

Thematic scope of "**Clinical Genetics**" course

for medical students

academic year 2023/2024

Lectures

1. Basic concepts and definitions: DNA structure, DNA replication, transcription and translation, chromosome structure, cell cycle and cell division. Types of mutations and their consequences. Basic molecular diagnostic tests. Anomalies in the number and structure of chromosomes. Basic methods of cytogenetic diagnostics. Mechanism and etiology of developmental malformations. Rules for conducting family interviews and pedigree preparation.
2. Diseases caused by chromosomal abnormalities: autosomal aneuploidy, sex chromosome aneuploidy, polyploidies, deletion and microdeletion syndromes, syndromes associated with chromosome instability. The concept of epigenetics and genomic imprinting.
3. Monogenic inheritance (Mendelian): autosomal dominant, autosomal recessive, dominant and recessive sex-linked, atypical - discussion on examples of selected diseases.
4. Mitochondrial diseases conditioned by mitochondrial and nuclear DNA mutations. Metabolic diseases. Neurogenetics (selected epileptic syndromes, ataxias, neuromuscular disorders and progressive brain disorders).
5. Multigenic inheritance, relationships between genetic predisposition and environmental factors - multifactorial diseases. The development of genetics and new disease classifications and therapeutic options - personalized medicine: discussion on clinical cases. Summary of the block of lectures on clinical genetics – final colloquium test.

Seminars

The thematic scope of seminars remains related to the topic of lectures. Seminars are aimed at consolidating the thematic scope of lectures and learning the practical use of knowledge acquired during the lecture block.

Thematic scope of the seminars: Genetic counseling. Chromosome aberrations. Microdeletion syndromes. Sex differentiation disorders. Phacomatoses, neurogenetics. Genetic prenatal diagnosis. Oncogenetics.

Classes

The thematic scope of classes remains related to the topic of lectures and seminars. Classes are aimed at consolidating the thematic scope of lectures and seminars and also learning the practical use of knowledge acquired during the lectures and seminars blocks.

Thematic scope of classes: Pedigree analysis. Morphological phenotype analysis. Cytogenetics. Selected syndromes genetically determined in clinical practice.