PLAN OF LECTURES AND PRACTICAL EXERCISES ON MEDICAL GENETICS

Section 1. Semiotics. Medical and genetic counseling

Topic 1. Semiotics of genetic diseases

Lecture 1. Semiotics of genetic diseases.

Question 1. Common semiotics.

Summary of 1 question. The subject of the General semiotics are the signs related to the General characteristic of proband or its sibs (sex, age, hereditary burden, past diseases, Constitution and Constitution, sexual and physical development, etc.), and its General condition (change in consciousness, body position, expression of the phenotype, face, skin color, etc.). Examples.

Question 2. Private semiotics.

Summary 2 questions. The subject of private semiotics of organs and systems of the body are the symptoms of specific diseases, mechanisms of their occurrence and development, diagnostic value, assessment of the degree of severity, features of their combination, the causes of possible absence. Examples.

Question 3. Phenogenetics.

Summary of 3 questions. Implementation of genetic information in the phenotype. The study of gene/genes by Hecker. Comparison of normal signs and phenotypic manifestations of a certain mutation that violates the development of signs. A detailed analysis of the manifestations of genetic information at each stage of its implementation (pathogenesis of a mutant trait or traits). Examples.

Laboratory lesson 1. Semiotics of genetic diseases

Summary of the laboratory lesson. Scheme of description of the phenotype. Inspection of individual parts of the body. Clinical and genealogical method: 1) genealogical analysis of pedigree, algorithm of pedigree; 2) risk groups depending on the type of possible hereditary pathology (in chromosomal pathology, monogenic diseases, multifactorial diseases); 3) graphical means of pedigree description, 4) methods of phenotype investigation.

Independent work: Semiotics of hereditary and congenital diseases. Make a map of your own phenotype.

Topic 2. Methods of clinical genetics.

Lecture 2. Methods of clinical genetics

Question 1. Clinical and genealogical method.

Summary of 1 question. Methods of collecting information and its features in various types of pathology. Possible error. The value of clinical and genealogical method in clinical practice. Examples.

Question 2. Cytogenetic methods.

Summary 2 questions. Stages of carrying out. Options, scope, value. Biochemical method. Levels of biochemical diagnosis. Indications for biochemical research for the diagnosis of hereditary diseases. Examples.

Question 3. Molecular-genetic methods.

Summary of 3 questions. The universality of DNA diagnostic methods, the possibility of their use. Characteristics of the main methodological techniques. Examples.

Laboratory lesson 2. Methods of clinical genetics.

Summary of the laboratory session. The methods of study of hereditary diseases, the main of which are: clinical and genealogical, twin, population, cytogenetic, biochemical and molecular genetic. Criteria for inclusion in risk groups by the nature of the trait (disease) inheritance are analyzed. Understand the indications for one of the main genetic methods.

Independent work: Graphic representation of the inheritance of one of the signs or disease.

Section 2. Hereditary disease

Topic 3. Chromosomal disorders, monogenic disorders and multifactorial diseases
Lecture 3. Chromosomal pathology, monogenic and multifactorial diseases.
Question 1. Classification of chromosomal diseases
Question 2. Frequency of occurrence. Symptoms, clinical signs.
Question 3. Mechanisms of chromosomal diseases development. Relationship with the age of the parents.
Laboratory lesson 3. Chromosomal disorders, monogenic disorders and multifactorial diseases
Summary of the laboratory session. Calculation of risk in pathology: 1) chromosomal, 2) monogenic, 3) multifactorial. The solution of typical tasks to determine the risk of genetic diseases. Analysis of phenotypic maps. Classification of genetic diseases (genetic, clinical, pathogenetic principles of classification). Features of the clinical picture of genetic diseases: 1) the variety of clinical manifestations; 2) the value of the age of the onset of the disease; 3) the progression of the clinical picture, the chronic nature of the disease; 4) the severity of the disease; 5) clinical polymorphism of genetic diseases (the period of onset of the disease, the completeness and severity of symptoms, duration of illness, degree of disability, tolerance to therapy, duration of life); 6) the causes of the clinical polymorphism (different biochemical effects of gene mutations, the phenomenon of anticipation, the dose of genes, genetic environment, the impact of the external environment). Etiology and pathogenesis of genetic diseases.
Independent work: solving problems to determine the risk of genetic diseases. Preparation of multimedia presentations on the topic of the lesson.

Topic 4. Hereditary metabolic diseases (mitochondrial, peroxisomal, lysosomal diseases)
Lecture 4. Hereditary metabolic diseases.
Question 1. The concept of orphan diseases. Mitochondrial diseases.
Summary of 1 question. Molecular genetic classification of mitochondrial diseases. Etiology and pathogenesis of mitochondrial diseases. Morphogenetic characteristics of individual diseases.
Question 2. Peroxisomal diseases.
Summary 2 questions. Molecular genetic classification of peroxisomal diseases. Etiology and pathogenesis of peroxisomal diseases. Morphogenetic characteristics of individual diseases.
Question 3. Lysosomal storage diseases.
Summary of 3 questions. Molecular genetic classification of lysosomal diseases. Etiology and pathogenesis of peroxisomal diseases. Morphogenetic characteristics of individual diseases.
Laboratory lesson 4. Hereditary metabolic diseases (mitochondrial, peroxisomal, lysosomal diseases)
Summary of the laboratory session. Classification of hereditary metabolic diseases (genetic, clinical, pathogenetic principles of classification). Construction of a graphological diagnostic algorithm for the diagnosis of orphan diseases. Nutrition therapy for genetic diseases.
Independent work: continuation of the pedigree. Preparation of multimedia presentations on the topic of the lesson.

Topic 5. Hereditary diseases characteristic of the Chuvash Republic.
Lecture 5. Hereditary diseases characteristic of the Chuvash Republic.
Question 1 prevention of hereditary pathology.

Short content 1voprosa. Complex of pre-conception prophylaxis. Indications for preconceptional prevention (Hollingsworth).
Question 2. Gene pool as a strategic resource. Genetic prediction. The concept of genofond.
Summary 2 questions. Genetic markers of susceptibility and resistance to certain socially significant diseases.
Question 3. Hereditary diseases characteristic of the Chuvash Republic.
Summary of 3 questions. General overview of endemic hereditary pathology characteristic of the Chuvash Republic. Congenital erythrocytosis, lethal osteopetrosis, congenital isolated hypotrichosis.
Laboratory lesson 5. Hereditary diseases characteristic of the Chuvash Republic.
Summary of the practical lesson. Familiarization with the semiotics of orphan diseases endemic to the Chuvash Republic. Prevention of orphan diseases.
Independent work. Ethical and deontological issues in clinical genetics (preparation of multimedia presentations).

Section 3. Diagnosis of hereditary diseases

Topic 6. Pharmacogenetics and pharmacogenomics

Lecture 6. Pharmacogenetics and pharmacogenomics.
Question 1. The concept of pharmacogenetics.
Summary of 1 question. The effect of heredity on the effects of drugs. The formation reactions of the organism to environmental impact (ecogenetics). Ways of xenobiotics biotransformation. Tasks pharmacogenetics, phenotyping and ensuring the safety of drugs.
Question 2. The concept of pharmacogenomics.
Summary 2 questions. Association of gene-specific expression or single-nucleotide polymorphism in the human genome with drug efficacy or toxicity.
Question 3. Principles of pharmacogenetic testing.
Summary of 3 questions.
Predictive (personalized) medicine. Requirements for pharmacogenetic tests for their use in clinical practice. Indications for pharmacogenetic testing. Principles of interpretation of pharmacogenetic testing results. Principles of choice of drugs and their dosing regimens taking into account the results of pharmacogenetic testing.

Laboratory lesson 6. Pharmacogenetics and pharmacogenomics.
Summary of the laboratory session. Familiarity with farmakokineticheskimi tests. Solving clinical problems.
Independent work: Introduction to the Internet resource on clinical pharmacogenetics, search for pharmacogenetic information about the drug (on the instructions of the teacher) on the international nonproprietary name of the drug, and / or on the name of the gene: http://www.pharmgkb.org/
The continuation of the compilation of the pedigree.

Topic 7. Prenatal diagnosis, prevention of hereditary and congenital diseases

Lecture 7. Prenatal diagnosis, prevention of hereditary and congenital pathology.
Question 1. The main methods of prenatal diagnosis.
Summary of 1 question. Direct (invasive and non-invasive) and indirect methods of prenatal diagnosis.
Question 2. Prenatal screening for chromosomal diseases.
Summary 2 questions. Markers in fetuses with down syndrome.
Question 3. Prenatal screening for hereditary metabolic diseases.
Summary of 3 questions. Prenat tangible markers of hereditary pathology.
Laboratory lesson 7. Prenatal diagnosis, prevention of hereditary and congenital diseases.

Summary of the laboratory session. Methods of prenatal diagnosis, indications, diagnostic capabilities and disadvantages. To identify groups of pregnant women with an increased risk of having children with hereditary defects.
Independent work:
The continuation of the compilation of the pedigree. Preparation of multimedia presentations on the topic of the lesson.

Topic 8. Diagnosis of multifactorial diseases. Teratogenes. Ethical issues of medical genetics

Lecture 8. Ethical issues of medical genetics.

Question 1 Genetic testing for monogenic diseases and its risks.
Short content 1voprosa. Characteristics of the term genetic testing. Social, economic, family, personal risks-arising as a result of genetic testing.
Question 2. International legislation in the field of genetic testing.
Summary 2 questions. The main provisions of the universal Declaration on the human genome and human rights (UNESCO, 1997), the Convention for the protection of the rights and dignity of the person in connection with the application of biology and medicine (1997), the Convention on human rights and bio-medicine (1997), recommendations of the Committee on public and professional policies of the European society of human genetics, International Declaration on human genetic data (UNESCO International Bioethics Committee, 2003), Resolution 2004/9 of the economic and social Council of the United Nations Genetic privacy and non-discrimination.
Question 3. Fundamentals of the legislation of the Russian Federation in the field of genetic testing.
Laboratory lesson 8. Diagnosis of multifactorial diseases. Teratogenes. Summary of the laboratory session. Deals with the search of candidate genes. Their use in order to determine the risk of multifactorial disease or cancer process of a particular body.
Independent work:
Preparation of multimedia presentations on the topic of the lesson.