

PAPER-3.1: CLINICAL GENETICS AND GENETIC TOXICOLOGY

Unit – I

Scope of clinical genetics; Monogenic diseases- Cystic fibrosis, Tay-Sachs syndrome, Marfan syndrome; Polygenic diseases- Hyperlipidemia, Diabetes mellitus, Atherosclerosis; Inborn errors of metabolism and their genetic bases- Phenylketonuria, Maple syrup urine syndrome, Mucopolysaccharidosis, Galactosemia.

Unit – II

Neurogenetic disorders- Major regions of human brain and nerve conduction, Charcot-Marie-Tooth syndrome, spinal muscular atrophy; Syndromes due to triplet nucleotide expansion -Alzheimer's disease; Genetic disorders of Haemopoietic systems- Sickle cell anemia, Thalassemias, Hemophilias.

Unit – III

Origin of genetic toxicology; historical prospective of genetic toxicology; fundamentals of genetic toxicity; mechanism of induction of chromosomal alterations and sister chromatid exchanges; mutagens-chemical, physical, biological, environmental and food; antimutagens.

Unit – IV

Mechanisms of gene mutations; germinal mutations and human genetic diseases; mutations and cancers; genetic toxicology and congenital malformations; consequences of genotoxic effects in humans.

Recommended Books				
1	Cox & Sinclair	Molecular Biology in Medicine	Blackwell	1997
2	DeGrouchy & Turleau	Clinical Atlas on Human Chromosomes	Wiley	1984
3	Jankowski & Polak	Clinical Gene Analysis and Manipulation	Cambridge	1996
4	David Brusick	Principles of Genetic Toxicology		
5	Pasternak	An Introduction to Molecular Human Genetics	Fritzgarald	2000
6	Albert P, Li, RH,Heflich	Genetic Toxicology		
7	Rimoin <i>et al</i>	Principles & Practice of Medical Genetics, vol. I-III	Churchill	2002
8	Robinson & Linden	Clinical Genetics Handbook	Blackwell	1994
9	Strachan & Read	Human Molecular Genetics	Wiley	1999
10	Wilson	Clinical Genetics: A Short Course	Wiley-Liss	2000

PAPER- 3.2: DNA TECHNOLOGY AND GENETIC ENGINEERING

Unit – I

Enzymes used in DNA technology, Restriction and modification enzymes, Other nucleases, Polymerases, Ligase, kinases and phosphatases. Nucleic acids, Isolation and purification of DNA (genomic and plasmid) and RNA, Gel electrophoresis of nucleic acids (denaturing and native), Pulse-field gel electrophoresis of DNA.

Unit – II

Cloning vectors, Plasmids, Phages, Cosmids, Artificial chromosomes, Shuttle vectors, Expression vectors, Cloning techniques, Construction of genomic and cDNA libraries, Positional cloning: RFLP mapping, chromosome walking and jumping, Screening and characterization of clones, Preparation of probes, Restriction mapping, Principles of hybridizations and hybridization based techniques (colony, plaque, Southern, Northern and in situ hybridizations).

Unit – III

DNA sequencing, Oligonucleotide synthesis, Polymerase Chain Reaction and its applications, Microarray technology, ELISA, western and south-western blotting, Promoter characterization: promoter analysis through reporter genes, electrophoretic mobility shift assay, DNA foot-printing, DNA fingerprinting, Mutagenesis, Site directed mutagenesis, Transposon mutagenesis, Construction of knock-out mutants.

Unit – IV

Gene transfer techniques, Electroporation and microinjection, Transfection of cells: Principles and methods, Germ line transformation in *Drosophila* and transgenic mice: Strategies and methods. Applications of Recombinant DNA Technology, Crop and live-stock improvement, Molecular genetic analysis of human diseases, DNA drugs and vaccines, Biosafety and ethical considerations.

Recommended Books				
	Ausubel <i>et al</i>	Short Protocols in Molecular Biology	Wiley	2002
	Brown	Essential Molecular Biology vol. I	AP	2000
	Brown	Essential Molecular Biology vol. II	AP	2000
	Brown	Gene Cloning - An Introduction	Stanley Thornas	1995
	Glick & Pasternak	Molecular Biotechnology	ASM Press	1998
	Kracher	Molecular Biology - A Practical Approach		
	Primrose	Molecular Biotechnology	Panima	2001
	Reischel	Molecular Diagnosis of Infectious Diseases	Humana	1998
	Robertson <i>et al</i>	Manipulation & Expression of Recombinant DNA	AP	1997
	Twyman	Advanced Molecular Biology	Viva	1999
	Watson <i>et al</i>	Recombinant DNA	Freeman	1992
	Sandhya Mitra	Genetic Engineering Principles and Practice	Macmillan	1996

PAPER- 3.3: GENETIC SCREENING, COUNSELING AND GENE THERAPY

Unit – I

Scope of genetic screening- Prenatal and Post natal screening. Population screening for genetic diseases, family screening.

Unit – II

Prenatal screening methods- Amniocentesis- Chronic Villous sampling, Ultrasonography, fetoscopy, maternal blood sampling.

Post-natal screening- chromosomal abnormalities, cytogenetic disorders and molecular methods.

Unit – III

Scope of genetic counseling- methods of genetic counseling, educating the counselee, presenting the risks and options and guiding. Social, ethical and legal issues. Patterns of inheritance and risk assessment, chromosomal disorders, autosomal dominant and recessive disorders, X-linked disorders, multifactorial-polygenic disorders. Reproductive failures, consanguinity.

Unit – IV

Gene Therapy- classification of gene therapy- class I, II, and III. Types of gene therapy- germ line gene therapy and somatic gene therapy.

SUGGESTED READINGS:

1. Human Molecular Genetics by T. Strachan and AP Read
2. Human Genetics by F. Vogel and A.G. Motulsky
3. Genetic Engineering by Sandhya A. Mitra
4. Medical Genetics by Jorde et al
5. Genetic Counseling by W. Fuhrmann and F. Vogel

PAPER- 3.4: GENOMICS AND PROTEOMICS

Unit – I

Introduction to genomics, genetic mapping of human chromosomes, mapping of genetic disease locus to chromosome location, multilocus mapping of human chromosome, physical mapping of human genome, cloning human disease genes, human genome project.

Unit – II

DNA sequencing, bio chips, DNA micro arrays, gene annotation, gene structure predictions, gene ontology consortium recommendations, structural and functional genomics.

Unit – III

Protein structure and its determination, structural hierarchy, domains, folds, motifs. Secondary structure prediction methods, fold recognition and abinitio structure prediction, homology- comparative modeling of proteins.

Unit – IV

Protein chip arrays, functional proteomics, docking, rational drug design.

Suggested Readings:

1	Malcolm Campbell and Laurie J. Heyer	Genomics, proteomics and Bioinformatics	Benjamin Cummings	2002
2	Lynn B. Jorde et al	Encyclopedia of Genetics, Genomics, Proteomics and Bioinformatics	Wiley	2006

