THEMATICAL PLAN FOR PRACTICAL LESSONS

№	Topic	Hours
1.	Methods of genetic study – pedigree, twins study, cytogenetic and DNA analysis.	2
2.	Single gene mutations. Autosomal dominant pattern of inheritance: pedigree criteria, genetic risks, general phenotypic features, examples.	2
3.	Autosomal recessive pattern of inheritance: pedigree criteria, genetic risks, general phenotypic features, examples.	2
4.	X-linked pattern of inheritance: pedigree criteria, genetic risks, general phenotypic features, examples. Carrier detection: obligate carriers, screening for carrier state.	2
5.	Inborn errors of metabolism: prevalence, inheritance, common metabolic disorders. Population screening programs – newborn screening.	2
6.	Molecular-genetic methods. Principles and approaches for DNA analysis. Direct DNA analysis and applications.	2
7.	Indirect DNA analysis and application in genetic disorders.	2
8.	Unusual pattern of inheritance – genomic imprinting, uniparental disomy, cytoplasmic inheritance. Clinical significance and examples.	2
9.	Chromosomal disorders I. Chromosome mutations – types, mechanism of occurrence, classification. Cytogenetic method and clinical indications	2
10.	Chromosomal disorders II. Autosomal and sex chromosomal abnormalities: main clinical features, incidence, examples, cytogenetic variants, recurrence risk.	2
11.	Multifactorial inheritance. Genetics of common disorders in adult and congenital anomalies in newborn babies. Cancer genetics.	2
12.	Genetics and congenital abnormalities. Dysmorphology and teratogenesis.	2
13.	Midterm examination.	2
14.	Genetic counselling. Part I. Chromosome disorders.	2
15.	Genetic counselling part II. Single gene and multifactorial disorders.	2