS

201 year

Purpose of the practice: to study human karyotype and to know how to make an ideogram of normal human karyotype; to group chromosomes according to the Denver classification.

CONTROL QUESTIONS

- 1. The concept of karyotype and ideogram.
- 2. Methods of studying the human karyotype.
- 3. Denver and Paris classifications of human chromosomes.

BASIC TERMS AND CONCEPTS

- 1. Autosomes –
- 2. Centromeric index (CI) –
- 3. Colchicine –
- 4. Karyotype –
- 5. Phytohaemagglutinin –
- 6. Sex chromosomes –

TESTS FOR SELF-CONTROL

- **1. Karyotype is:** a) haploid complement of chromosomes; b) chromosome complement of a somatic cell; c) chromosome complement of a gonad cell; d) diploid complement of chromosomes; e) set of genes in diploid chromosome complement.
- **2. Ideogram is:** a) non-systematized karyotype; b) systematized karyotype; c) location order of genes in a chromosome; d) location order of nucleotides in a gene; e) chromosomes of a karyotype arranged by decreasing of their size.
- **3. Denver classification of human chromosomes considers:**a) size of chromosomes; b) presence of a centromere; c) coloration peculiarities of chromosomes; d) centromeric index; e) number of chromatids.
- **4. Centromeric index is:** a) number of chromosome centromeres; b) length ratio of the short arm and long arm of the chromosome; c) ratio of the short arm's length and all length of the chromosome; d) length ratio of the long arm and short arm of the chromosome; e) ratio of the long arm's length and all length of the chromosome.
- **5. Paris classification of human chromosomes takes considers:** a) size of telomere; b) number of chromatids; c) coloration peculiarities of chromosomes; d) centromeric index; e) presence of secondary constriction and satellites.
- **6.** According to the Denver classification, chromosomes of the group A are: a) large submetacentric; b) small submetacentric; c) small metacentric; d) large metacentric; e) small acrocentric.
- **7.** According to the Denver classification, chromosomes of the group B are: a) large submetacentric, (CI) 24–30; b) small submetacentric, (CI) 24–30; c) small metacentric, (CI) 27–35; d) large metacentric, (CI) 34; e) small acrocentric, satellite.

- **8.** According to the Denver classification, chromosomes of the group C are: a) large submetacentric, (CI) nearby 15; b) submetacentric of moderate size, (CI) 27–35; c) small metacentric, (TSI) 36–46; d) large metacentric, (CI) 27–35; e) small acrocentric, (CI) 13–33.
- **9.** According to the Denver classification, chromosomes of the group D are: a) large submetacentric, (CI) 27–35; b) small metacentric, (CI) 13–33; c) large metacentric, satellite; d) acrocentric of moderate size, (CI) nearby 15; e) small acrocentric, (CI) nearby 15.
- **10.** According to the Denver classification, chromosomes of the group E are: a) large submetacentric; b) small submetacentric; c) small metacentric; d) large metacentric, X-chromosome; e) small acrocentric.
- **11.** According to the Denver classification, chromosomes of the group F are: a) large submetacentric, (CI) 36–46; b) small submetacentric, (CI) 36–46; c) small metacentric, (CI) 13–33; d) large metacentric, (CI) 34, satellites; e) small acrocentric, (CI) 13–33.
- **12.** According to the Denver classification, chromosomes of the group G are: a) large submetacentric; b) small submetacentric and Y-chromosome; c) small metacentric, (CI) 13–33; d) large metacentric, (CI) 26–40; e) small acrocentric.

- **1.** Classification of chromosomes based on the methods of their differential staining is called ...
- **2.** Classification of chromosomes based on the size of chromosomes, their form and the position of centromere is called ...

- **3.** The difference in the size, genes, location of centromere between pairs of chromosomes is called the rule of ...
- **4.** According to the Denver classification, chromosomes of a medium size with the centromere index 27–35 belong to the group ...
- **5.** According to the Denver classification, the Y-chromosome belongs to the group
- **6.** According to the Denver classification, group D includes chromosome pairs number ...
- 7. Chromosomes with secondary constrictions are called ...
- **8.** According to the Denver classification, X-chromosome belongs to the group....
- **9.** Make a record of gene localization: long arm of the 1st chromosome, 1st band of the third region.
- **10.** Make a record of gene localization: short arm of the 6th chromosome, 3th band of the 2nd region ...

PRACTICAL WORK ARRANGING HUMAN CHROMOSOMES ACCORDING TO THE DENVER CLASSIFICATION Task I. Study the ideogram of normal human karyotype. **Groups and** Morphologic Centrocharacteristics of pairs of Pairs of chromosomes meric chromosomes chromosomes Index A (1-3) B (4-5) C(6-12, X)17 15 16 18 D (13-15) E (16-18) Y 21 The Denver human chromosome classification. F (19-20) G (21-22, Y) Task II. Analyze the human karyotype and group the chromosomes according to the Denver classification **Conclusion: Teacher's signature**

Practice 5. Topic: ORGANIZATION OF HEREDITARY MATERIAL (I)

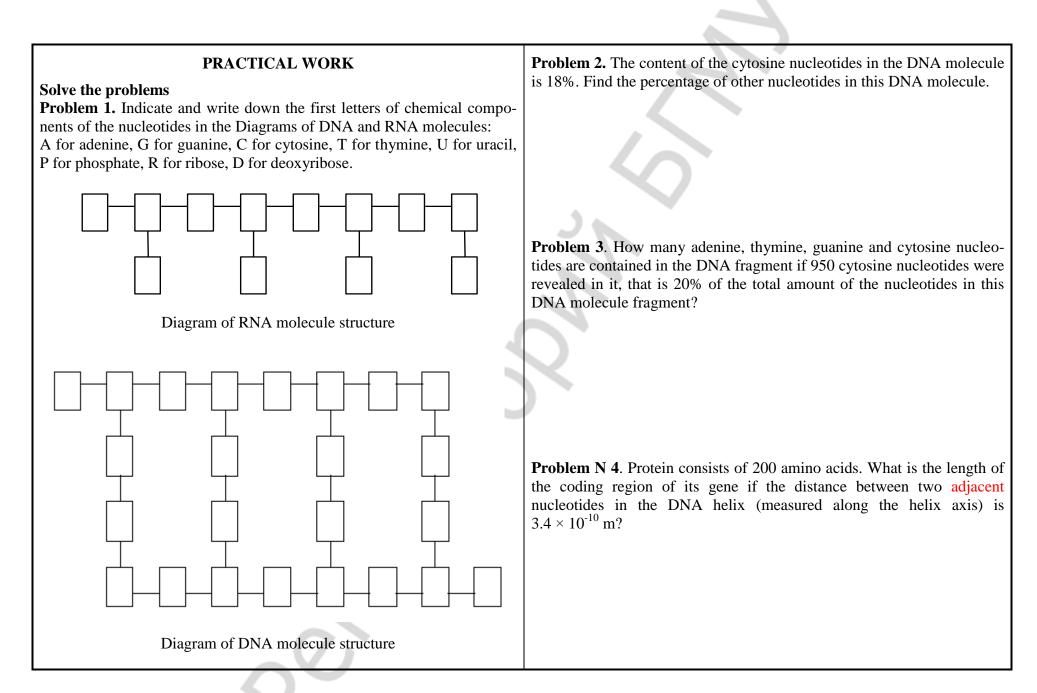
201	vear
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Purpose of the practice: to study the molecular basis of a gene, its properties, to solve problems in the context of DNA and RNA structure, to learn chromosome replication, transcription, translation.

<u> </u>	
CONTROL QUESTIONS	6. Gene –
1. Nucleic acids (DNA and RNA): the structure and functions. Chargaff	
rules.	7. Elongation –
2. Proofs of the nucleic acids role in transmission hereditary information.	
3. Properties of genes.	8. Initiation –
4. DNA replication.	o. Initiation –
5. The genetic code and its properties. Protein biosynthesis.	
6. The central dogma of Molecular Biology.	9. Lability of the gene –
BASIC TERMS AND DEFINITIONS	
1. Avirulent strain –	10. Nucleotide –
1. Avirulent strain –	
	11. Stability of the gene –
2. Anti-codon –	
	12. Termination –
3. Bacteriophage –	12. Termination –
3. Bacteriophage	
	13. Transduction –
4. Codon –	
	14. Transformation –
5. Complementarity of nitrogenous bases –	
2. Complementality of introgenous bases	15. Virulent strain –
	13. VII uiciit Straiii —

- **1. Amount of A+G is equal to amount of:** a) A + T; b) C + T; c) G + T; d) A + C; e) G + C.
- **2.** Bonds between complementary nucleotides in a two-strand DNA are: a) hydrogen; b) covalent; c) phosphodiester; d) peptide; e) disulfide.
- **3. DNA functions are:** a) storage and reproduction of genetic information; b) transport of amino acids to ribosomes; c) transmission of genetic information to daughter DNA molecules; d) transport of amino acids; e) determination of r-RNA synthesis.
- **4. Functions of t-RNA are:** a) storage of genetic information; b) transport of amino acids to ribosomes; c) transmission of the genetic information to daughter t-RNA molecules; d) direct participation in assembling of polypeptides; e) transfer of the genetic information from DNA to the ribosome.
- **5. Gene properties are:** a) stability and lability; b) integrity and pleiotropy; c) integrity, specificity and unambiguity; d) discretion and absence of specificity; e) specificity, tripletness and universality.
- **6. Specificity is the gene property to:** a) mutate; b) determine synthesis of the certain polypeptide; c) be responsible for exhibiting several characters; d) vary the degree of its phenotypic manifestation; e) have different frequency of phenotypic manifestations.
- **7. Pleiotropy is the gene property to:** a) mutate; b) determine synthesis of the certain polypeptide; c) be responsible for exhibiting several characters; d) vary the degree of its phenotypic manifestation; e) have different frequency of phenotypic manifestations.
- **8. Elementary structural unit of a gene is:** a) nitrogenous base; b) pair of complementary nucleotides; c) codon; d) one nucleotide; e) triplet of nucleotides.
- **9. Elementary functional unit of a gene is:** a) one nucleotide; b) pair of complementary nucleotides; c) codon; d) transcripton; e) triplet of nucleotides.
- **10. Heterosynthetic function of a gene is:** a) transcription and replication; b) translation and transcription; c) DNA replication and reparation; d) transformation and translation; e) only translation.

- **1.** The ability of one bacteria culture to absorb and use DNA parts of another bacteria acquiring its properties is called ...
- **2.** The property of bacteriophages to transfer the genetic information from one bacteria to another is called ...
- **3.** ... performed experiments on tobacco mosaic virus and proved the role of nucleic acids in transfering the genetic information.
- **4.** The autosynthetic function of the DNA molecule is its ...
- **5.** The DNA-polymerase can move along the matrix strand from the ... end to the ... end.
- **6.** The direction of the genetic information reading from the 5' to the 3'-end is the property of the genetic code called ...
- 7. The identification process by tRNA of its amino acid is called ...
- **8.** There is the initiating mRNA triplet ... in the peptidylic center of a ribosome during the translation.
- **9.** The process which begins with the first peptide bond formation and ends up with addition of the last amino acid to the polypeptide molecule is called ...
- **10.** Antibiotics are ... of protein biosynthesis.



Correspondence of mRNA codons and amino acids

Second nitrogenous base

_			occond me	ogenous base		
		U	C	A	G	
		Phenylalanine	Serine	Tyrosine	Cysteine	U
	U	Phenylalanine	Serine	Tyrosine	Cysteine	\mathbf{C}
		Leucine	Serine	non	non	A
ě		Leucine	Serine	non	Tryptofane	G
First nitrogenous base		Leucine	Proline	Histidine	Arginine	\mathbf{U}
ns	C	Leucine	Proline	Histidine	Arginine	\mathbf{C}
no		Leucine	Proline	Glutamine	Arginine	A
age		Leucine	Proline	Glutamine	Arginine	G
itr		Isoleucine	Threonine	Asparagine	Serine	\mathbf{U}
t n	A	Isoleucine	Threonine	Asparagine	Serine	\mathbf{C}
irs		Isoleucine	Threonine	Lysine	Arginine	A
1		Methionine	Threonine	Lysine	Arginine	G
		Valine	Alanine	Aspartic acid	Glycine	\mathbf{U}
	G	Valine	Alanine	Aspartic acid	Glycine	\mathbf{C}
		Valine	Alanine	Glutamic acid	Glycine	A
		Valine	Alanine	Glutamic acid	Glycine	\mathbf{G}
						_

Problem 5. One of the DNA chains has the following nucleotide sequence: **GAGGCTCTAGGTACCAGT**

- a) find the sequence of the nucleotides in the complementary chain.
- b) find the order of mRNA codons synthesized using the complementary chain.
- c) find the sequence of the amino acids in the polypeptide encoded by this gene.

Original DNA GAGGCTCTAGGTACCAGT

a

b)

Third nitrogenous base

c)

Problem 6. The part of the protein molecule has the following structure:

Serine – **Lysine** – **Histidine** – **Valine.** How many different variants of the DNA fragment could code for this part of protein molecule?

Teacher's signature

Practice 6. Topic: ORGANIZATION OF HEREDITARY MATERIAL (II)

201	vear

Purpose of the practice: to study properties of genes and their classification, levels of packaging of hereditary material, principles of cytoplasmic heredity, mechanisms of regulation of gene functions and to know how to solve typical problems concerning the regulation of gene functioning.

CONTROL OVINGRIANIS	4.0
CONTROL QUESTIONS	6. Operon –
1. Levels of genetic material packing.	
2. Classification of genes.	
3. Transcription regulation in prokaryotes.	7. Processing –
4. Transcription regulation in eukaryotes (the diagram of G. P. Georgiev).	
5. Cytoplasmic heredity.	8. Promoter –
BASIC TERMS AND CONCEPTS	
1. Exon –	
	9. Repressor –
2. Gene-operator –	
2. Gene-operator –	10. Solenoid –
3. Inductor –	11 Culining
	11. Splicing –
4. Intron –	
	12. Transcripton –
5. Nucleosome –	
3. Ivucicosome	13. Transposon –
	-

- **1. The genes are classified into:** a) structural, modifiers and repressors; b) introns, exons and inhibitors; c) functional and structural; d) co-repressors and operators; e) regulators and intensifiers.
- 2. The role of structural genes is to: a) contain the information about the structure of a protein-repressor; b) contain the information about structure of the proteins-enzymes; c) contain the information about structure of proteins-histones; d) contain the information about RNA structure; e) contain the information about structure of RNA and protein-repressor.
- **3.** The role of functional genes is to: a) contain the information about the structure of a protein-repressor; b) contain the information about structure of the proteins-enzymes; c) contain the information about structure of the proteins-histones; d) contain the information about m-RNA structure, regulate work of structural genes; e) contain the information about r-RNA structure.
- **4.** A transcripton consists of: a) exons and genes-operators; b) genes-operators and genes-regulators; c) structural gene and initiator; d) promotor, terminator and repressor; e) initiator and genes-regulators.
- 5. Pre-mRNA maturation includes processes: a) reading the nucleotides order from one strand of DNA; b) transfer of pre-mRNA in cytoplasm; c) enzymic destruction of non-informative segments of pre-mRNA; d) splicing of exons; e) splicing of introns.
- **6. Intron functions are:** a) regulation of translation and DNA replication; b) regulation of transcription; c) participation in the crossing-over and regulation of translation; d) storage of supply information that provide variability; e) regulation of translation.
- 7. Criteria of cytoplasmic heredity are: a) splitting characters in fillies according to Mendel's laws; b) absence of splitting characters in fillies according to Mendel's laws; c) it is possible to reveal linkage groups; d) inheritance goes on mother's line; it is not possible to reveal linkage groups; e) identical results of recurrent crossings.
- **8. Features of human mitochondrial genome are:** a) circular DNA contains 16 500 pairs of nucleotides; b) circular DNA contains 500 pairs of nucleotides and includes r-RNA genes; c) both strands are transcribed, contains gene of cytochrome b; d) one strand is transcribed; includes r-RNA genes; e) contains information about 22 t-RNA, circular DNA contains 160 pairs of nucleotides.

- **1.** A segment of the DNA molecule together with a histone octamer form ...
- **2.** The length of the DNA molecule decreases by ... times at the first level of the its packaging.
- **3.** The decrease of the DNA length by 10–20 times occurs at the ... level of its packaging.
- **4.** As a result of all packaging levels the DNP molecule is shortened by ... times.
- **5.** ... contain the reserve information providing variability.
- **6.** The genes-regulators carry information for the synthesis of such proteins as ...
- 7. During the structural genes «expression» genes-operators get rid of ...
- 8. The substance which is broken by enzymes coded in the operon is ...
- **9.** Reactions of binding informative pre-mRNA fragments to form mature mRNA is ...
- 10. Leber disease is caused by mutations of ... genes.

PRACTICAL WORK

Task 1. Make indications in the pictures:

5 – 6 –

7 –

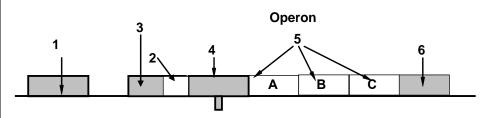


Fig. 1. Structure of an operon

123456-

Transcripton

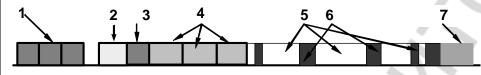


Fig. 2. Structure of a transcripton

Task 2. Solve the problems:

Problem 1. The molecular weight of the phage nucleic acid (consists of one strand) is about 10⁷. How many proteins are encoded in it if its typical protein consists on average of 400 monomers and the molecular weight of the nucleotide is about 300.

Problem 2. Speed of enzymes performing DNA replication is 0.6 mkm/min. How much time will DNA replication take if it consists of 500 replicons? The length of each replicon is 60 mkm.

Problem 3. Let's take the relative weight of one nucleotide as 1. Find the weight of bacterial operon where the promoter and initiator (together) consist of 10 nucleotide pairs, each of operator and terminator consists of 10 nucleotide pairs and each of three structural genes code for a polypeptide that consists of 50 amino acids.

Teacher's signature

Practice 7. Topic: GENETIC ENGINEERING

201 ____ year

Purpose of the practice: to study the principles of genetic engineering and organism cloning, to know how to solve problems in the context of genetic engineering.

engineering.	
CONTROL QUESTIONS 1. Stages of genetic engineering methods.	6. Hybridization of primers –
2. Obtaining of genetic material.	
3. Insertion of DNA fragments into the molecule-vector.	7. Liposomes –
4. Incorporation of the recombinant DNA into the cell-recipient.	7. Exposomes –
5. Using methods of genetic engineering in medicine.	
5. Using methods of genetic engineering in medicine.	8. Phasmids –
BASIC TERMS AND CONCEPTS	6. Fliashius –
1. "Sticky ends" –	
1. Sucky chus –	9. Plasmids –
	9. Plasmids –
2 Autorodiogram	
2. Autoradiogram –	10. P. 4 1 4 1 2 4 2
	10. Restriction sites –
2 Cosmide	
3. Cosmids –	11 D 4 C 4
	11. Restrtictases –
1 DVI	
4. DNA-probe –	
	12. Vector –
5. Genome dactyloscopy –	

- **1. Purposes of genetic engineering are:** a) designing of genetic structures according to a plan; b) decoding of the nucleotide orders of DNA; c) creation of organisms with the new genetic program; d) revealing of linkage groups; sequenation of genes; e) construction of a chromosome genetic map.
- 2. Main stages of genetic engineering are: a) obtaining genetic material; b) construction of a chromosome genetic map; c) decoding of the nucleotide order of a DNA site and building of recombinant DNA; d) selection of the transformed cells; e) incorporation of a recombinant DNA molecules in a chromosome.
- **3. Ways of obtaining genes for transplantation:** a) synthesis of simple genes by chemical reactions; b) synthesis of genes on the base of a protein molecule; c) synthesis of complex genes by reverse transcription; d) making of a genetic map of a chromosome; e) cutting out of genes by restrictases.
- **4.** Recombinant DNA molecules can be received by embedding the gene in: a) protein; b) bacteria plasmid; c) virus genome; d) lipid; e) a bacteriophage genome.
- **5. The enzymes used in gene engineering are:** a) DNA-polymerase; b) lipase and restrictase; c) revertase and restrictase; d) restrictase and amylase; e) ligase.
- **6. Genetic engineering allowed us to receive:** a) the strains of Escherichia coli, capable to synthesize inulin; b) the strain of Escherichia coli, capable to synthesize somatotropinum; c) plants, capable to acquire atmosphere nitrogen; d) microorganisms, capable to synthesize carbohydrates of oil from alimentary proteins; e) antiviral serums.
- 7. The future of gene engineering is based on the following achievements of molecular biology: a) ability to transmit genetic information by sexual way in eukaryotes; b) receiving of modifications with help of chemical mutagens; c) sequenation of genes; d) substitution of defective genes; e) including artificially synthesized genes in the human genome.
- **8.** The chemical basis of plasmids is: a) RNA; b) DNA; c) proteins; d) lipids; e) polysaccharides.

- **1.** Enzymes called ... are used in the genetic engineering for obtaining genes.
- **2.** Enzymes capable of cutting the DNA molecule in certain sites and form «sticky ends» are called ...
- **3.** The method of genes synthesis by reverse transcription is called
- **4.** Bacterial plasmids, phage and viral genome, phasmids and ... can be used as vector molecules in genetic engineering.
- **5.** Hybrid vectors capable of developing both as a phage and as a plasmid are called ...
- **6.** The plasmids containing cos-sites (sticky ends) of phage λ DNA are called ...
- **7.** Size of the DNA fragments which can be cloned in cosmids is about ... thousand nucleotide pairs.
- **8.** The basic vector for the animal genes cloning is the genome of the virus ...
- **9.** Restrictase Eco R I forms ... when cuts the DNA.
- **10.** Restrictase Hind II forms ... when cuts both DNA strands in same places.

Restrictases		
№	Restrictase	Definition sites and DNA cut points
1.	Bal I	5'-T G G C C A-3' 3'-A C C G G T-5'
2.	Bam H I	5' - G' G A T C C - 3' $3' - C C T A G G - 5'$
3.	Eco R I	5'- G A A T T C-3' 3'- C T T A A G-5'
4.	Hind III	5'- A' A G C T T-3' 3'- T T C G A A-5'
5.	Sal I	5'-G' T C G A C-3' 3'- C A G C T G-5'
6.	Xba I	$5'$ - T \checkmark C T A G A-3' 3'- A G A T C \uparrow T-5'

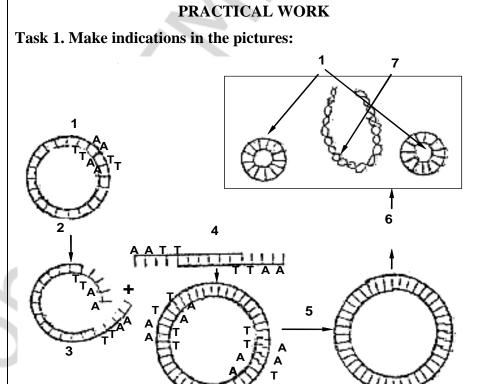
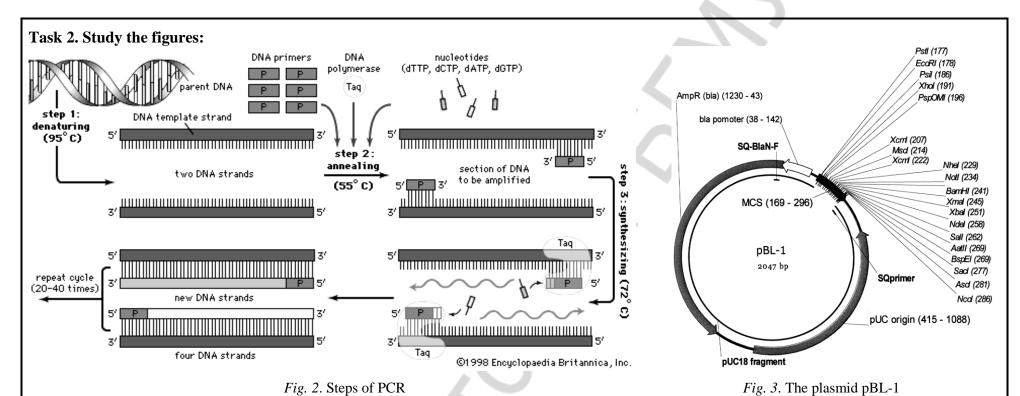


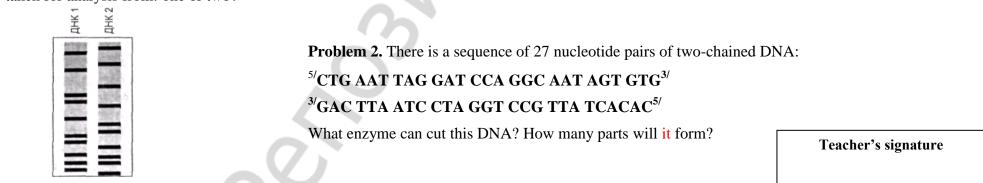
Fig. 1. Scheme of gene incorporation in the plasmid and injection of recombinant DNA into a bacteria

6 – 7 –



Task 3. Solve the problems:

Problem 1. DNA samples treated with restrictases are analyzed with the method genome dactyloscopy using a radioactive probes, complementary to the minisatellite DNA. The scheme of radiogram is shown in the figure. Basing on the spectrum character indicate how many persons the DNA was taken for analysis from: one or two?



Practice 8. Topic: CONTROL PRACTICE IN CYTOLOGY AND MOLECULAR BIOLOGY

_201___ year

Purpose of the practice: to control the student's knowledge of studied material and fix the skills of solving problems in molecular biology

CONTROL QUESTIONS

- 1. Origin of life. Evidence of the organic world evolution.
- **2.** Organization levels of living things. Properties and characters of living things.
- **3.** Methods of studying living things (methods of biological sciences).
- **4.** The significance of Biology for medicine.
- **5.** The position of the human in the animal world system. Humans as biological and social beings.
- **6.** The subject, tasks and methods of cytology. The present state of the cellular theory.
- 7. Differentiating signs of pro- and eukaryotic cells.
- **8.** The structure (models) of elementary membrane, its properties and functions. Methods of passing substances into the cell.
- **9.** Anabolic system of the cell. Catabolic system of the cell.
- 10. Energy exchange in the cell. Enzymatic systems of mitochondria.
- 11. The structure and functions of the nucleus.
- 12. Types of chromosomes. The structure of a metaphase chromosome.
- **13.** Cell and mitotic cycles. Interphase, characteristic of its periods. Reasons of mitosis.
- **14.** Characteristic and significance of mitosis.
- **15.** Characteristic and significance of meiosis.
- 16. Amitosis.

- **17.** The concept of karyotype and ideogram. Methods of studying the human karyotype.
- 18. The Denver and Paris classifications of human chromosomes.
- **19.** Nucleic acids (DNA and RNA): the structure and functions. Chargaff's rules.
- **20.** Proofs of the nucleic acids role in transmission hereditary information.
- 21. Properties of genes. DNA replication.
- 22. The genetic code and its properties. Protein biosynthesis.
- 23. The central dogma of Molecular Biology.
- 24. Levels of packing genetic material. Classification of genes.
- **25.** Transcription regulation in prokaryotes.
- **26.** Transcription regulation in eukaryotes.
- 27. Cytoplasmatic heredity.
- 28. Stages of genetic engineering methods. Obtaining genetic material.
- **29.** Introduction of DNA fragments into the molecule-vector.
- **30.** Insertion of recombinant DNA in the cell-recipient.
- **31.** Using methods of genetic engineering in medicine.

Practice 9. Topic: INHERITANCE REGULARITIES. INTERACTION	OF GENES201 year
Purpose of the practice : to study the inheritance laws of the mono- and posolve standard problems that demonstrate the objective laws of mono- and positive laws of mono- and mono- a	
CONTROL QUESTIONS	4. Genotype –
 Genetics as a science. Basic concepts of Genetics. Peculiarities of the hybridological method. Inheritance regularities in monohybrid cross. Hypothesis of purity of gametes and its cytological foundation. Analyzing crossing. Phenotypical radical. Regularities of inheritance in polyhybrid crossing. The law of independent assortment. Conditions limiting the Mendel's laws. Pleiotropy. Semi-lethal and lethal genes. Intra-allelic interaction of genes. Inheritance of blood groups. Inter-allelic interaction of genes. 	 5. Homozygous organism – 6. Multiple allelism – 7. Phenotypic radical –
BASIC TERMS AND CONCEPTS	
1. Allelic genes –	8. Phenotype –
2. Complementation –	9. Polygenic inheritance –
3. Genome –	10. Super-dominance –

- 1. The main features of G. Mendel's hybridological method are: a) one or of two pairs of alternative alleles are analyzed; b) many alternative alleles are analyzed; c) analysis starts with cross of homozygous organisms; d) several generations are analyzed; e) one generation is analyzed.
- 2. Concepts of the hypothesis of purity of gametes: a) genes of one allelic pair of a hybrid organism are hybridized; b) genes of one allelic pair of a hybrid organism are not hybridized; c) genes of different allelic pairs can be hybridized; d) both allelic genes get in one gamete; e) from each pair of allelic genes one gene gets into gamete.
- 3. The conditions necessary for actuality of Mendel's laws:
 a) codominance; b) semidominance; c) presence of lethal genes;
 d) equiprobable formation of gametes and zygotes of different types;
 e) genes of different allelic pairs are in one chromosome.
- **4. Analyzing cross is performed to reveal:** a) mutations; b) a phenotype of the individual; c) a genotype of the individual with a recessive character; d) a genotype of the individual with dominant character; e) lethal genes.
- **5. Features of incomplete dominance are:** a) a dominant gene does not completely suppress the action of a recessive gene; b) the dominant gene completely suppress the action of a recessive one; c) homo-and heterozygotes are identical phenotypicly; d) homo-and heterozygotes are not identical phenotypicly; e) the dominant gene in a heterozygous state express stronger, than in homozygous.
- **6. Features of co-dominance are:** a) the dominant gene does not completely suppress the action of recessive gene; b) it is a type of interaction of allelic genes, genes are equivalent; c) homo- and heterozygotes are identical phenotypicly; d) it is a type of interaction of non-allelic genes; e) the dominant gene in a heterozygous state express stronger, than in homozygous.
- 7. Features of polymeria are: a) mutual influence of different alleles that occupy adjacent loci of one chromosome; b) 2 dominant genes of different allelic pairs are responsible for a new character; c) 2 recessive genes of different allelic pairs are responsible for a new character; d) one gene is responsible for different characters; e) genes from different allelic pairs have an effect on a manifestation degree of one character.

Fill in the gaps:

- 1. Characters with different qualitative states are called ...
- 2. The second and third Mendel's laws require the gene penetrance ... %.
- **3.** Bombay phenomenon is an example of the genetic interaction which is called ...
- **4.** Phenotypic segregation in ratio 9:7 in crossing diheterozygotes result from interallelic gene interaction called
- **5.** Independent combination of two pairs of allelic genes during an analyzing cross result in phenotypic segregation ... in the first generation of offsprings.
- **6.** Alleles presented in the populations more than in two states are called ...

PRACTICAL WORK

Solve the problems:

Problem 1. How many and what type of gametes would be formed in the organisms with the genotypes:

P: AaBbDd AAbbCCddRR?

Problem 2. Brown color of eyes is dominant human character while blue color is recessive. The parents of a blue-eyed child have got brown eyes. Find the genotypes of all members of the family.

Character	Gene	Genotype
Brown eyes	В	BB; Bb
Blue eyes	b	bb

Problem 3. The allele of brown eyes' color dominate over the allele of blue color and the allele of right-handedness (a habit to use mostly the right hand to perform usual work) dominate over the allele of left-handedness. The genes of both characters are situated in the different chromosomes. A parents are brown-eyed right-handed diheterozygotes. What characters would their children get and what is their percentage ratio?

Problem 4. A woman has blood group I (0), Rh-, MN, her husband has blood group IV (AB), Rh+ (homozygote), N. Which combinations of blood groups by all systems will their children get?

Human's blood group inheritance

Character	Gene	Genotype	Character	Gene	Genotype
AB0 system				MN system	
Group I (0)	I_{O}	$I_{O}I_{O}$	Group M	L^{M}	$L^{M}L^{M}$
Group II (A)	I^{A}	I^AI^A , I^AI^O	Group N	L^{N}	$L^{N}L^{N}$
Group III (B)	I^{B}	I^BI^B , I^BI^O	Group MN	L^{M} and L^{N}	$L^{M}L^{N}$
Group IV (AB)	I^A and I^B	$I^A I^B$			
Rh system					
Rh+	D	DD, Dd			
Rh-	d	dd]		

Problem 5. Angiomatosis of retina is caused by a dominant autosomal gene with the penetrance 50%. What is the probability of giving birth to a sick child if both parents are heterozygous?

Problem 6. Rare gene a causes anophthalmia (the absence of eyeballs). Its allele A is responsible for the normal of eyeballs, but heterozygotes have smaller eyeballs. Find all the phenotypes and genotypes (with %) of all children whose parents have undersized eyeballs.	
Problem 7. Human congenital deafness can be determined by recessive	
genes d and e . Only presence of both dominant alleles (D and E) provides normal hearing. In a family both parents are deaf and their seven children have normal hearing. Find out the genotypes of the parents.	
	Teacher's signature

Practice 10. Topic: GENETIC LINKAGE

Purpose of the practice: to get acquainted with T. Morgan experiments in the linked inheritance, to study the inheritance in cases of autosomal and gonosomal linkage, to know how to write gametes and solve the problems in gene linkage, to compose and analyze chromosome maps

CONTROL QUESTIONS

- 1. Experiments of T. Morgan. Complete and incomplete genetic linkage.
- 2. Autosomal and gonosomal linkage groups.
- 3. Crossing-over, crossover and non-crossover gametes.
- 4. Basic concepts of the chromosome theory of inheritance.
- 5. Maps of eukariotic chromosomes (genetic and cytological).

BASIC TERMS AND CONCEPTS

- 1. A cytological map of a chromosome –
- 2. Crossover gametes –
- 3. Genetic map of the chromosome –
- 4. Linkage of genes –
- 5. Non-crossover gametes –
- 6. Recombinants -

TESTS FOR SELF-CONTROL

201 year

- 1. The phenomenon of genetic linkage is observed when genes of different allelic pairs are situated: a) in the same chromosome; b) in the different chromosomes; c) only in the autosomes; d) only in the X-chromosome; e) only in the Y-chromosome.
- 2. Complete genetic linkage is observed: a) in a female Drosophila and a male silkworm; b) if non-allelic gnes are located in different chromosomes; c) if crossing-over occurs; d) if crossing-over does not occur; e) in a male Drosophila and a female silkworm.
- **3. Incomplete genetic linkage is observed:** a) if genes of different allele pairs are located in one chromosome; b) if non-allelic gnes are located in different chromosomes; c) if crossing-over occurs; d) if crossing-over does not occur; e) in a male Drosophila and a female silkworm.
- 4. The main concepts of the chromosome theory of inheritance are:

 a) allelic genes are located in the linear order in identical locus's of homologous chromosomes; b) allelic genes occupy different locus's of homologous chromosomes; c) the number of linkage groups is equal to monoploid set of chromosomes; d) the number of linkage groups is equal to diploid set of chromosomes; e) between homologous chromosomes of Drosophila male the crossing-over is possible.
- **5. Phenotypic** segregation ratio for monohybrid cross of homozygotes at complete dominance: a) is absent; b) 3:1; c) 1:2:1; d) 9:3:3:1; e) 1:1.
- **6. Phenotypic segregation ratio for incomplete genetic linkage in Morgan's experiences:** a) 3:1; b) 1:2:1; c) 9:3:3:1; d) 1:1; e) 41.5:8.5:8.5:41.5.
- 7. Phenotypic segregation ratio for complete genetic linkage in Morgan's experiences: a) 41.5:8.5:8.5:41.5; b) 3:1; c) 1:2:1; d) 9:3:3:1; e) 1:1.

Fill in the gaps:

- 1. Conditions limitting Mendel's 3rd law are: incomplete penetrance of genes, lethal and semi-lethal genes, unequal formation of different types of gametes and zygoteges, genes' pleyotropy, interaction of genes apart from complete dominance and ...
- 2. If a diheterozygous organism forms only 2 types of gametes, then genetic linkage is...
- 3. If a diheterozygous organism forms 4 types of gametes, then genetic linkage is...
- 4. If crossing-over occur between the genes of a pair of homologous chromosomes, then genetic linkage is...
- **5.** Biological phenomenon breaking the genetic linkage is...
- **6.** The distance between genes measured in morganids is equal to % of ...
- 7. The maximal probability of crossing-over for linked genes is ... %.
- **8.** Individuals formed from crossover gametes are called
- **9.** The number of human's autosomal linkage groups is ...

PRACTICAL WORK

Genetic experiment of T. Morgan:

Gene	Character
В	Grey colour of body
b	Black colour of body
V	Long wings
V	Short (vestigial) wings

Experiment 1.

- P. BBVV x bbvv



BbVv Grey colour of body with Vestigial wings - 100 %

Experiment 2.

P. bbvv x	BbVv
G. (bv)	BV bv
F ₁ . bbvv	BbVv
50%	50%

Experiment 3

- P. BbVv x bbvv
- F₁. BbVv; Bbvv; bbVv; bbvv 41,5% 8,5% 8,5% 41,5%

Solve the problems

Problem 1. How many and what types of gametes are formed in the organism of:

- a) Drosophila with the genotypes:
- **= 2**. Female **= =** 1. Male a b a b
 - 3. Male **=** 4. Female

a b

- b) a man with the genotype
- c) a woman with the genotype $\frac{AB}{ab} \frac{D}{d}$

Problem 2. The human's dominant gene of elliptocytosis (**El**) and the gene that code for the Rh-antigen on the erythrocytes (**D**) are situated in the same autosome at the distance 3 centimorgans. What and how many gametes are produced in the organism of:

a) the woman with the genotype:

b) the man with the genotype:

 $\frac{\text{Eld}}{\text{elD}}$

ElD eld

c) One of the spouses is heterozygous for both characters (Rh+ was heredited from the one parent and the elliptocytosis from the other one). The other spouse has the Rh- and normal erythrocytes. Find out the percentage of the possible genotypes and phenotypes of the children in this family.

Character	Gene	Genotype	Gene location
Rh+	D	D-	
Rh-	d	dd	One autosome
Elliptocytosis	El	El-	Distance D-El = 3 Morgan units
Norm	el	elel	

Problem 3. Genes **L**, **M** and **N** are referred to one linkage group. It was revealed during the experiment that the distance between genes **L** and **M** is 5 centimorgans, and between genes **M** and **N** is 3 centimorgans. Is it possible to find the distance between genes **L** and **N**? During the additional experiment it was revealed that the distance between **L** and **N** is 2 centimorgans. Show the location of the genes **L**, **M** and **N** in the chromosome by a diagram.

Problem 4. While crossing heterozygous female drosophilae with male recessive the following results were obtained:

1) AB : Ab : aB : ab = 25% : 25% : 25% : 25%;

2) AB : Ab : aB : ab = 45% : 5% : 5% : 45%;

3) AB : Ab : aB : ab = 5% : 45% : 45% : 5%.

Which cases belong to the linked inheritance and free segregation? What is the position of these genes in the chromosomes? Find the distance between the genes A and B when possible.

Practice	11.	Topic:	VARIATION
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______201____ year

Purpose of the practice: to study the main forms of variation, its reasons, medical and biological importance, mechanisms of gene, chromosome and genomic mutations, genetic material repair and biological principles of oncogenesis.

CONTROL QUESTIONS	5. Inversion –
1. Variation and its types.	
2. Phenotypic variation. The reaction norm.	
3. Genotypic variation and its types.	6. Reaction norm –
4. Mutagenic factors.	o. Reaction norm –
5. Classification of mutations.	
6. Genome, chromosome and gene mutations.	
7. Stability and repair of genetic material; anti-mutagens.	7. Ring chromosomes –
8. Biological basis of cancerogenesis.	
BASIC TERMS AND CONCEPTS	
1. Reading frame shift –	8. Transgenations –
2. Cancerogenesis –	9. Transitions –
3. Deletions –	10. Translocations –
4. Duplications –	

- **1. The properties of modifications:** a) have adaptive character; b) are inherited; c) are not inherited; d) are the matter for natural selection; e) are the matter for artificial selection.
- **2. Biological mutagens cause:** a) structural defects of genes and chromosomes; b) polyploidy; c) formation of thymine dimers; d) haploidy; e) embedding of its DNA in DNA of the host cells.
- **3.** Characteristic features of gametic mutations are: a) occur in sex cells; b) occur in somatic cells; c) manifest in the individual; d) pass to offsprings by sexual reproduction; e) pass to offsprings by asexual reproduction.
- **4. Types of functional genes mutations:** a) a transposition; b) impairment of the alternation of recognition and terminations; c) impairment of the alternation of initiation and elongation; d) impairment of the alternation of induction and repression; e) transitions.
- **5. Polyploidy is:** a) not multiple of a haploid complement increase of the chromosome number; b) multiple of a haploid complement increase of the chromosome number; c) not multiple of a haploid complement decrease of the chromosome number; d) multiple of a haploid complement decrease of the chromosome number; e) haploid set of chromosomes.
- **6. Haploidy is:** a) a positive mutation; b) nullsomy; c) monosomy; d) absence of one chromosome; e) a haploid set of chromosomes.
- **7. Kinds of structural genes mutations:** a) transductions; b) a transpositions; c) translocations; d) reading frame shift; e) transitions.
- 8. Stages' order of excision repair of a DNA: 1) synthesis of a new DNA strand fragment; 2) ligation of the synthesized strand with the main strand; 3) recognition the damaged DNA strand; 4) cutting out of the damaged DNA fragment; 5) replication of a DNA molecule: a) 1–5–2–3; b) 5–1–3–2; c) 3–4–5–2; d) 3–4–2–1; e) 3–4–1–2.
- **9.** According to the oncogene concept, the basis of carcinogenesis is: a) protooncogenes received from parents or introduced into the cell genome by viruses; b) chromosome mutations of somatic cells; c) presence of protooncogenes in somatic cells of an organism; d) genome mutations of somatic cells; e) incorporations of viral DNA in the genome of somatic cells.

Fill in the gaps:

- **1.** Enzymes capable of cutting out the damaged part of the DNA during the repair are ...
- **2.** Transgenation when one purine base is replaced with another purine base is called ...
- **3.** ... of the terminal parts of chromosomes leads to formation of ring chromosomes.
- **4.** Mutation of ... genes leads to the impairment of alternation of repression and induction of genes.
- **5.** Non-disjunction of chromosomes during the mitosis or meiosis leads to ... mutations.
- **6.** Aneuploidy when only one chromosome of a pair is present in the karyotype is called ...
- **7.** Genome mutation when somatic cells have single chromosome set is called ...
- **8.** Disease caused by the infringement of DNA repair mechanisms and is characterized by insufficiency of red bone marrow functions resulting in deficit of blood cells and hyperpigmentation is called ...

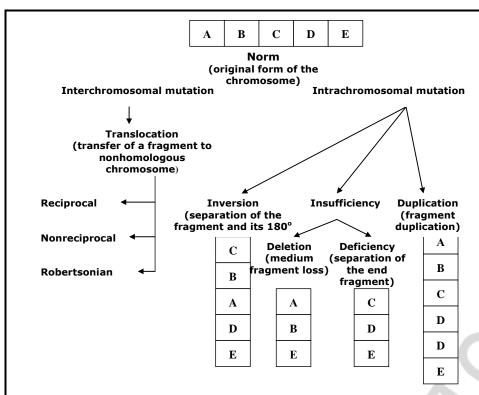


Fig. 1. Change of chromosome structure

Task I. Solve the problems

Problem 1. Some cells of a sick person have normal karyotype, others – 47 or 45 chromosomes. What are the name and possible mechanisms of this phenomenon?

Problem 2. The father has blue eyes, the mother has brown ones and the daughter has one brown eye and the other is blue. How can it be explained?

Task II. Study the preparations of drosophila flies mutations and add the missing elements.

Wings Body color Eyes Yellow Bar Curly Narrow, I chromo-Bend, II chromosome, Yellow, I chromosome, dominant chardominant character, some, recessive acter, chromosome gene mutation character, gene mutation mutation **Vestigial** White **Black** White, I chromosome, Vestigial, II chromo-Black, II chromorecessive character, some, recessive some, recessive gene mutation character, gene character, gene mutation mutation Normal Red eyes, normal wings, grey body color **Teacher's signature**

Practice 12. Topic: **BIOLOGY AND GENETICS OF SEX**

__201___ year

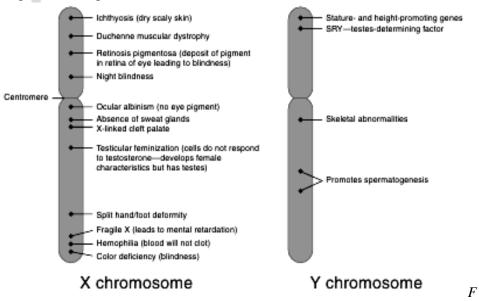
Purpose of the practice: to study the laws of sex inheritance, principles of its differentiation and determination, mechanisms of chromosomal diseases of sex, to know how to solve problems in the context of the characters linked with the X-chromosome and holandric ones

CONTROL QUESTIONS	5. Holandric characters –
1. Sex as a biological character. Sex characters.	
2. Sex-controlled and sex-limited characters.	
3. X-linked and holandric characters.	6. Klinefelter syndrome –
4. Chromosome theory of sex determination.	
5. Peculiarities of sex determination in humans and its impairments.	
6. Sex chromatin.	7. Morris syndrome –
7. Sex chromosome disorders.	
8. Primary, secondary and tertiary ratios of sexes.	
BASIC TERMS AND CONCEPTS	8. Physical sex determinants –
1. Sex-controlled characters –	
2. Sex-limited characters –	9. Shereshevsky-Turner syndrome –
3. X-linked characters –	10. Transsexualism –
4. Hermafroditism –	11. Trisomy X syndrome –

- **1. Formation of gonad primordium proceeds untill the week of embryogenesis:** a) 1st; b) 2nd; c) 3rd; d) 4t^h; e) 5th.
- **2.** The differentiation of donads' primordia into the gonads occurs during the weeks of embryogenesis: a) from 1st to 4th; b) from 4th to 6th; c) from 4th to 8th; d) from 4th to 12th; e) from 10th to 15th.
- **3.** Till 4th week of an embryogenesis, formation of gonad primordiua goes under the control of genes of: a) autosomes; b) one X-chromosome; c) two X-chromosomes; d) Y-chromosomes; e) X-and Y-chromosomes.
- **4.** The differentiation of donads' primordia into the gonads occurs under the control of genes of: a) autosomes; b) one X-chromosome; c) the second X-chromosome; d) Y-chromosomes; e) cytogene.
- 5. In case of absence of the second gonosome in karyotype, gonads:
 a) are differentiated; b) are not differentiated; c) connective tissues are formed on their place; d) partially atrophy; e) completely atrophy.
- **6. Physical abnormality of sex the determination in humans:** a) a genetic gender; b) homosexuality; c) transvestism; d) gametic gender; e) hermaphroditism.
- **7.** Transvestism is a phenomenon, when the person: a) chooses the sexual partner of the other gender; b) chooses the sexual partner of the same gender; c) the sexual satisfaction is reached by wearing clothes of the opposite gender; d) wishes to change his/her gender; e) infertile.
- **8.** The karyotype at Shereshevsky-Turner syndrome is :a) 46, XY, 5p; b) 45, X0; c) 47, XXY; d) 47, XX, 21 +; e) 46, XX, 9p +.
- **9.** The karyotype at Klinefelter syndrome is :a) 47, XXY; b) 45, X0; c) 47, XXX; d) 46, XY; e) 46, XY, 9p +.
- **10. A Barr's body is:** a) an activated Y-chromosome; b) inactivated Y-chromosome; c) activated X-chromosome; d) inactivated X-chromosomes.

Fill in the gaps:

- **1.** Two Barr bodies in the nucleus of a female somatic cell are typical for the ... syndrome.
- **2.** Female phenotype, low position of ears, short neck with a skin fold are typical for the ... syndrome.
- **3.** Men with female phenotype, gynecomastia and impairment of spermatogenesis suffer from ... syndrome.
- **4.** Phenomenon when sexual excitement and satisfaction are reached while wearing clothes of the opposite sex is called ...
- **5.** Human chromosomal diseases of sex result from the impairmnt of the process called.
- **6.** Charactrs determined by genes located in the non-homologous part of the Y-chromosome are called ...
- **7.** Persistent discordance of person's sexual self-conscious and his real genetic and gonad sex is called ...



ig. 1. Sex chromosomes

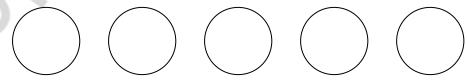
Task I. Solve the problems

Problem 1. An albino woman (autosome recessive character) married a daltonian man (recessive X-linked character). The rest of their genotype is normal. Which combinations of genotypes and characters are possible for their children?

Problem 3. Genes of hemophilia (h) and daltonism (d) are located in the X chromosome at the distance of 10 centimorgans. A woman whose father is sick with both diseases and the mother don't have such genes married a healthy man. Find the probability of giving birth to a child suffering from both diseases.

Problem 2. Recessive gene of hemophilia is located in the X-chromosome. There is a girl whose father is sick with hemophilia and mother is healthy and have no cases of hemophilia in the family. The girl marries a healthy man. What is the probability of giving birth to sick children in this family?

Task II. Draw the nuclei with different content of X chromosomes, indicate the norm or the name of the disease and the content of sex chromosomes.



- A. Female chromatin-positive nucleus
- B. Male chromatin-negative nucleus
- C. Female chromatin- negative nucleus
- D. Male chromatin-positive nucleus
- E. Female double chromatin-positive nucleus

Practice 13. Topic: FUNDAMENTALS OF HUMAN GENETICS (I)

Purpose of the practice: to study the purposes and the main methods of the human genetics at the present day, to learn how to solve problems in the context of composition and analysis of the family trees, finding the role of heredity in the character formation.

CONTROL QUESTIONS

- 1. Present tasks of human genetics.
- 2. The human as an object of genetic investigations.
- 3. Clinical-genealogical method.
- 4. Twin method.
- 5. Cytogenetic method. Biochemical methods.
- 6. Methods of recombinant DNA. The Human genome project.

BASIC TERMS AND CONCEPTS

- 1. Concordance –
- 2. Discordance –
- 3. Dizygotic twins –
- 4. Genealogy –
- 5. Monozygotic twins –
- 6. Proband -
- 7. Sequencing –

TESTS FOR SELF-CONTROL

201 year

- **1. Difficulties of studying human genetics are:** a) simple karyotype; b) early puberty; c) small amount of offsprings; d) a plenty of offsprings; e) an experimentation opportunity.
- **2.** The stages of genealogic analysis: a) the taking the anamnesis; b) definition of frequencies of genes and genotypes in a population; c) making genetic maps of chromosomes; d) studying the role of the environment in exhibiting character; e) analysis of a family tree.
- -3. Order of stages of the cytogenetic method: 1) processing of the cells by hypotonic solution NaCl; 2) staining of chromosomes; 3) stopping mitosis (with colchicine) at the stage of metaphase; 4) cultivation of cells on artificial nutrient mediums; 5) stimulation of mitosis by PHA: a) 1–5–3–4–2; b) 4–5–3–1–2; c) 4–1–5–3–2; d) 5–3–4–1–2; e) 4–5–1–3–2.
- **4.** Holzinger's formula is used for calculation: a) frequencies of genes and genotypes in a population; b) quotient of inheritance; c) roles of environment in exhibiting an attribute; d) probabilities of inheritance; e) degree of enetic risk.
- **5. What is studied by biochemical methods of human genetics?**a) general blood analysis;
 b) activity of enzymes of a blood plasma;
 c) activity of enzymes of a gastric juice;
 d) structure of primary urine;
 e) regional frame of enzym.
- **6. Methods of recombinant DNA are based on:** a) use of mathematical expression of the law of Hardy-Weinberg; b) extracting DNA fragments and determining nucleotide sequenceces in them; c) analysis of family trees; d) analysis of enzyme systems activity; e) microscopic karyotype studying.

Fill in the gaps:

- **1.** Man from whom medical-genetic examination of family and compiling genealogy start is called ...
- **2.** If parents are heterozygous (complete dominance, type of inheritance is autosomal-dominant and gene penetrance 25%), then the probability of giving birth to a sick baby is ... %
- **3.** If a mother is heterozygous and a father is healthy (X-linked dominant inheritance, gene penetrance is 40%), then the probability of giving birth to a sick baby is ... %.
- **4.** Determining the order of nucleotides and finding a pathologic gene is possible by the method of nucleic acids' ...
- **5.** Type of inheritance when the father transmits his character to all daughters, but neither to sons is called ...
- **6.** Method of human genetic that allows to reveal the role of heredity and environment in the formation of a character is called ...
- **7.** Genetic method that allows to reveal genome and chromosome mutations is called ...
- **8.** Biochemical ... tests allow to reveal heterozygous carriers of a pathologic gene.

PRACTICAL WORK

Solve the problems

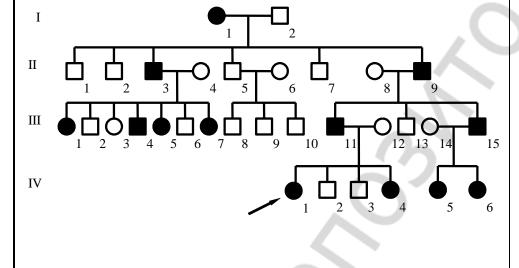
Problem 1. The concordance of monozygotic twins according to the body weight is 80 %, and the concordance of dizygotic ones is 30 %. What is ratio of heredity and the environment in the formation of this character?

Problem 2. Draw and analyze the family tree: the proband is a boy suffering from Duchenne muscle dystrophy. His parents and two sisters are healthy. In the father's line: two uncles, aunt, grandfather and grandmother are healthy. Cousins (two uncle's daughters and the aunt's son) are healthy. In the mother's line: one of the two uncles (the elder one) was sick with the distrophia. The second uncle (the healthy one) had two healthy sons and a healthy daughter. The aunt had a sick son. The grandfather and grandmother are healthy.

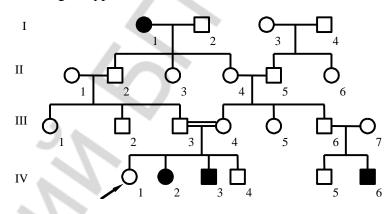
Problem 3. Which of the answers characterize X-linked dominant type of inheritance?

a) equal number of women and men suffer from the disease; b) the disease is transferred from the parents to the children in each generation; c) all the daughters of an ill father are ill; d) sons never inherit the disease from their fathers; e) if the mother is ill, then the possibility of an ill child birth is 50% without dependence of the sex?

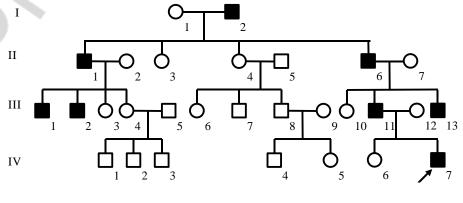
Problem 4. Analyze the family tree, determine the inheritance type and find the genotypes of the members.



Problem 5. Analyze the family tree, determine the inheritance type and find the genotypes of the members.



Problem 6. Analyze the family tree, determine the inheritance type and find the genotypes of the members.



Practice 14. Topic: FUNDAMENTALS OF HUMAN GENETICS (II)	201 year
Purpose of the practice: to study the methods of human genetics: mode	
disease prenatal diagnostics methods, to learn to solve problems using the F	lardy-Weinberg law.
CONTROL QUESTIONS	5. Guthrie test –
1. Modeling methods. A law of N. I. Vavilov.	
2. Characteristic of human populations. Types of marriages.	
3. Genetic processes in large populations. The law of Hardy-Weinberg.	
4. Genetic processes in small populations.	6. Panmixia –
5. Genetic load and its biological nature.	
6. Methods of prenatal diagnostics of hereditary diseases.	
7. Express-methods.	
BASIC TERMS AND CONCEPTS	7. Population –
1. Amniocentesis –	
2. Chorion biopsy –	8. Ultrasonography –
3. Demes –	9. α-fetoprotein –
4. Genetic drift –	

- **1.** Characteristic features of an ideal population are: a) great number of individuals; b) small number of individuals; c) complete panmixia; d) absence of mutations; e) presence of mutations.
- **2.** In mathematical expression of the Hardy–Weinberg law, p denotes the frequency of: a) dominant gene; b) recessive gene; c) dominant homozygotes; d) recessive homozygotes; e) heterozygotes.
- **3.** In mathematical expression of the Hardy–Weinberg law, q denotes frequency of: a) dominant gene; b) recessive gene; c) dominant homozygotes; d) recessive homozygotes; e) heterozygotes.
- **4.** In mathematical expression of the Hardy–Weinberg law, 2pq denotes frequency of: a) dominant gene; b) recessive gene; c) dominant homozygotes; d) recessive homozygotes; e) heterozygotes.
- **5. Microbiologic tests allow to:** a) build genetical maps of human chromosomes; b) determine the number of X-chromosomes; c) determine the number of Y-chromosomes; d) reveal some chromosome mutations; e) reveal some metabolism defects.
- **6. Dermatoglyphic analysis allow to:** a) study of patogenesis of skin diseases; b) develop measures for prophylaxis of skin diseases; c) determine the causes of skin diseases; d) reveal hereditary components of disease; e) diagnose metabolic defects.
- **7. Direct noninvasive methods of prenatal diagnostics are:** a) definition of the concentration of alpha-fetoprotein; b) ultrasonography; c) chorion biopcy; d) aminoicenthesis; e) fetoscopy.
- **8.** Optimal terms for carrying out direct noninvasive methods of prenatal diagnostics are: a) 6–8 weeks; b) 8–10 weeks; c) 12–20 weeks; d) 23–30 weeks; e) 30–35 weeks.
- **9. The genetic load is:** a) saturation of the population by positive mutations; b) saturation of the population by mutations, reducing adaptability of individuals; c) saturation of the population by neutral mutations; d) saturation of the population by negative mutations; e) absence of mutations in populations.

Fill in the gaps:

- **1.** Chorion biopsy is performed within ... weeks of pregnancy.
- **2.** Predicting changes of genetic structure of human populations can be carried out by with the ... method.
- 3. Level of α -fetoprotein in the blood of a pregnant woman ... in case of Down syndrome of the fetus.
- **4.** Each pregnant woman compulsory undergoes ... a direct non-invasive method of prenatal diagnostics.
- **5.** Mother's age of over 37 years, spontaneous abortions and stillbirth in the anamnesis, children with congenital malformations are indications for carrying out ... methods of prenatal diagnostics.
- **6.** Sex chromatin Y is determined by staining the cells of buccal epithelium by ...
- 7. In norm, the main palmar angle is not more than ...
- **8.** Human populations with the number not exceeding 1500 people where intragroup marriages surpass 90 % are called ...
- **9.** Genetic load has no phenotypic manifestation when ... of a pathological gene is observed.

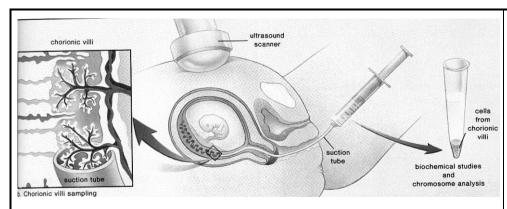
Solve the problems

Problem 1. In the USA, the 30% of persons of the examined population feel the bitter taste of phenylthiocarbamide (PTC) and the 70% do not. The ability to feel its taste is determined by the recessive gene **a**. Find out the frequency of the alleles **A** and **a** in the population.

Problem 3. Find out the frequency of albinos in the large African population where the concentration of the recessive pathology gene is 10%.

Problem 2. An aboriginal population of 127 (including children) persons lives in the jungle of the South America. The frequency of the M blood group is 64% here. Is it possible to find out the frequencies of N and MN blood groups in this population?

Problem 4. The rate of the disease gout is 2% and it is conditioned by the dominant autosomal gene. According to some information (V. Efroimson, 1968), gene penetrance in men is 20% and 0% in women. Find out the genetic structure of the population.



Problem 5. What method of prenatal diagnostics is shown in the figure? What are the indications for it?

Problem 6. Name the ridge patterns:



Problem 7. The results of the dermatoglyphic analysis of a patient are the following: the single transverse palmar crease on both palms, radial loops on the 4th and 5th fingers of both hands, the main palm angle is 77⁰. Is it possible to suspect that this man has a hereditary disorder according to this information?

Practice 15. Topic: CONTROL PRACTICE IN GENETICS

__201___ year

Purpose of the practice: to assess of the students' knowledge of genetics and the ability to solve typical problems

CONTROL QUESTIONS

- 1. Genetics as a science. Basic concepts of Genetics.
- 2. Peculiarities of the hybridological method.
- 3. Inheritance regularities in monohybrid cross.
- **4.** Hypothesis of purity of gametes and its cytological foundation.
- **5.** Analyzing cross. The concept of a phenotypic radical.
- **6.** Inheritance regularities in polyhybrid cross. The law of independent assortment.
- **7.** Conditions limiting the manifestation of Mendel's laws. Pleiotropy. Semi-lethal and lethal genes.
- **8.** Intra-allelic interaction of genes. Inheritance of blood groups.
- **9.** Inter-allelic interaction of genes.
- 10. Experiments of T. Morgan. Complete and incomplete genetic linkage.
- **11.** Autosomal and gonosomal linkage groups. Crossing-over. Basic concepts of the chromosomal theory of heredity.
- 12. Maps of eukaryotes' chromosomes (genetic and cytological).
- 13. Variation and its types. Phenotypic variation. The reaction norm.
- 14. Genotypic variation and its forms. Mutagenic factors.
- **15.** Classification of mutations. Genome, chromosome and gene mutations.
- **16.** Stability and repair of genetic material; anti-mutagens.
- 17. Biological basis of cancerogenesis.
- **18.** Sex as a biological character. Sex characters. Sex-controlled and sex-limited characters. X- linked and holandric characters.

- 19. Chromosome theory of sex determination.
- 20. Peculiarities of sex determination in humans and its impairments.
- 21. Sex chromatin. Chromosomal sex disorders.
- 22. Primary, secondary and tertiary ratios of sexes.
- **23.** Present tasks of human genetics. The human as an object of genetic investigations.
- **24.** Clinical-genealogical methods. Twin method.
- 25. Cytogenetic method. Biochemical methods.
- 26. Methods of a recombinant DNA. Human genome project.
- 27. Modeling methods. A law of N. I. Vavilov.
- 28. Characteristics of human populations. Types of marriages.
- **29.** Genetic processes in the large populations. The law of Hardy-Weinberg.
- **30.** Genetic processes in the small populations.
- 31. Genetic load and its biological nature.
- **32.** Methods of prenatal diagnosis of hereditary disorders. Expressmethods.

Practice 16. Topic: HUMAN GENETIC AND CHROMOSOMAL DISEASES		
CONTROL QUESTIONS	5. Syndactylia –	
 Gene mutations as a cause of metabolic diseases. Characteristic of gene human disorders. Chromosome and genome mutations as a cause of chromosomal human disorders. Characteristic of chromosomal human diseases. 	6. Trisomy –	
BASIC TERMS AND CONCEPTS 1. Hemophilia –	7. Enzymopathy –	
2. Microphthalmia –	8. Chromosomal diseases –	
3. Microcephaly –	9. Ceruloplasmin –	
4. Monosomy –	10. Epicanthus –	

- 1. Diagnostic symptoms of phenylketonuria are: a) mice odor, intellect is not disturbed; b) increased muscular irritability and tone, mental retardation; c) low muscular irritability and tone, reduced skin pigmentation; d) convulsive epileptiform attacks, hemorrhages in joints; e) increased contents of phenylalanine hydroxylase in the blood.
- **2. Diagnostic symptoms of albinism are:** a) hyposensitivity to ultraviolet rays; b) milky-white skin color; c) hair depigmentation; d) hair pigmentation; e) decreased acuity of vision.
- **3. Diagnostic symptoms of galactosemia are: a)** jaundice of newborns; b) vomiting, diarrhea, hepatomegaly and splenomegaly; c) depigmentation of skin and hair; d) propensity to self-damages; e) mental retardation.
- **4. Diagnostic symptoms Wilson–Konovalov disease are:** a) increased concentration of copper in the blood; b) increased concentration of iron in the blood; c) accumulation of copper in the liver and brain leading to their degeneration; d) accumulation of iron in the liver and brain leading to their degeneration; e) impairment of functions of liver and central nervous system.
- **5. Diagnostic symptoms of hemophilia A are:** a) time of blood coagulation is 5–6 minutes; b) nasal bleedings and paralysis of legs; c) plural hematomas; d) hemorrhages in large joints and intellect decrease; e) blood in urine and high arterial pressure.
- **6. The karyotype for Patau syndrome is:** a) 47, XXY; b) 47, XX, 18+; c) 47, XXX; d) 48, XYY; e) 47, XY, 13+.
- **7. Diagnostic symptoms of Edward syndrome are:** a) macrocephaly; b) congenital heart defects; c) big lower jaw and oral opening; d) throat underdevelopment; e) rocker bottom foot.
- **8.** The karyotype for Down syndrome is: a) 45, XX, 21-; b) 47, XY, 13+; c) 47, XX, 21+; d) 47, XY, 21+; e) 46, XX, 5q-.
- **9.** The karyotype for Cat cry (cri du chat) syndrome: a) 45, XX, 5-; b) 46, XY, 5-; c) 47, XX, 18+; d) 47, XY,5+; e) 46, XX, 5q-.

Fill in the gaps:

- 1. Increased concentration of copper in blood in Wilson–Konovalov disease is caused by mutation of the gene responsible for synthesis of protein ...
- 2. Sickle-cell anemia is caused by the mutation leading to replacement of glutamic acid with ... in 6^{th} position of the β -chain
- **3.** Increased level of uric acid and its salts n the organism caused by deficit of the enzyme catalyzing the addition of purine bases to nucleotides, is a symptome of a ... syndrome.
- 4. Hereditary deficiency of the enzyme tyrosinase leads to the ...
- **5.** Deficit of ceruloplasmin results in the ... disease
- **6.** Genetic diseases caused by the impairment of lipid exchange in the blood plasm due to defects of enzymes or cells' receptors are called ...
- **7.** Mutations associated with changes of chromosome number or impairment of their structure cause ... diseases.
- **8.** ... syndrome results from trisomy on the 18th pair of autosomes.

Solve the problems

Problem 1. The parents (the wife aged 45, the husband aged 50) gave birth to a full-term child. The child has a flat face, low backward-sloping forehead, big head, upslanting palpebral fissures, epicanthus, light spots on the iris, thick lips, thick tongue protruding from the mouth, underdeveloped low-set auricles, high palate, improper growth of the teeth, unclosed interatrial septum, a single transverse palmar crease, main palmar angle 65°; the significant mental retardation is observed.

What disease can be suspected? Which methods should be used to make a right diagnosis? What is the future viability prognosis for this child? Which methods of prenatal diagnostics could be used to diagnose this disease?

Problem 2. In the family of healthy parents who are second-cousins, a full term child was born who was breast-fed by the mother. The vomit and diarrhea, jaundice, mental retardation, hepatomegalia and lien enlargement, general dystrophy, cataract gradually appeared and got stronger in the course of time.

What disease can be suspected? What laboratory research should be made? Is it possible to stop the disease progression? What is the possibility of the second ill child birth in this family?

Problem 3. Which symptoms of the listed ones are the diagnostic characters of Edwards' syndrome a) mental retardation, hepatomegalia and lien enlargement, general dystrophy, cataract; b) macrocephaly, microphthalmia, double-sided cleft of lip and palate, toe dactylion, ventricular septal defect of the heart, mental retardation; c) semiluxation of the crystalline lens, cardiac failures, tall height, long thin fingers, "funnel chest"; d) blue sclera, congenital deafness, fragility of bones; e) congenital defects, low ear auricles, elongated skull, abnormal development of the footsteps, mental retardation?

Problem 4. The family has a child aged 5 with the mice odour, increased muscle tone, convulsive epileptic attacks, mental retardation, macrocephaly, weak pigmentation of the skin and hair. What disease can be suspected? How can it be diagnosed? What is the possibility of giving birth to the next child with this pathology?	Problem 6. In a young family a child was born whose cry sounds like the cat's cry. He has a moon-like face, muscular hypotony, microcephaly, antimongoloid eyes, epicanthus, squint, deformed low ear auricles, mental retardation. What disease can be suspected? Which methods should be used to diagnose it? What is the future viability forecast for this child?
Problem 5. In the family of healthy parents a full term child with low body weight (2600 g) was born. The baby has microcephaly, low backward-sloping forehead, narrow eye splits, microphthalmia, aglia, deformed ear auricles, double-sided cleft of lip and palate, toe dactylion,	
single transverse palmar creases, ventricular septal defect of the heart, significant mental retardation. What disease can be suspected? What methods of the prenatal diagnostics are necessary to be used to diagnose this disease?	
	Teacher's signature

Practice 17. Topic: GENETIC COUNSELLING Purpose of the practice: to study the aims of genetic counselling, the stage ical genetic counselling service, principles of hereditary disease there problems.	
CONTROL QUESTIONS	6. Gene therapy –
1. The aim and tasks of genetic counselling.	
2. Characteristic of the stages of making genetic prognosis.	
3. Treatment principles of hereditary human pathology.	7. Substitution therapy –
BASIC TERMS AND CONEPTS	
1. Mild genetic risk –	
	8. Pathogenic therapy –
2. Medium genetic risk –	
3. High genetic risk –	9. Symptomatic therapy –
4. Diet therapy –	10. Etiotropic therapy –
5. Metabolic inhibition –	

- 1. The main aims of genetic counselling are: a) estimating of a genetic risk degree in the examined family; b) to decrease the frequency of all diseases; c) to decrease the frequency of genetic diseases; d) to decrease the frequency of congenital malformations; e) to increase the birthrate.
- **2. High genetic risk is:** a) up to 5 %; b) 5–10 %; c) 10–20 %; d) 20-30 %; e) about 50 %.
- 3. Indications for directing a family to-genetic counselling are:
 a) presence of similar hereditary diseases at several family members;
 b) arrested physical growth of the child; c) infection disease in the family; d) parasitic disease in family; e) divorce of spouses.
- **4.** Examples of symptomatic treatment of hereditary disorders are:
 a) pain killers for inflammatory processes; b) antibiotics for pain syndrome; c) sedatives for excitement; d) excluding substance that is not-metabolized in the organism from a diet; e) surgical correcting of congenital defects.
- **5. Hereditary diseases, corrected by special diets are:** a) Down syndrome; b) phenylketonuria; c) mucoviscidosis; d) galactosemia; e) Duchenne myodystrophy.
- 6. Examples of pathogenic treatment of hereditary disorders are:
 a) pain killers for a pain syndrome; b) metabolic inhibition; c) gene therapy; d) excluding substance that is notmetabolized in the organism from a diet; e) restriction of non metabolic substance in the diet.
- 7. Examples of etiological treatment of hereditary disorders are:
 a) metabolic inhibition; b) antibiotics; c) substitution therapy;
 d) excluding substance that is not metabolized in the organism from a diet; e) gene therapy.
- **8. Metabolic inhibition includes:** a) restriction of substance receipt with food; b) elimination of the substrate of pathological reaction from the organism; c) compensation of not synthesized product; d) suppression of pathological substrate's synthesis; e) protection of an organ against receipt of catabolic products.

Fill in the gaps:

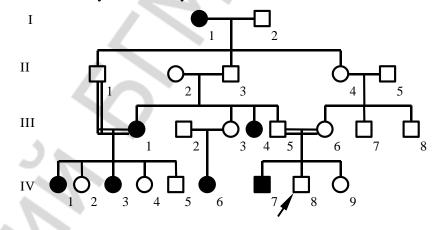
- **1.** Substitution therapy is an example of the ... treatment of hereditary disorders.
- 2. Dietotherapy is an example of the ... treatment of hereditary disorders.
- **3.** Prescribing anesthetics is an example of the ... treatment of hereditary disorders.
- **4.** Surgical removal of the 6-th finger is an example of the ... treatment of hereditary disorders.
- **5.** Gene therapy is an example of the ... treatment of hereditary disorders.

Task I. Solve the problems

Problem 1. The son of American banker Twister suffered from three diseases: hemophilia, daltonism and total absence of teeth. These diseases are caused by the genes located in the X chromosome. Twister junior has been living in Paris for many years, far away from the parents, where he died in 1944. After his death, a French woman with a 15-year-old boy came to Twister senior. The boy had hemophilia, daltonism and the absence of teeth. The woman told that this boy is a son of passed Twister junior and he was his rightful heir but the documents proving that had been lost. Despite the absence of the documents, Twister senior recognized the boy to be his grandson. The family doctor convinced him that such coincidence of three rare hereditary diseases proved that the boy was his grandson. Are you agree with the doctor's opinion?

Problem 2. In a family, a man and son are suffering from hemophilia. The man's wife is pregnant. Being afraid that she will give birth to a son with haemophilia, she consults to genetic counselling to clear up the sex of the fetus and abort the fetus if it is a boy. Having talked to her the doctors recommended to abort the fetus without carrying out the amniocentesis. Is this recommendation right?

Problem 3. Analyse the family tree:



Define the type of inheritance. What is the probability of a sick child birth if an ill girl (IV, 1) marries a heterozygous man? What methods of prenatal diagnostics can be used for making the diagnosis of hereditary pathology of the fetus? Which recommendations should the geneticist make?

Practice 18. Topic: CONTROL PRACTICE IN CYTOLOGY, MOLECULAR BIOLOGY AND GENETICS _______201_____year

Purpose of the practice: to assess the students' knowledge of cytology and genetics and the ability to solve typical problems.

CONTROL QUESTIONS

- 1. Origin of life. Evidence of the organic world evolution.
- 2. Organization levels of living things. Properties and characters of living things.
- 3. Methods of studying living things (methods of biological sciences).
- 4. The significance of Biology for medicine.
- 5. The position of the human in the animal world system.
- 6. Humans as biological and social beings.
- 7. The subject, tasks and methods of cytology.
- 8. Magnifying devices and their purpose. Arrangement of a light microscope.
- 9. Rules of working with the microscope.
- 10. The present state of the cell theory.
- 11. Distinguishing features of pro- and eukaryotic cells.
- 12. The structure (a models) of elementary membrane, its properties and functions.
- 13. Ways of passing substances into the cell.
- 14. Anabolic system of the cell.
- 15.Catabolic system of the cell.
- 16. Energy exchange in the cell. Enzymatic systems of mitochondria.
- 17. The structure and functions of the nucleus.
- 18. Types of chromosomes. The structure of a metaphase chromosome.
- 19. Cell and mitotic cycles.
- 20.Interphase, characteristic of periods. Reasons of mitosis.
- 21. Characteristic and significance of mitosis.

- 22. Characteristic and significance of meiosis.
- 23. Amitosis.
- 24. The concept of karyotype and ideogram.
- 25. Methods of studying the human karyotype.
- 26. Denver and Paris classifications of human chromosomes
- 27. Nucleic acids (DNA and RNA): the structure and functions. Chargaff rules.
- 28. Proofs of the nucleic acids role in transmission hereditary information.
- 29. Properties of genes.
- 30. DNA replication.
- 31. The genetic code and its properties. Protein biosynthesis.
- 32. The central dogma of Molecular Biology/
- 33. Levels of genetic material packing.
- 34. Classification of genes.
- 35. Transcription regulation in prokaryotes.
- 36. Transcription regulation in eukaryotes (the diagram of G. P. Georgiev).
- 37. Cytoplasmic heredity.
- 38. Stages of genetic engineering methods.
- 39. Obtaining of genetic material.
- 40. Insertion of DNA fragments into the molecule-vector.
- 41. Incorporation of the recombinant DNA into the cell-recipient.
- 42. Using methods of genetic engineering in medicine.
- 43. Genetics as a science. Basic concepts of Genetics.
- 44. Peculiarities of the hybridological method.
- 45. Inheritance regularities in monohybrid cross.

- 46. Hypothesis of purity of gametes and its cytological foundation.
- 47. Analyzing crossing. Phenotypical.
- 48. Regularities of inheritance in polyhybrid crossing. The law of independent assortment.
- 49. Conditions limiting the Mendel's laws. Pleiotropy. Semi-lethal and lethal genes.
- 50. Intra-allelic interaction of genes. Inheritance of blood groups.
- 51. Inter-allelic interaction of genes.
- 52. Experiments of T. Morgan. Complete and incomplete genetic linkage.
- 53. Autosomal and gonosomal linkage groups.
- 54. Crossing-over, crossover and non-crossover gametes.
- 55. Basic concepts of the chromosome theory of inheritance.
- 56. Maps of eukariotic chromosomes (genetic and cytological).
- 57. Variation and its types.
- 58. Phenotypic variation. The reaction norm.
- 59. Genotypic variation and its types.
- 60. Mutagenic factors.
- 61. Classification of mutations.
- 62. Genome, chromosome and gene mutations.
- 63. Stability and repair of genetic material; anti-mutagens.
- 64. Biological basis of cancerogenesis
- 65. Sex as a biological character. Sex characters.
- 66. Sex-controlled and sex-limited characters.
- 67. Characters linked with an X-chromosome and the holandric ones.
- 68. Chromosome theory of sex determination.
- 69. Peculiarities of sex determination in humans and its impairments.
- 70. Sex chromatin.

- 71. Sex chromosome disorders.
- 72. Primary, secondary and tertiary ratios of sexes.
- 73. Present tasks of human genetics.
- 74. The human as an object of genetic investigations.
- 75. Clinical-genealogical method.
- 76. Twin method.
- 77. Cytogenetic method. Biochemical methods.
- 78. Methods of recombinant DNA. The Human genome project.
- 79. Modeling methods. A law of N. I. Vavilov.
- 80. Characteristic of human populations. Types of marriages.
- 81. Genetic processes in large populations. The law of Hardy-Weinberg.
- 82. Genetic processes in small populations.
- 83. Genetic load and its biological nature.
- 84. Methods of prenatal diagnostics of hereditary diseases.
- 85. Express-methods.
- 86. Gene mutations as a cause of metabolic diseases.
- 87. Characteristic of gene human disorders.
- 88. Chromosome and genome mutations as a cause of chromosomal human disorders.
- 89. Characteristic of chromosomal human diseases.
- 90. The aim and tasks of genetic counselling.
- 91. Characteristic of the stages of making genetic prognosis.
- 92. Treatment principles of hereditary human pathology.

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