**Synopsis in MEDICAL GENETICS for the academic year 2015-2016**

1. Methods for studying genetic disorders. Pedigree analysis: construction and ascertainment.
2. Methods for studying single-gene disorders. DNA analysis: DNA extraction, techniques – Southern blotting, PCR, RFLP analysis.
3. Methods for studying chromosomal disorders. Steps in cytogenetic preparation. Differential and selective staining, application.
4. Chromosomal disorders–incidence (spontaneous abortions, stillbirths, newborns); general clinical phenotypes (autosomal and sex chromosomal disorders).
5. Indications for cytogenetic analysis.
6. Chromosomal disorders – autosomal abnormalities.
7. Chromosomal disorders – sex chromosomal abnormalities.
8. Patterns of inheritance – autosomal dominant inheritance: characteristics of pedigree, genetic risks, clinical features, examples.
9. Patterns of inheritance – autosomal recessive inheritance: characteristics of pedigree, genetic risks, examples.
10. Patterns of inheritance – X-linked (dominant and recessive) inheritance: characteristics of pedigree, genetic risks, examples.
11. The inborn errors of metabolism (IEM) – prevalence, inheritance, biochemical basis of IEM, common clinical features, examples.
12. The inborn errors of metabolism – population (newborn) screening: criteria for a screening program.
13. Multifactorial inheritance – common disorders.
14. Multifactorial inherited congenital anomalies
15. Congenital anomalies – malformation, deformation and disruption. Examples. Etiology, incidence and clinical significance.
16. Congenital anomalies – syndrome, association, sequence. Examples. Etiology, incidence and clinical significance.
17. Classification of genetic disease. The impact and incidence of genetic disease.
18. Organization of human genome. Gene structure and function.
19. Mutations as cause of genetic disorders.
20. Haemoglobinopathies. Disorders of haemoglobin structure – types of mutations, general clinical features of structural variants of haemoglobin. Sickle cell disease.
21. Haemoglobinopathies.Disorders of haemoglobin synthesis. α- and β-thalassaemias.

22. Multifactorial inheritance. The liability / threshold model.
23. Recurrence risks in multifactorial disorders. Factors increasing risk to relatives in multifactorial disorders.
24. Inherited immunodeficiency disorders. Examples of primary immunodeficiency disorders (defects at specific stages of differentiation of stem cells).
25. Inherited immunodeficiency disorders. Examples of secondary (associated) immunodeficiency disorders.
26. Unusual pattern of inheritance. Uniparental disomy and genomic imprinting. Examples: Prader–Willi syndrome and Angelman syndrome
27. Unusual pattern of inheritance. Anticipation and triplet repeat expansion. Examples: Huntington's disease, Myotonic dystrophy and Fragile X syndrome.
28. Genetic heterogeneity. Allelic heterogeneity. Examples.
29. Genetic heterogeneity. Locus heterogeneity. Examples.
30. Pleiotropy. Examples. Variable expression and reduced penetrance. Examples.
31. Approaches for prevention of genetic disorders. Genetic screening (types). Maternal serum screening.
32. Genetic counseling – definition and goals. Indications for genetic referral.
33. Steps in genetic counseling.
34. Prenatal diagnosis, goals. Standard techniques used in prenatal diagnosis (techniques, optimal time in weeks, disorders diagnosed).
35. Cancer genetics – tumor suppressor genes. Retinoblastoma. Wilms' tumor.
36. Cancer genetics – oncogenes. Examples.
37. Cancer genetics – mendelian disorders with strong predisposition to cancer. Chromosome abnormalities and neoplasia.
38. Cancer genetics – common cancers: breast and colon cancers.