# **Syllabus for M.Phil in Human Genetics**

## HG-601 Paper-I Research Methodology in Human Genetics

#### **Unit-1: Chromosome Analysis**

Karyotyping, Automated Scanning System, Software studies, FISH, m-FISH, Different types of probes in FISH and its application, Staining Methods, Inverted microscope and its application.

#### **Unit-2: Molecular Analysis**

PCR, RT-PCR and its application, DNA sequencing-Manual and Automated, Microarray techniques, quantification and qualitative techniques and application.

### **Unit-3: Bioinformatics**

Genomics: Meiotic recombination and cytogenetic Maps, Chromosome Painting, Physical mapping of Genome, Data collection and interpretation, Mapping Databases, Sequence Database, Mapping of Whole genome, Ethical, Legal and Social Issues, Synthetic Genome.

Proteomics: Proteomics and protein function, 2D PAGE, Isotope coded affinity tags (ICAT), Determination the function of a protein - Phylogenic profile methods, Method of correlated gene neighbour, Analysis of fusion, Protein-Protein interaction, Protein Array.

### **Unit-4: Population Genetics**

Gene and Gene frequencies – Methods of measuring Genotype frequencies, Hardy Weinberg Law, Variation in Population, DNA sequencing polymorphism, Changes in Gene frequencies in population.

## **HG-602** Paper-II Clinical Genetics

## **Unit-1 Chromosomal Anomalies**

Microdeletion, Congenital anomalies, Chromosome marker (specific) Syndromes, X-linked mental retardation, Androgen insensitivity Syndrome, Infertility, Y-chromosome microdeletion.

#### **Unit-2 Metabolic Disorders**

Inborn errors of purine metabolism (Lesch-Nyhan Disease, Hyperuricemia in other HGPRT variants, PRPP synthetase abnormalities, Adenosine deaminase deficiency, Purine nucleoside phosphorylase deficiency, Xanthinuria)

Disorders of Amino acid metabolism, Disorders of carbohydrate metabolism, Disorders of lipid metabolism.

#### **Unit-3 Endocrine Abnormalities and Clinical Testing**

Congenital Adrenal Hyperplasia, Thyroid-Hyperthyroidism and Hypothyriodism, Enzyme deficiency disorders, Pseudohypoadosteronism, Pseudohypothyroidism, Testicular Feminization.

Testing in clinical genetics: Biochemical techniques (enzymes and hormones), tissue biopsy (reproductive, muscular etc), Physical examination, IQ test.

## **Unit-4 Cancer and Immunogenetics**

Oncogenes, Tumor suppressive genes, Molecular genetics of cancer, Autoimmune disorders, Immune deficiency syndrome- Chemistry, symptoms, therapy,HIV,H1N1.

## HG-603 Paper-III Nanotechnology and Stem cell Biology

## **Unit-1 Basics of Nanotechnology**

Introduction of Nanosciences and Nanotechnology, Types of Nanostructures, Synthesis of Nanostructural materials, Bionanotechnology, DNA nanotechnology, Protein based Nanostructures, Nanobiosensors for Biological Labelling and Cellular Imaging, Biosynthesis of Nanoparticle, Microbial Nanoparticle production.

## **Unit-2 Nanomedicines and its Importance**

Nanoparticles surface modification, Bioconjugation, Antibodies, Cell-Specific targeting and controlled drug release, Multi-functional Nanoparticles for Drug Delivery, Targeting through angiogenesis, Targeting organ or Tumor types, Tumor-specific targeting: Breast cancer, Liver, Targeting Tumor vasculature for Imaging, Delivery of specific anticancer agents: such as Paclitaxel, Doxorubicin, 5-Fluorouracil etc., Resent advances in Biomedical Nanotechnology.

## **Unit-3 Introduction to Stem cell Biology**

Introduction to Stem cell Biology, Types Characteristics, Isolation, and Maintenance, Application with recent perspective, Methodology, Cultures.

## **Unit-4 Advances in Stem cell Biology**

Regeneration medicines and stem cells, Stem cell and Epigenetics, Stem cell and Disease therapy, Stem cell and Immunomodulation, Stem cell and Genetics, Stem cell and Drug Discovery, Stem cell and Nanotechnology, Stem cell and Dentistry, Stem cell and Gamate Production.

## **HG-604 INTERNAL PAPER**

## It includes Seminars, Assignments etc.

#### **Suggested Reference Books:**

- 1. Human Cytogenetics: Constitutional Analysis, D E Rooney, 3<sup>rd</sup> edition.
- 2. Inborn Metabolic Diseases: Diagnosis and Treatment, Walter, 4<sup>th</sup> edition.
- 3. Medical Biochemistry, Baynes and Dominiczak, 2005
- 4. Chromosome, Sumner, 1990
- 5. Human cytogenetics: A practical approach (Vol. I & II), Rooney and Czepułkowski, 1992
- 6. Bioinformatics: Methods and protocols, Misner & Krawetz, 2000
- 7. Recent Advances in Bioinformatics, Khan & Kanum, 2001
- 8. Emery's Elements of medical genetics, Mueller and Young, 1998
- 9. Medical Instrumentation and their applications & design Webster.
- 10. Clinical Biochemistry: Clinical and Metabolic Aspects, 2<sup>nd</sup> Edition, Marshall and Bangert, Churchill, 2008.
- 11. Medical Biochemistry by Baynes and Dominiczak, 2<sup>nd</sup> Edition, 2005.
- 12. Basic and Clinical Endocrinology by Greenspan et al., 1983, ELBS, Singapore.
- 13. Endocrinology by De Groot Vol I, II & III, 2<sup>nd</sup> Edition, Saunders Publications, 1989.
- 14. Essentials Endocrinology by Laycock and Wise, 2<sup>nd</sup> Edition, ELBS, Singapore, 1981.
- 15. Immunology, 6<sup>th</sup> Edition by Kuby, Freeman Publications.
- 16. Cellular and molecular immunology by Abbas et al., 6<sup>th</sup> Edition, 2007, Elsevier Publications.
- 17. Elements of Immunology by Khan 2009, Dorling Kindsley, Delhi.
- Clinical Cancer Genetics, Risk counseling and management ,Kenneth-Liss, Wiley-Liss, 1998
- 19. Principles of cancer Genetics ,Fred Bung ,Springer,2008
- 20. Molecular Biology of Cancer, Lauren Pecorino, Oxford Uni. Press, 2008
- 21. Nanotechnology, Niemeger CM & CA. MIRLCIU, 2004
- 22. Bionanotechnology, D.S. Goodsell, 2004
- 23. Advanced Nanotechnology (I&II), S. K. Prasad, 2008
- 24. The Potential of Stem cells: An Inventory, Nikolaus Knoepffler, 2007