# Methodical instructions for practical classes

**Discipline: "Genes and Heredity" (Medical Genetics)**

**Discipline code: GN 1204**

**EP: 6B10115 "Medicine".**

**Volume of study hours/credits: 120 hours/4 credits (12 h)**

**Course and semester of study: 1-2**

**Shymkent - 2023**

The Methodological instructions for practical classes was developed in accordance with the working curriculum of the EP "Genes and heredity", the discipline "Medical genetics" and discussed at the meeting of the department.

Protocol no. __18_ of «_13__» ___06_____ 2023 y.

Head of Department, Professor _____ Yessirkepov M.M.

**№1**

**1. Topic:** Fundamentals of Medical Genetics

**2. Purpose:** Familiarization with the methods of studying human genetics, with the application of research methods of human genetics.

**3. Learning objectives:** The concept of environment and its influence on the formation of diseases. Familiarization with methods of studying human genetics, with application of methods of studying human genetics.

**4. Main questions of the topic:**

**1.** Specifics of the study of human genetics.

2. Methods of studying human genetics:

* twins,
* chiromancy and dermatoglyphics,
* genetics of somatic cells,
* population-statistical,
* biochemical,
* cytogenetic methods:
* analysis of telophase and anaphase chromosomes

o analysis of metaphase chromosomes

o metaphase analysis of all chromosomes

o stages of cytogenetic methods:

o obtaining metaphase chromosomes in preparations

o staining of preparations

o identification of chromosomes

12. principles of analysing relatives:

a) determination of the heritability of traits

b) determination of inheritance types

c) calculation of genetic risk

**5. Learning and teaching methods/techniques:** Work with microphotographs, charts, tables.

**6. Assessment methods/techniques:** Testing, oral questioning on the control measuring instruments materials.

**7. Literature:** see appendix 1

**8. Control:**

1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

**№2**

**1. Topic:** Fundamentals of general genetics.

**2. Objective:** Research methods of human genetics. Detection of hereditary diseases

**3. Learning objectives:** To be able to solve problems according to Mendel's laws. To apply methods of genealogical analysis by genealogical method

**4. Main topic questions:**

1. History of development of genetics
2. Principles of analyzing relatives:

a) determination of heredity of traits

b) determination of inheritance typesв) расчет генетической опасности
3 History of the origin of genetics
4. The genealogical method
5. Gemini methods
6. Biochemical methods
7. Heterozygote
8. Homozygote
9. Cytogenetic methods
**5. Learning and teaching methods/techniques:** Work with microphotographs, diagrams, tables, problem solving.
**6. Assessment methods/techniques:** Testing, oral questioning on the materials of control measuring instruments.
**7. Literature:** see appendix 1
**8. Control:**
1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

**№3**
**1. Topic:** Basics of general genetics. Linked inheritance.
**2. Objective:** Linked inheritance. Study of the chromosomal theory of heredity
**3. Learning Objectives:** Morgan's Law. To be able to compose genetic problems
**4. Main questions of the topic:**
1. Genetic maps
2. Morgan's law
3. What is the genome of combined genes?
4. The complete combination
5. Incomplete combination
6. Crossing over
7. Chromosomal sex determination
8. Inheritance of traits in combination with Sex
9. Types of inheritance of traits
10. Chromosome theory of heredity
**5. Learning and teaching methods/techniques:** Work with microphotographs, diagrams, tables, problem solving.
**6. Methods of assessment:** Testing, oral questioning on the materials of control and measurement tools.
**7. Literature:** see appendix 1
**8. Control:**
1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

**№4**
**1. Topic:** Chromosomal diseases

**2. Objective:** to find out the contribution of the process of variability to the formation of organism pathology; to study the genetic mechanisms of chromosomal diseases.
**3. Learning objectives: students should know** the essence of the concept of hereditary diseases, mechanisms of their occurrence, types and methods of prevention; be able to operate with this knowledge when studying the basics of genetics; classify the types of chromosomal diseases and diseases with non-Mendelian type of inheritance; **be able to operate** with this knowledge to understand and be able to diagnose these diseases.
**4. The main questions of the topic:**
1. Hereditary diseases.Mechanisms of occurrence.
2. Classification of chromosomal diseases.
3. Chromosomal diseases:
* Monsomia X-chromosome syndrome,
* Polysomy X-chromosome syndrome in males and females,
* Y-chromosome polysome syndrome,
* Autosomal monosomy syndrome *
* Autosomal polysomy syndrome,
4. Types of specific (non-traditional) hereditary diseases:
* hereditary diseases in combination with sex
* mitochondrial diseases,
* genomic imprinting diseases,
* threenucleotide repeat expansive diseases,
* prion diseases.
* hereditary metabolic diseases
**5. Learning and teaching methods/techniques:** Work with microphotographs, diagrams, tables.
**6. Assessment methods/technologies:** Testing, oral questioning on the materials of control and measurement tools.
**7. Literature:** see appendix 1
**8. Control:**
1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

**№5**
**1. Topic:** Congenital malformations.
**2. Objective:** Study of human hereditary diseases. Study of etiology, pathogenesis and epidemiology of hereditary diseases
**3. Learning objectives:** the learner should know the mechanisms of occurrence of the nature of congenital hereditary diseases, mechanisms of their prevention
**4. Main questions of the topic:**
1. Human hereditary diseases. Their classification. Ways of prevention
2. Role of heredity and occurrence of human pathology in the environment
3. Methods of prevention of hereditary diseases.
4. What causes birth defects?
- Genetic factor
- Environmental factors
- Multifactorial causes
5. Non-genetic causes

6. What are the risk factors for birth defects?
7. How are birth defects diagnosed?
8. How are birth defects treated?
9. How are birth defects prevented?
10. Teratogens
11. Teratogen factors
5. Learning and teaching methods/techniques: Work with microphotographs, diagrams, tables.
6. Assessment methods/techniques: Testing, oral questioning on the materials of control and measurement tools.
7. Literature: see appendix 1
8. Control:
1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

**№ 6**
**1. Topic:** Prenatal diagnosis of hereditary diseases.
**2. Objective:** to familiarize students with modern methods of laboratory diagnosis and prevention of hereditary diseases and the basics of medical and genetic counseling. Study of genetic processes in the population.
**3. Learning objectives:** students should know the essence of the basic methods of prenatal diagnosis of hereditary diseases and be able to apply this knowledge in practice.
**4. Main questions of the topic:**
1. Genetic basis for the prevention of hereditary diseases:
- primary prevention
- secondary prevention
- tertiary prevention
- gene expression management
- elimination of embryos and inherited foetal pathologies
- genetic engineering at the level of dead cells
- family planning
- environmental protection
2. Medical and genetic counseling
3. Prenatal diagnosis:
- identification of the pregnant woman with biochemical markers by screening
- invasive methods
- amniocentesis
- cordocentesis
- chorion and placentobiopsy
- non-invasive methods
- ULTRASOUND
4. Preimplantation diagnostics
5. Preclinical diagnostics, preventive treatment
**5. Learning and teaching methods/techniques:** Work with microphotographs, diagrams, tables.
**6. Assessment methods/techniques:** Testing, oral questioning on the materials of control and measurement tools.
**7. Literature:** see appendix 1

**8. Control:**
1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

**№7**
**1. Topic:** Diseases with non-Mendelian type of inheritance.
**2. Objective:** To form students' knowledge and ideas about the causes and mechanisms of monogenic diseases caused by mitochondrial (nuclear) genes, expansion (dynamic mutation) of nuclear genes, genomic imprinting, mutations of prion genes.
**3. Learning objectives:** to know - to disclose the essence of the causes and mechanisms of mitochondrial diseases; - to disclose the causes and mechanisms of diseases based on the expansion of three nucleotide genes (with dynamic mutation); - to disclose the causes and mechanisms of diseases based on genomic imprinting; - to identify the causes of diseases caused by prion gene mutations and mechanisms of development; - to familiarize students with the main clinical features of monogenic diseases
**4. The main questions of the topic are:**
1. Mitochondrial diseases, mechanism of occurrence, types and features of inheritance
2. Mechanism and essence of occurrence of dynamic mutations.
3. Diseases, mechanism of origin, types and features of inheritance, caused by expansion of three nucleotide repeats
4. Diseases of genomic imprinting, mechanism of occurrence, types and features of inheritance
5. Genetic mechanism of prion diseases.
**5. Learning and teaching methods/techniques:** Work with microphotographs, diagrams, tables.
**6. Assessment methods/techniques:** Testing, oral questioning on the materials of control and measurement tools.
**7. Literature:** see appendix 1
**8. Control:**
1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

**№8**
**1. Topic:** Principles of prevention of human hereditary pathology. Medical and genetic counseling
**2. Objective:** Study of etiology, pathogenesis and epidemiology of hereditary diseases. Study of medical and genetic counseling.
**3. Learning objectives:** the learner should know the mechanisms of hereditary diseases, mechanisms of their prevention.
**4. Main questions of the topic:**
1. Hereditary diseases
2. Hereditary diseases by genetic load
- monogenic
- chromosomal
- multifactorial (polygenic)

3. treatment of hereditary diseases
4. Ineffectiveness of inbreeding
5. Medical and genetic counseling

**5. Learning and teaching methods/techniques:** Discussions of key issues, video training, presentation
**https://www.youtube.com/watch?v=Xh_RplAaNBQ&feature=youtu.be** mono, poly, chromosomal diseases
**https://www.youtube.com/watch?v=dFOwvD1Xb0w&feature=youtu.be** monogenn

**6. Assessment methods/techniques:** Tests oral and written questioning

**7. Literature:** see appendix 1

**8. Control:**
1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

**№ 9**

**1. Topic:** Monogenic diseases arising from changes in protein structure.

**2. Objective:** Molecular and genetic mechanisms of pathogenesis of monogenic diseases

**3. Learning objectives:** the learner should be able to characterize monogenic diseases

**4. Main questions of the topic:**
1. Normal state of protein metabolism
2. Change of protein balance in the process of personal development
3. Change of protein metabolism in pathological conditions.
4. Diseases of ion channels
5. Collagenopathies
6. Hereditary diseases of amino acid metabolism
- Phenylalanine
- Haemoglobiopathies
7. Hereditary diseases of carbohydrate metabolism
8. Hereditary diseases of lipid metabolism
9. Lysosomal diseases
10. Peroxisomal diseases
11. Mitochondrial diseases

**5. Learning and teaching methods/techniques:** Work with microphotographs, diagrams, tables.

**6. Assessment methods/techniques:** Testing, oral questioning on the materials of control and measurement tools.

**7. Literature:** see appendix 1

**8. Control:**
1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

**№10**

**1. Topic:** Polygenic diseases.

**2. Objective:** Pathogenesis and etiology of polygenic diseases (diseases prone to heredity).
**3. Learning objectives:** the learner should be able to characterize polygenic diseases
**4. Main questions of the topic:**
1. General characterization and classification
2. Diseases prone to heredity
3. Mechanism of molecular-genetic analysis of diseases origin
4. Some genes predisposing to multifactorial diseases.
5. Some inherited diseases with Clinical and genetic specificity:
(a) arterial hypertension
c) bronchopulmonary diseases
d) diabetes mellitus
e) gastric and duodenal ulcer disease
f) Alzheimer's disease
d) immune diseases
h) infectious diseases
i) malignant neoplasm
**5. Learning and teaching methods/techniques:** Work with microphotographs, diagrams, tables.
**6. Assessment methods/techniques:** Testing, oral questioning on the materials of control and measurement tools.
**7. Literature:** see appendix 1
**8. Control:**
1. Answers to test questions.
2. Solving situational tasks.
3. Filling in cards on the topic.
4. Answering oral questions.

**№11**
**1. Topic:** Chromosomal diseases
**2. Objective:** Study of epidemiology and pathogenesis, etiology of chromosomal diseases.
**3. Learning Objectives:** the student should be able to describe chromosomal diseases
**4. Main questions of the topic:**
1. Classification of chromosomal diseases, mechanism of their occurrence.
2. Etiology, clinical and genetic syndrome, monosomy associated with X-chromosome.
3. Clinical seers
4. Etiology, clinic and genetics of X chromosome polysomy in women and men.
5. Clinical seers
6. Etiology of diseases of Y-chromosome polysomia
7. Clinic and genetics
8. Etiology, etiology, clinic of autosomal monosomy syndrome and genetics of syndromes associated with autosomal polysomy.
9. Etiology, clinic and genetics of syndromes due to partial autosomal monosomy.
**5. Learning and teaching methods/techniques:** Work with microphotographs, diagrams, tables.
**6. Assessment methods/techniques:** Testing, oral questioning on the materials of control and measurement tools.
**7. Literature:** see appendix 1
**8. Control:**
1. Answers to test questions.
2. Solving situational tasks.

3. Filling in cards on the topic.
4. Answering oral questions.

**№ 12**

**1. Topic:** Fundamentals of human population genetics

**2. Objective:** To familiarize students with the processes of genetics occurring in a population. To disclose the ecological characteristic of the population structure, to study the genetic structure of the population, the property of the gene pool: genetic unit (Hardy-Weinberg law) and genetic diversity (genetic polymorphism). Study the origin of genetic polymorphism in the human population.

**3. Learning objectives:** the student should know the ecological structure of the population, its gene pool properties; to study the genetic process, the provision of genetic polymorphism in the population; to be able to make a characterization of genetic polymorphism and genetic polymorphism of humans

**4. The main questions of the topic are:**

1. Population, definition.
2. Elementary evolutionary factors
3. Genetic structure of a population: genetic unity and genetic polymorphism
4. Characteristics of a genetic population:

- gene pool (the totality of genotypes of all individuals in a population),
- gene and genotype frequencies,
- frequencies of phenotypes, mating system,
- factors that alter gene frequencies.

4. Human population structure, its characterisation and types: Mendelian, demes, isolates.
5. Genetic unity (Hardy-Weinberg law) of a population. Hardy-Weinberg law and its significance for medicine
6. Genetic polymorphism - characteristic of genetic diversity of a population.
7. Types of genetic polymorphism: adaptation and balanced.
8. Genetic load - source of appearance of recessive alleles.
9. Genogeography of hereditary diseases.

**5. Learning and teaching methods/techniques:** Work with microphotographs, diagrams, tables.

**6. Assessment methods/techniques:** Testing, oral questioning on the materials of control and measurement tools.

**Annex 1**

**In Russian:**

**Basic:**

1. Genetics. Textbook for universities/edited by Academician RAMS V.I. Ivanov - M.: ICC "Akademkniga", 2006-638p.
2. Muminov T. Fundamentals of molecular biology: a course of lectures. -Almaty: Effekt, 2007.

**Additional:**

1. Ivanyushkin A.Y., Ignatiev V.N., Korotkikh R.V., SiluyanovaI.V.Izd-vol. Progress, M. 2008 y.
2. U. Clague, M. Cummings. Fundamentals of Genetics - M.: Technosphere, 2009.
3. Fundamentals of molecular biology of the cell. Textbook. 3 volumes. B. Alberts et al, OZON.RU Publishing House, 2018.

| № | Name | Links |
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| 1 | Electronic library | http://lib.ukma.kz |
| 2 | Electronic library of republican higher | http://rmebrk.kz/ |

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| 3 | Electronic library of “Student Adviser” Medical University library | http://www.studmedlib.ru |
| 4 | “Paragraph” information system “Medicine” department | https://online.zakon.kz/Medicine |
| 5 | Scientific electronic library | https://elibrary.ru/ |
| 6 | «BooksMed» electronic library | http://www.booksmed.com |
| 7 | «Web of science» (Thomson Reuters) | http://apps.webofknowledge.com |
| 8 | «Science Direct» (Elsevier) | https://www.sciencedirect.com |
| 9 | «Scopus» (Elsevier) | www.scopus.com |
| 10 | PubMed | https://www.ncbi.nlm.nih.gov/pubmed |

**Internet resours:**

1. Genetics. Textbook for Higher Education Institutions / Edited by Academician of RAMS V.I. Ivanov - Moscow: ICC "Akademkniga", 2011-638 p.
2. Mushkambarov N.N., Kuznetsov S.N. Molecular biology. Textbook for students of medical universities, 3rd edition, Moscow: Nauka, 2016, 660 p.
3. У. Clague, M. Cummings. Fundamentals of genetics - M.: Technosphere, 2009.
4. Kurchanov.A. Human genetics with the basics of general genetics: textbook -SPb, 2009.
5. Alberts B. B., Bray D., Hopkin K. Fundamentals of molecular biology of the cell. Textbook. 2nd ed., revised, per. from Engl. 768 p. 2018 y.
6. Spirin A.S. Protein biosynthesis, the RNA World and the origin of life.
7. Spirin A.S. Molecular Biology. Structure of ribosomes and protein biosynthesis. - M.: (electronic textbook).