

ABSTRACT

OF THE ACADEMIC DISCIPLINE

"MODERN METHODS OF GENETIC DIAGNOSIS"

COURSE DESCRIPTION

The course "Modern methods of genetic diagnostics" is designed for 3rd year students. During the course, practical classes are held, covering a wide range of important problems of medical genetics.

PREREQUISITES AND CO-REQUISITES OF THE DISCIPLINE

Prerequisites. The study of the discipline involves the prior mastering of disciplines in medical biology, normal and pathological anatomy, normal and pathological physiology, biochemistry, microbiology.

Postrequisites. The main provisions of the discipline should be applied in the study of related disciplines during 3 years of study, is the basis for preparation for the licensing exam EDKI, preparation for study in higher education institutions in the programs of the third educational and scientific level of higher education.

THE PURPOSE AND OBJECTIVES OF THE DISCIPLINE

Objective: to provide training of highly qualified specialists in the field of medicine, able to solve complex problems of diagnosis of congenital and hereditary pathology.

The main objectives of the course are for students to acquire competencies in accordance with the general and professional competencies of the educational-professional program "Medicine" of the second level of higher education in the specialty 222 Medicine.

TOPICS OF PRACTICAL CLASSES

SECTION 1. SYNDROMOLOGICAL ANALYSIS. MODERN CYTOGENETIC METHODS OF DIAGNOSIS OF CONGENITAL AND HEREDIC PATHOLOGY.

Semantic section 1. Syndromological analysis.

Topic 1. Clinical and genealogical analysis. Methods of compiling a pedigree.

Topic 2. Syndromological analysis. Application of syndromological analysis in the diagnosis of hereditary pathology.

Semantic section 2. Cytogenetic methods of diagnosis of congenital and hereditary pathology.

Topic 3. Cytogenetic research methods in the clinic. Chromosomal abnormalities (numerical, structural).

Topic 4. Chromosomal polymorphism, chromosomal instability, gonadal mosaicism, single-parent disomy.

Topic 5. Molecular cytogenetic diagnostic methods (FISH). DNA sequencing.

SECTION II. MODERN BIOCHEMICAL METHODS OF DIAGNOSIS OF CONGENITAL AND HEREDIC PATHOLOGY.

Content section 3. Biochemical methods of diagnosis of congenital and hereditary pathology

Topic 6. Diagnosis of hereditary metabolic diseases.

Topic 7. Mass screening programs in the early diagnosis of hereditary pathology.

Topic 8. Selective screening programs in the diagnosis of SHO.

Topic 9. Modern methods of clarifying the diagnosis of SHO. Interpretation of high performance liquid chromatography results.

Topic 10. Modern methods of clarifying the diagnosis of SHO. Interpretation of gas chromatography - mass spectrometry results. Modern methods of clarifying the diagnosis of SHO. Interpretation of tandem mass spectrometry results.

SECTION III. MODERN MOLECULAR-GENETIC METHODS OF DIAGNOSIS OF HERITABLE PATHOLOGY.

Semantic section 4. Molecular genetic methods of diagnosis of hereditary pathology.

Topic 11. Modern methods of DNA diagnosis of hereditary pathology.

Topic 12. The latest technologies in molecular diagnostics (DNA analysis on microchips).

Content section 5. Prenatal diagnosis of congenital and hereditary pathology.

Topic 13. Methods of prenatal diagnosis.

Topic 14. Prenatal ultrasound diagnosis of congenital malformations.

Topic 15. Invasive methods of prenatal diagnosis.

Topic 16. Laboratory methods of prenatal diagnosis.