

Approved at the methodic meeting
of the Department of Propaedeutic Pediatrics and Medical Genetics
on the August, 30, 2023 (Minutes No 1)

**MODERN METHODS OF GENETIC DIAGNOSTICS
(CURRICULUM)
(Medical Faculty, the 3rd Year of Study, 2023-2024)**

Lectures: 12 hours
Practice: 18 hours
Self-works: 60 hours
Total: 90 hours

Lecture Curriculum

No	Topic	Hours
<i>Part 1. Syndromological analysis</i>		
1	Clinical and genealogical analysis. Syndromological analysis. Application of syndromological analysis in the diagnosis of hereditary pathology	2
<i>Part 2. Cytogenetic methods of diagnosis of congenital and hereditary pathology</i>		
2	Cytogenetic research methods in the clinic.	2
<i>Part 3. Biochemical methods of diagnosis of congenital and hereditary pathology.</i>		
3	Diagnosis of hereditary metabolic diseases.	2
<i>Part 4. Molecular genetic methods of diagnostics of hereditary pathology.</i>		
4	Modern methods of DNA-diagnostics of hereditary pathology.	2
<i>Part 5. Prevention of congenital and hereditary pathology.</i>		
5	Levels of prevention of congenital and hereditary pathology	2
<i>Part 6. Principles of treatment of hereditary diseases.</i>		
6	Basic principles and methods of treatment of hereditary pathology	2
	<i>TOTAL for discipline</i>	12

Practice Curriculum

No	Topic	Hours
<i>Part 1. Syndromological analysis</i>		
1	Clinical and genealogical analysis. Method of compiling pedigree.	2
2	Analysis of familial morbidity. Methods of compiling a legend to the pedigree	2
3	Methods for calculating the risk of predisposition to hereditary and multifactorial diseases	2
	<i>TOTAL</i>	6
<i>Part 2. Cytogenetic methods of diagnosis of congenital and hereditary pathology</i>		
4.	Cytogenetic research methods in the clinic.	2
	<i>TOTAL</i>	2
<i>Part 3. Biochemical methods of diagnosis of congenital and hereditary pathology</i>		
5.	Diagnosis of hereditary metabolic diseases	2
	<i>TOTAL</i>	2
<i>Part 4. Molecular genetic methods of diagnostics of hereditary pathology</i>		
6.	Modern methods of DNA-diagnostics of hereditary pathology	2
	<i>TOTAL</i>	2
<i>Part 5. Prevention of congenital and hereditary pathology.</i>		
		2

7.	Mass neonatal screening programs.	2
8.	Programs of preconception prevention and prenatal diagnosis	2
	TOTAL	4
Part 6. Principles of treatment of hereditary diseases		
9	Modern methods of etiological and pathogenetic therapy of hereditary pathology	2
	TOTAL	2
	TOTAL for discipline	18

Self-work Curriculum

No	Topic	Hours
Part 1. Syndromological analysis		
1.	Method of compiling pedigree	2
2.	Methodology of examination of a patient with a suspicion of hereditary pathology. Conducting analysis of phenotypic features of proband and its family members	3
3.	Syndromological analysis. Application of syndromological analysis in diagnostics of hereditary pathology	2
	TOTAL	7
Part 2. Cytogenetic methods of diagnosis of congenital and hereditary pathology		
1.	The structure and functions of chromosomes.	2
2.	Cytogenetic research methods in the clinic.	2
3.	Chromosomal polymorphism, chromosomal instability, gonadal mosaicism, single-parental dysmosis	3
4.	Molecular-cytogenetic diagnostic methods (FISH)	3
5.	DNA sequencing	3
	TOTAL	13
Part 3. Biochemical methods of diagnosis of congenital and hereditary pathology		
1	Diagnosis of hereditary diseases of metabolism	3
2	Massive screening programs in the early diagnosis of hereditary pathology.	3
3	Programs of selective screening in diagnostics of HMD.	3
4	Modern methods of clarifying the diagnostics of HMD. Interpretation of the results of high performance liquid chromatography	3
5	Modern methods of clarifying the diagnostics of HMD. Interpretation of the results of gas chromatography - mass spectrometry	3
6	Modern methods of clarifying diagnostics. Interpretation of the results of tandem mass spectrometry	3
7	Interpretation of the results of enzyme diagnosis of lysosomal diseases accumulation	3
	TOTAL	21
Part 4. Molecular genetic methods of diagnostics of hereditary pathology		
1	Structure and functions of DNA	2
2	Modern methods of DNA-diagnostics of hereditary pathology.	2
3	New technologies in molecular diagnostics (DNA analysis on microchips)	3
	TOTAL	7
Part 5. Prevention of congenital and hereditary pathology		
1	Prenatal diagnosis of congenital and hereditary pathology.	3
2	Prenatal ultrasound diagnosis of birth defects	3
3	Invasive methods of prenatal diagnosis.	3
4	Laboratory methods of prenatal diagnostics.	3

	<i>TOTAL</i>	12
	<i>TOTAL for discipline</i>	60

Head of the Department of Propaedeutic Pediatrics
and Medical Genetics, Professor, MD

_____ Olena LYCHKOVSKA