लोक सेवा आयोग

नेपाल स्वास्थ्य सेवा, मेडिसिन समूह, क्लिनिकल जेनेटिक्स उपसमूह, नवौं (९) तह, कन्सल्टेन्ट क्लिनिकल जेनेटिशिष्ट पदको खुला तथा आन्तरिक प्रतियोगितात्मक परीक्षाको पाठ्यक्रम

Paper II: Technical Subject

1. **Cytogenetics**
   - 1.1. Cell division
     - 1.1.1. Mitosis, meiosis
     - 1.1.2. Medical relevance of mitosis and meiosis
   - 1.2. Introduction to embryology and clinical teratology
     - 1.2.1. Human gametogenesis and fertilization
   - 1.3. Chromosomal Disorders
     - 1.3.1. Incidence and Mechanisms of Chromosomal disorder
     - 1.3.2. Chromosomal abnormalities.
     - 1.3.3. Types of chromosomes abnormalities.
     - 1.3.4. Chromosome abnormalities and malignancies.
     - 1.3.5. Chromosomal breakage studies and malignancies
   - 1.4. Human Karyotype
     - 1.4.1. Cytogenetic nomenclature
     - 1.4.2. Introduction of Cytogenetic methods.
     - 1.4.3. Indication of Cytogenetic analysis.
   - 1.5. Disorders of the Autosomes and Sex Chromosomes
   - 1.6. Genetic basis of cancer
   - 1.7. Applying Genomics to individualize Cancer Therapy
   - 1.8. Molecular cytogenetics
     - 1.8.1. Definition
     - 1.8.2. Types including FISH, Comparative Genomic Hybridization, Array CGH
2. **Molecular genetics**
   - 2.1. Gene
     - 2.1.1. Gene organization, structure and function
     - 2.1.2. DNA and RNA structure and function
   - 2.2. Fundamentals of gene expression
     - 2.2.1. Transcription, translation, proteomics, Integrative multi- 'omics'
   - 2.3. The Human Genome
   - 2.4. Gene structure and function
   - 2.5. Epigenetics and epigenomic aspects of gene expression
   - 2.6. Variation in gene expression and its relevance to medicine
   - 2.7. Genetic variation, inherited variation and polymorphism in DNA
   - 2.8. Mutations : Definition, types of mutations and their consequences, dynamic mutations
   - 2.9. Genotypes, phenotypes
     - 2.9.1. The Hardy –Weinberg Law
   - 2.10. Pattern of Single-Gene Inheritance : Pedigrees
   - 2.11. Inheritance : Pattern of Inheritance, Mendelian Inheritance, Types and Characteristics of Autosomal patterns of Mendelian Inheritance

2.12. Gene mapping, gene identification, positional cloning.
2.13. Sex- Linked Inheritance : Definition, Pseudo-autosomal Inheritance.
2.14. Mosaicism and Chimerism.
2.15. Pattern of origin effects on inheritance patterns.
2.16. Genetics of complex disorder
  2.16.1. Qualitative and quantitative traits.
  2.16.2. Familial aggregation and correlation.
  2.16.3. Determining the relative contributions of genes and environment to complex disease
2.17. Techniques of DNA analysis and application, DNA cloning, Genome sequencing
2.18. Mitochondrial Genetics : Definition, types of diseases associated with mitochondrial genetics
2.19. Evolutionary /Human Developmental Genetics

3. Special topics

3.1. Pharmacogenetics
  3.1.1. Biochemical modification and kinetics of drug metabolism
  3.1.2. Pharmacogenomics- Definition and importance in medicine
  3.1.3. Personalized medicine
3.2. Immunogenetics
  3.2.1. Different types of immunity and related diseases
  3.2.2. HLA polymorphism and disease association
  3.2.3. Importance and relation of HLA in transplant genetics
  3.2.4. Inherited immunodeficiency disorders
3.3. Blood grouping and molecular basis of blood grouping
  3.3.1. Hemoglobin and structure of globin gene
  3.3.2. Hemoglobinopathies- structure variants
  3.3.3. Hemolytic anemias, Thalassemia and sickle cell anemias
3.4. Cancer genetics
  3.4.1. Genetic and environmental factors in cancer.
  3.4.2. Oncogenesis and identification of oncogenes, tumor suppressor genes and its functions
  3.4.3. Epigenetics and cancer
  3.4.4. Common genetic associated cancer like colorectal carcinoma, breast carcinoma, prostate carcinoma and ovarian carcinoma, etc
  3.4.5. Genetic counseling in familial cancer
  3.4.6. Screening in familial cancer
3.5. Community genetics
  3.5.1. Genetic variation in population
  3.5.2. Genotypes and phenotypes in population
  3.5.3. Ethnic differentiation in the frequency of genetic diseases
  3.5.4. Genetics and ancestry

3.6. Molecular microbiology and metagenomics : Introduction, importance, uses in disease diagnosis and its pitfalls
3.7. Human genome project : Introduction and importance of human genome project
3.8. Stem cell biology and regenerative medicine and genetic cloning : Introduction and applications in modern medicine
3.9. Synthetic biology : Introduction and its uses
3.10. Animal models of genetic diseases.

4. Screening for genetic diseases and carriers

4.1. Criteria for screening
4.2. Prenatal screening: Indications for prenatal testing, techniques used in prenatal diagnosis
4.3. Special problems in prenatal diagnosis
4.4. Neonatal screening
4.5. Pre-symptomatic screening of adults
4.6. Ethical considerations in carrier detection and predictive testing

5. Congenital abnormalities, dysmorphology and learning disability

5.1. Incidence, Definition and classification of birth defects
5.2. Genetic causes of malformations
5.3. Environmental agents (teratogens) including drugs causing malformations
5.4. Malformations of unknown cause
5.5. Neurodevelopmental disabilities, global developmental delay,
5.6. Intellectual disability and autism

6. Inborn errors of metabolism

6.1. Disorders of amino acid metabolism
6.2. Disorders of carbohydrate metabolism
6.3. Disorders of steroid metabolism
6.4. Disorders of lipid and lipoprotein metabolism
6.5. Lysosomal storage disorders
6.6. Peroxisomal disorders
6.7. Disorders of metabolism of purines and pyrimidines
6.8. Disorders of metabolism of fatty acid and ketone body metabolism
6.9. Disorders of metabolism of porphyrins and heme metabolism
6.10. Disorders of metabolism of trace elements and metals
6.11. Disorders of energy metabolism

7. Chromosomal abnormalities

7.1. Incidence of chromosomal abnormalities
7.2. Trisomies: (Down syndrome, Edward syndrome, Patau syndrome)

7.3. Disorders of sex chromosomes: (Klinefelter syndrome, Turner syndrome, Fragile X syndrome)
7.4. Chromosomal deletion and microdeletion syndromes: (Prader Willi syndrome, Angelman syndrome, Retinoblastoma, DeGearge syndrome)
7.5. Chromosomal breakage syndromes: (Fanconi anemia, Ataxia telangiectasia, bloom syndrome, Xeroderma pigmentosa)
7.6. Disorders of sexual differentiation: (True hermaphroditism, Male pseudo hermaphroditism, Female pseudo-hermaphroditism)
7.7. Indications for chromosomal analysis

8. Genetic counselling

8.1. Definition
8.2. Indications for genetic counselling
8.3. Establishing the diagnosis
8.4. Pedigree analysis and calculating and presenting risk
8.5. Discussing the options
8.6. Communication including breaking bad news, handling complaints and communication with colleagues and support
8.7. Directive or non-directive Genetic counselling
8.8. Ethical, legal and social consideration in genetic counselling
8.9. Outcomes in genetic counselling
8.10. Special problems in genetic counselling: (Consanguinity, Incest, Adoption, Disputed paternity)

9. Reproductive genetics

9.1. Termination of pregnancy
9.2. Preimplantation genetic diagnosis
9.3. Assisted reproductive techniques and implications for genetic diseases
9.4. Assisted conception and the law
9.5. Prenatal treatment

10. Treatment of genetic diseases

10.1. Enzyme/Protein replacement therapy
10.2. Gene therapy
10.3. Drug therapy
10.4. Genetic engineering
10.5. Tissue transplantation/Stem cell therapy
10.6. Approach to diagnosis and treatment of genetic diseases
10.7. Basics of multidisciplinary management of genetic diseases

11. Ethical and legal issues in medical genetics

11.1. General principles
11.2. Ethical dilemmas in medical genetics

11.3. Ethical dilemmas in a wider context including ethical policies
11.4. Privacy of genetic information

12. Introduction to research methods

12.1. Scientific thinking, reading, critiquing including search and critical review of medical literature
12.2. Scientific writing
12.3. Bioinformatics tools for research
12.4. Information technology and the computer: Their role in research
12.5. Research designs including clinical trial.
12.6. Informed consent and confidentiality issues in conducting research
12.7. Planning experiments for appropriate statistical analysis
12.8. Statistical methods including genetic statistics and linkage analysis
12.9. Data presentation, clinical database and tools for clinicians
12.10. The ethics of use of human and animal subjects in research and related ethical dilemmas
12.11. Plagiarism and major research and publication misconducts
12.12. Detection of fraud and implication

13. Recent advances in clinical genetics