CLINICAL MOLECULAR GENETICS

Duration : 30 Hours	Exam Marks : 35

Module I: Introduction to human molecular genetics: — **03Hours**

- Basic Human Genetics and Genomics
- DNA as the genetic material, genetic code, flow of genetic information, mutation
- Principles of inheritance and probability rules
- Pedigree construction & family study
- Complications in pedigree analysis (variable expressivity, heterogeneity, penetrance, anticipation, epigenetics, mosaicism)

Module II:Non-Mendelian inheritance Polygenic inheritance — **04Hours**

- Multifactorial trait
- Threshold trait
- Genetic Susceptibility & risk factors P
- Mitochondrial genome and disorders
- Human genome, genome organization, annotations and databases, markers (microsatellites, SNPs) Pharmacogenomics,
- Mapping and identification of disease genes (linkage analysis, LOD score, association study)
- Allele frequency in population, estimation of carrier frequency

Module III: Basics of cell culture — **04Hours**

- Techniques of cell cultures (short term lymphocyte, primary and secondary cell cultures, maintenance of cell lines)
- Techniques of chromosome analysis
- Chromosome preparation from cultured lymphocytes, cell lines and solid tumors
- Karyotyping, C-,G-banding and fluorescence banding, nomenclatures of bandings.

Module IV: Chromosomal anomalies and disorders — **03Hours**

- Numerical (polyploidy, aneuploidy, autosomal, sex- chromosomal)
- Structural (deletion, duplication, translocation, inversion, isochromosome, ring chromosome)
- Chromosomal abnormalities in cancer

MODULE-V Clinical Genetics — **04Hours**

- Genetic Disorders
- Classification of genetic disorders
- Single gene Disorders (Cystic Fibrosis, Marfan's syndrome)
- Multifactorial disorders (Diabetes, Atherosclerosis, Schizophrenia)

MODULE-VI Molecular Techniques — **04Hours**

- PCR-RFLP
- ARMS-PCR
- Multiplex-PCR
- DNA Sequencing

MODULE-VII : Disease identification and Genetic tests for following disorders — **02Hours**

• Thalassemia, Sickle Cell anaemia, • Duchenne Muscular Dystrophy/Becker's Muscular Dystrophy, Huntington's disease • Allelic susceptibility test for multifactorial disorders (Neural Tube Defect, Cleft Lip and Palate, Cardio Vascular Disorder, Male infertility)	
MODULE-VIII	**03Hours**
• Prenatal and pre-implantation diagnosis. • Non-invasive prenatal testing for 13, 18 and 21	
MODULE-IX	**01Hours**
Molecularbasis of cancer	
MODULE-X Genetic Counselling	**02Hours**
• Principles of genetic counselling • Causes and factors for seeking counselling • Ethical and legal issues in genetic counselling	

CLINICAL MOLECULAR GENETICS PRACTICAL

Duration : 26 Hours	Exam Marks : 20

1. Universal safety precaution, importance of personal hygiene, disposal of organic waste washing, cleaning of glass ware, sterilization of glassware and lab ware
2. Drawing blood, preparation of blood film and identification of cells
3. Use of OMIM
4. Cleaning, sterilization and maintenance of culture chamber, cleaning and maintenance of laminar flow hood, preparation of culture medium
5. Collection and storage of samples (blood, biopsy material), culturing lymphocytes, stem cells
6. Culture of lymphocytes from blood
7. Metaphase chromosome preparations from blood
8. Chromosome preparation from lymphocyte culture
9. G-banding, C-banding , R-banding
10. Karyotyping
11. Detection of mutation using ARMS-PCR (e.g.; Thalassemia) and microsatellite markers (e.g.; fragile-X syndrome)
12. **Tests for genetic disorders**
    - SNP (e.g.; cleft lip and palate)
    - STS (e.g.; male infertility: Y-chromosome micro deletion)
    - Triplet repeat polymorphism (e.g.; Huntington's disease)
    - Molecular markers for tumor detection
    - Bcr-abl (RT-PCR)
13. Haplotyping of risk alleles in multifactorial disorders (PCR)