M.Sc. MOLECULAR GENETICS

Course Structure - (w.e.f. 2009-2010)

Semester – I

Paper	Title of the paper	Internal	Semester End	Total	Credits
1.1	Principles of Genetics	15	85	100	4
1.2	Cell Biology	15	85	100	4
1.3	Medical Biochemistry	15	85	100	4
1.4	Molecular Genetics	15	85	100	4
1.5	Practical - I	--	100	100	4
1.6	Practical - II	--	100	100	4

Semester – II

Paper	Title of the paper	Internal	Semester End	Total	Credits
2.1	Human Cyto and Medical Genetics	15	85	100	4
2.2	Population Genetics and Biostatistics	15	85	100	4
2.3	Genomics and Proteomics	15	85	100	4
2.4	DNA Technology and Genetic Engineering	15	85	100	4
2.5	Practical - III	--	100	100	4
2.6	Practical - IV	--	100	100	4
2.7	Basic Human Genetics (Optional Paper/Non-core subject - for other Departments)	15	85	100	4

Semester – III

Paper	Title of the paper	Internal	Semester End	Total	Credits
3.1	Biochemical and Immunogenetics	15	85	100	4
3.2	Bioinformatics	15	85	100	4
3.3	Prenatal Diagnosis and Gene Therapy	15	85	100	4
3.4	Somatic Cell and Cancer Genetics	15	85	100	4
3.5	Practical -V	--	100	100	4
3.6	Practical -VI	--	100	100	4
3.7	Advanced Human Molecular Genetics (Optional Paper/Non-core subject - for other Departments)	15	85	100	4

Semester – IV

4.1	Dissertation based on Project work			100	5
4.2	Comprehensive Viva – Voce			100	5

Grand Total : I – IV Semesters 2200 -90

I – SEMESTER

Paper-1.1: Principles of Genetics

Unit – I

Introduction to Genetics Mendelism, Mendel and his experiments, Law of segregation, Law of independent assortment, Application of laws of probability (product rule, sum rule), Chromosomal basis of segregation and independent assortment

Unit – II

Extensions of Mendelism, Allelic variation and gene function- Dominance relationships, basis of dominant and recessive mutations, Multiple allelism, allelic series, Testing gene mutations for allelism: complementation test, intragenic complementation, Visible, sterile and lethal mutations, Genotype to phenotype: effect of the environment on phenotype development- Penetrance and expressivity, phenocopy, Gene interactions and modifying genes, Pleiotropy.

Unit – III

Sex-linked inheritance, Linkage and crossing over, Genetic recombination and construction of genetic maps in *Drosophila*, Interference and coincidence,Cytological demonstration of crossing over in *Drosophila*, Mitotic recombination, Intragenic recombination.

Unit – IV

Inheritance of quantitative traits, Continuous and discontinuous variation, Polygenic inheritance, Genetic variance, heritability (narrow sense and broad sense), Cytoplasmic inheritance, maternal effects, inheritance due to parasites and symbionts.

Recommended Books				
1.	Atherly *et al*	The Science of Genetics	Saunders	1999
2.	Brooker	Genetics – Analysis and Principles	Benjamin/Cumings	1999
3.	Fairbanks *et al*	Genetics	Wadsworth	1999
4.	Gardner *et al*	Principles of Genetics	John Wiley	1991
5.	Griffiths *et al*	Modern genetic Analysis	Freeman	2002
6.	Griffiths *et al*	An Introduction to Genetic Analysis	Freeman	2004
7.	Hartl & Jones	Genetics – Principles and Analysis	Jones & Bartlett	1998
8.	Snustad *et al*	Principles of Genetics	Wiley and sons	1998
9.	Strickberger	Genetics	Mcmillan	1985
10.	Tamarin	Principles of Genetics	WCB	1996
11.	Peter Sydbery	Human Molecular Genetics Second Ed.,	Pearson Education	2002
12.	Avinash&K.Upadhyay	Fundamentals of Molecular Biology	Himalaya Publishing House	2005

Paper – 1.2: Cell Biology

Unit - I

Plasma Membrane: Fluid mosaic model, structure of membrane, transport across membrane and mechanisms of endocytosis and exocytosis, Endomembrane system, General organization of protein transport within and outside the cell **,**Protein sorting and secretion , Mechanism of intracellular digestion.

Unit – II

Cytoskeleton, Microfilaments: Structural organization, cell motility and cell shape, Microtubule: Structural and functional organization, cilia, flagella, centriole, Intermediate filaments, Mitochondria, Ultrastructure, Chemiosmotic theory and respiratory chain complexes.

Unit - III

Structure and function of peroxisome, Structure and biosynthesis of ribosome , Cell cycle and its regulation, Cell-Cell Interaction , Cell adhesion molecules, Cellular junctions, Extracellular matrix.

Unit – IV

Signal transduction, Intracellular receptor and cell surface receptors, Signalling via G-protein linked receptors (PKA, PKC, CaM kinase), Enzyme linked receptor signaling (Growth factor receptor signaling; JACK-STAT pathway), Network and cross-talk between different signal mechanisms, Programmed cell death

Recommended Books				
1.	Alberts *et al*	Essential Cell Biology	Garland	1998
2.	Alberts *et al*	Molecular Biology of the Cell	Garland	2002
3.	Cooper	The Cell: A molecular Approach	ASM Press	2000
4.	Gilbert	Developmental Biology	Saunders	2003
5.	Jeremy *et al*	Biochemistry	Freeman	2002
6.	Karp	Cell and Molecular Biology	John Wiley	2002
7.	Lewin	Genes VIII	Pearson	2004
8.	Lodish *et al*	Molecular Cell Biology	Freeman	2004
9.	Pollard & Earnshaw	Cell Biology	Saunders	2002
10.	Tobin & Morcel	Asking about Cells	Saunders	1997
11.	Wilson & Hunt	The Cell: A Problems Approach	Garland	2002

Paper –1.3: Medical Biochemistry

Unit - I

Carbohydrates: Mucopolysaccharides and related disorders, Glycolysis, Krebs cycle Gluconeogenesis, Glycogenesis and glycogenolysis, Disorders of glycogen metabolism-Galactosemia**.**

Lipids: Fatty acids: synthesis and oxidation of fatty acids, Ketogenesis, Metabolism of cholesterol , Lipoproteins: role in lipid transport and storage Prostaglandins: structure and function. Familial Hypercholesteremia.

Unit - II

Amino acids and peptides

Essential and non-essential amino acids, Amino acids related disorders

Small peptides and their biomedical importance. Proteins: Structure-conformation-function relationship (exemplified by Myoglobin, Hemoglobin, and Collagen) Protein degradation, Enzymes: General properties, Enzyme inhibition , Mechanism of action and Regulation of enzyme activity. Phenylketonuria, Alkaptonuria.

Unit - III

Bioenergetics: Second law of thermodynamics, High energy compounds and Oxidative Phosphorylation. Hormones: Characteristics, Mechanism of action of peptide and steroid hormones, Hormone receptors and diseases. Vitamins : water and lipid soluble- their structure and function.

Unit - IV

Nucleic Acids: structure and conformations, Nucleotide Metabolism, Synthesis and degradation of pyrimidine and purine nucleotides and Disorders of nucleotide metabolism.

Recommended Books				
1.	Berg *et al*	Biochemistry	Freeman	2002
2.	Bhagwan	Medical Biochemistry	AP	2004
3.	Devlin	Biochemistry: with clinical correlations	Wiley	2002
4.	Higgins *et al*	Biochemistry for the Molecular Sciences	Longman	1994
5.	Murray *et al*	Harpers Illustrated Biochemistry	Prentice Hall	2003
6.	Nelson *et al*	Lehninger’s Principles of Biochemistry	McMillan	2000
7.	Voet & Voet	Biochemistry Vol. 1 and 2	Wiley	2004
8.	Zubay *et al*	Principles of Biochemistry	WCB	1995
9.	S. N. Jogdand	Medical Biotechnology	Himalaya Publ.	2008

Paper – 1.4: Molecular Genetics

Unit-I

An overview of molecules involved in the flow of genetic information; Double helical structure of DNA, Alternate forms of DNA double helix, Denaturation and renaturation of DNA, DNA binding proteins, factors affecting DNA stability. Types and structure of RNA, RNA-DNA hybrid helices.

Unit-II

Genome Replication: Experimental proof of the Watson–Crick scheme for DNA replication, The Messelson-Stahl Experiment, The topological problems, Mechanisms of replication in prokaryotes and eukaryotes, DNA polymerases and topoisomerases, Regulation of eukaryotic replication, DNA modification systems, DNA repair systems.

Unit-III

Transcription Machinery: Prokaryotic RNA polymerases, Promoter, Initiation and termination, Eukaryotic RNA polymerases, transcription factors and promoters. Posttranscriptional processing of RNA, 5' capping 3' poly-adenylation, splicing, RNA editing. Genetic Code: General features of the genetic code. Degeneracy and universatility of genetic code.
Translation Machinery: RNA, prokaryotic and eukaryotic ribosomes, Activation of amino acids, Initiation, elongation and termination of polypeptide chains, Inhibitors of protein synthesis, Post-translation modifications. , Structure of proteins

Unit-IV

Regulation of transcription initiation, operon and regulon, positive and negative regulation, enhancers and promoters Attenuation and antiterminations, post transcriptional regulation, alternate splicing, Gene silencing, Transport and targeting of RNA, Regulation of translation.

Books Recommended:

1	Alberts, B., Bray, D., Lewis, J., Raff, M., Roberts, K. and Watson, J.D.	Molecular Biology of the Cell	Garland Press	1999
2	Brown, T.A	Genomes:	Wiley	1999
3	Lewin, B.	Genes VII	Oxford	1997
4	Lodish, H., Baltimore, D., Berk, A., Zipursky, S.L., Matsudaira, P. and Daniell, J.	Molecular Cell Biology	Freeman and Co	2000
5	Singer, M. and Berg, P	Genes and Genomes	Blackwell Scientific	1991
6	Weaver, R.F.	Molecular Biology	McGraw Hill	2002

Paper 1.5: PRACTICAL – I

Part – A

1. **Genetics of Blood Groups**
    a) ABO –typing
    b) Rh (D) typing
    c) ABH typing

2. **Genetic Traits**
    a) Colour Blindness
    b) Phenyl Thio Carbamide (PTC)
    c) Dermatoglyphics

Part – B

1. Spotters – Eukaryotic Cell Organelle-Membrane, Nucleus, Nucleolus, Mitochondria, Endoplasmic Reticulum, Golgi apparatus, Chloroplasts, Ribosomes.

2. Mitotic Chromosomes in Onion Root Tips

3. Meiotic Chromosomes in Onion Flowers

Paper 1.6: PRACTICAL – II

Part – A

1. Red Cell Enzyme estimations
2. Plasma Protein estimations
3. Estimation of cholesterol
4. Estimation of urea
5. Paper chromatography for separation of carbohydrates
6. Paper chromatography for separation of Amino Acids

Part – B

1. Isolation of DNA from peripheral blood
2. Isolation of DNA from Leaf
3. Isolation of DNA from Tissue
4. Isolation of RNA from Lymphocytes
5. Quantification of Genomic DNA
6. Quality check of Genomic DNA

II – SEMESTER

Paper 2.1: Human Cyto and Medical Genetics

Unit – I:

History and development of Human Cytogenetics. Different conferences and chromosomal nomenclature. Setting up of cultures, harvesting and making chromosomes – Karyotyping. Chromosomes in mitosis and meiosis and interphase. Non-disjunction.

Unit – II:

Chromosomal abnormalities – Numerical and structural leading to syndromes – Monosomy, trisomy, triploidy, tetraploidy, Turner, Klinefelter, Down, Edwards, Patau, cri-du-chat, Females with multiple X chromosomes and males with XYY chromosomes. Gonadal dysgenesis, true hermophrodites, testicular feminization, chromosomes in malignancies and spontaneous abortions. Fragile X syndrome, genetics of fragile X syndrome, molecular biology of fragile X chromosome.

Unit – III:

Scope of Medical Genetics. Skin- Ichthyosis, baldness, psoriasis, hereditary Hemorrhagic telangiectasia, epiloia, multiple neurofibromatosis, the porphyrias, blooms syndrome. The skeletal system – Marfan's syndrome, Nail patella syndrome, Brachydactyly, syndactyly, Polydactyly, Spina bifida and anencephaly, Ankylosing spondylitis, Rheumatoid arthritis, Osteogenesis imperfecta. Muscle – Muscular dystrophies, Myotonia.

Unit – IV:

Eye – Glaucoma, ptosis, squint, nystagmus, cataract, retinoblastoma. Jaws – Hare lip and palate. Ears - Deafness. Alimentary system – Hypertrophic pyloric stenosis, gastric and duodenal ulcers, peptic ulcers, cirrhosis of liver. Respiratory system – Cystic fibrosis.
Cardio vascular system – congenital heart disease, coronary heart diseases and Hypertension Central nervous system – The ataxias and familial spastic paraplegia. Kidney and urinogenital tract – Cystinosis, polycystic kidney disease.Endocrine system – Cretinism, goiter, diabetes.

SUGGESTED READINGS:

a. Human Genetics by A.G. Motulsky and F. Vogel
b. Medical Genetics by Lynn B. Jorde et al
c. Genetic counseling by Fuhrman and F. Vogel
d. Text book of Human Genetics by Fraser and Mayo
e. Molecular structure of Human Chromosome by J J Yunis
f. Human Cytogenetics (Vol I and II) by J.L. Hamerton

Paper 2.2 : Population Genetics and Biostatistcs

Unit – I:

Mendelian Population and scope of population genetics. Gene and genotype frequencies, mating patterns, Hardy-Weinberg principle, heterozygotes, extention of H-W principle to multiple alleles, sex-linked alleles. Non-random matings, inbreeding and assortative matings, inbreeding coefficient. Factors that change allelic frequencies.

Unit – II:

Genetic polymorphism, transient and stable and factors responsible stable polymorphism. DNA markers and populations differences. Application of population genetics. Role of population genetics in genetics counseling. Genetics origin and evolution of human races. Genetic Demography, age and gender specific death and birth rates, intrinsic rate of natural increase. Index of opportunity for natural selection.

Unit – III:

Importance of population studies, sampling techniques, classification of data and tabulation. Measures of central tendency- mean, median and mode. Measures of dispersion - variance and standard deviation.

Unit – IV:

Probability – laws of probability for independent events – permutations and combinations – conditional probability. Binomial distribution. Tests of significance – chi square test and 't' test.

Books suggested:

1. The Genetics of Human Populations by LL Cavalli-Sforza and WF Bodmer

Freeman and Company, 1971.

2. Population Genetics Theory by James F. Crow and W. Kimura, Harper and Row, 1970.

3. Introduction to Biostatistics by P.S.S. Sundara Rao and J. Richard, New Delhi

4.Introduction to Biostatistics by Robert R. Sokel and James F. Kohlf

5. Quantitative Genetics 4th Ed., -Douglas S. Falconer and FC Mackay (Pearson Eduction)

Paper 2.3: 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	BenjaminCummings	2002
2	Lynn B. Jorde et al	Encyclopedia of Genetics, Genomics, Proteomics and Bioinformatics	Wiley	2006
3	V.S. Gomase & .J.Chikhale	Proteomics Theory and Practice	Himalaya Publishing House	2009
4	V.S. Gomase & .J.Chikhale	Bioinformatics Theory and Practise	Himalaya Publishing House	2007

Paper 2.4: 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) , Expression based screening, Interaction based screening: yeast two-hybrid system

Unit – III

S1 nuclease and RNase mapping, 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, Gene therapy: somatic and germ line gene therapy, DNA drugs and vaccines, Biosafety and ethical considerations

Suggested Readings

Recommended Books				
1.	Ausubel *et al*	Short Protocols in Molecular Biology	Wiley	2002
2.	Brown	Essential Molecular Biology vol. I	AP	2000
3.	Brown	Essential Molecular Biology vol. II	AP	2000
4.	Brown	Gene Cloning - An Introduction	Stanley Thornas	1995
5.	Glick & Pasternak	Molecular Biotechnology	ASM Press	1998
6.	Kracher	Molecular Biology - A Practical Approach		
7.	Krenzer & Massey	Recombinant DNA and Biotechnology	ASM	2000
8.	Micklos & Freyer	DNA Science	CSHL	1990
9.	Primrose	Molecular Biotechnology	Panima	2001
10.	Reischel	Molecular Diagnosis of Infectious Diseases	Humana	1998
11.	Robertson *et al*	Manipulation & Expression of Recombinant DNA	AP	1997
12.	Sambrook *et al*	Molecular Cloning Vol. 1, 2, 3	CSHL	2001
13.	Twyman	Advanced Molecular Biology	Viva	1999
14.	Watson *et al*	Recombinant DNA	Freeman	1992
15.	P,K. Gupta	Molecular Biology and Genetic Engineering	Rastogi Publ.	2008
16.	S. RamReddy K.Venkateswarlu V. Krishna Reddy	A Text Book of Molecular Biotechnology	Himalaya Pub. House	2007
17.	P,K. Gupta	Genetics	Rastogi Publ.	2009

Paper 2.5: PRACTICAL – III

Part – A

1. Chromosome Nomenclature
2. Karyotyping
3. Metaphase drawing
4. Drumstick and Barr body identification
5. Demonstration of Human blood lymphocyte culture :

    a. Washing and sterilization of glassware
    b. Medium preparation
    c. Setting up of lymphocyte culture
    d. Harvesting and slide preparations
    e. Identification of individual chromosomes
    f. Preparation of G-banded chromosomes

Part – B

(Assignment)

1. Measures of Central Tendency
2. Measures of Dispersion
3. Correlation
4. Probability
5. Binomial Distribution
6. Tests of Significance
7. Gene frequencies-Hardy Weinberg Equilibrium
8. Genetic Polymorphism
9. Mutational Rates

Paper 2.6 : PRACTICAL – IV

Part – A

1. Primer designing
2. Insertion deletion polymorphism
3. DNA Finger printing – RFLPs and VNTRs
4. Amplification and purification of DNA fragments
5. ARMS-PCR
6. Multiplex PCR
7. Nested PCR
8. DNA sequencing methods

Part – B

1. Preparation of Plasmids
2. Restriction digestion and mapping
3. Ligation of DNA fragments
4. Transformation
5. Bacterial cultures
6. Construction of cDNA libraries
7. Purification and identification of clones
8. Expression of cloned genes
9. Construction of genomic libraries
10. Purification and identification of recombinant plasmid DNA
11. Blotting Techniques: a) Southern b) Western c) Northern

Paper 2.7: Basic Human Genetics

(*Optional Paper / Non Core Subject)*

UNIT- I :

Introduction to genetics Mendelism and Mendels laws of inheritance; Dominant and Recessive mutations: Multiple allelism; Penetrance and expressivity; Phenocopy and Pleotrophy; Sexlinked inheritance; Linkage and Crossing over; Poly geneic inheritance.

UNIT- II :

Scope of population genetics; gene and genotype frequencies; H.W. principle; genetic polymorphism (balanced and transient); Random matting and its consequences; DNA markers and population differences; Inbreeding – types of consanguineous marriages..

UNIT- III :

Role of population genetics in genetic councelling; Scope of genetic screening: Prenatal and postnatal screening; Population screening for genetic diseases; Family screening; Prenatal and postnatal screening methods.

UNIT- IV :

The concept of biochemical polymorphism, enzyme and protein polymorphisms- ACP1and HP; Hemoglobinopathies;Metabolic disorders-Phenylketonuria, Alkaptonuria, Albinism and Homocystinuria; Hereditary disorders with altered drug resposes-G6PD.

Suggeted Books:

1, Principles of Human Genetics – Curt Stern
2. Human Genetics – F. Vogel and A. G. Motulsky
3. Basic Human Genetics – Mange and Mange
4. Genetic Counselling - W. Fuhramann and F. Vogel
5. Genetics of Human Populations – Cavallis S. forza and Bodmer
6. Principles of Human Biochemical Genetics – H. Harris

III SEMESTER

Paper 3.1: Biochemical and Immunogenetics

Unit – I

The concept of Biochemical polymorphism, enzyme and protein polymorphisms – Hemoglobin, Acid Phosphotase and Haptoglobin. Metabolic disorders, Phenylketonuria, Hypercholesteremia, Lasch Nyhan Syndrome, Orotic aciduria, mukopolysaccharidoses, DNA studies of PKU and Hemoglobinopathies.

Unit – II

Immune responses – innate immune system and adaptive immune system. Immunoglobulins. The major histocompatability complex – HLA and Complement system.Human blood group systems. Immunodeficiency diseases – autoimmunity and acquired immunodifficiences. DNA level studies in HLA systems.

Unit – III:

Genetics in drug metabolism. Genetic Variations revealed solely by the drug – succinyl choline sensitivity. Hereditary disorders with altered drug responses – G-6-PD deficiency.

Unit – IV

Ecogenetics – alpha -1- antitripsin.

SUGGESTED READINGS:

1. Principles of Human Biochemical Genetics by H. Harris

2. Human Genetics by A.G. Motulsky and F. Vogel

3. The metabolic basis of inherited diseases by Scriver et al.

4. Medical Genetics by Lynn B. Jordee et al

Paper 3.2 : Bioinformatics

Unit – I

Basics of computers (CPU, I / O units) and operating systems. Introduction and scope of Bioinformatics. Computer networking, Internet and e-mail, concept of home pages and web-sites, www, uniform resource locations

Unit – II

Archiving and retrieval of information- search ingines, data bases, medline, NCBI, nucleic acid sequences, genomes, protein sequence and structures, Bibliographic

Unit – III

Access to molecular biology data bases: Entrez, Sequence Retrieval System (SRS), protein identification resources (PIR), sequence alignments and phylogenetic trees.

Micro arrays and genome wide expression analysis. Pharmacogenomice, patenting, Intellectual property rights and bioinformatics patents.

Unit – IV

Human Genome Project: Goals of HGP, the human genetic material, benefits from HGP, Ethical, legal and social implications of HGP. Sequence of Human Genome. Bioethics and Genethics.

SUGGESTED READINGS:

1	Attwood and Parry Smith	Introduction to Bioinformatics	pearson	2002
2	Barnes and Gray (ed)	Bioinformatics for Geneticists	Wiley	2003
3	Lesk	Introduction to Bioinformatics	Oxfore	2003
4	Mount	Bioinformatics: Sequence and analysis	CBC	2003
5	Rashidi and Buchler	Bioinformatics Baiscs	CBC	2000
6	Rastogi et al	Bioinformatics: Concepts, skills and applications	CBC	2003
7	Westhead et al	Bioinformatics Instant Notes	Viva Books	2003
8	Azariah et al	Bioethics in India	Eubios Ethics	1998
9	Bryant et al	Bioethics for Scientists	Wiley	2002

Paper 3.3: Prenatal Diagnosis and Gene Therapy

Unit – I

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

Unit – II

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 – III

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

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

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 Jordee et al
5. Genetic Counseling by W. Fuhramann and F. Vogel
6. Genetics in Medicine by Thomson and Thomson.

Paper 3.4: Somatic Cell and Cancer Genetics

Unit – I

Somatic cell genetics: Cell cultures, somatic cell hybridization, making cell lines with different chromosomes, use of somatic cell hybrids in gene mapping. Other methods of transfer of genetic information. Complementation, mutations in cell cultures, studies of differentiated cell functions.

Unit – II

Cancer Genetics: Characteristics of cancer cells, Chromosomes in neoplasias, cancer as a genetic disorder, cancer in families, loss of cell cycle control. Inherited versus sporadic cancers.

Unit – III.

Molecular changes in proto-oncogenes, tumor suppressor genes-Knudson's Hypothesis, Retinoblastoma, Lung cancer, Colon cancer, Brain cancer, Breast cancer, Prostrate cancer, cervical and esophageal cancers.
Cancer and environment: physical, chemical and biological carcinogens.

Unit – IV

Cancer prevention, Diet – cancer associations, diagnosis and treatment.

Recommended Books				
1.	Alberts *et al*	Molecular Biology of the Cell	Garland	2002
2.	Cowell	Molecular Genetics of Cancer	Bios	2001
3.	Ehrlich	DNA Alterations in Cancer	Eaton	2000
4.	Gersen & Keagle	Principles of Clinical Cytogenetics	Humana	1999
5.	Lewin	Genes VIII	Pearson	2004
6.	Lodish *et al*	Molecular Cell Biology	Freeman	2004
7.	Stillman	Molecular Genetics of Cancer	CSHL	1994
8.	Thompson and Thompson	Genetics in Medicine	Saunders	1986
9.	Ricki Lewis	Human Genetics – Concepts and applications	McGraw Hill	
10	S.N. Jogdand	Medical Biotechnology	Himalaya Publishers	2008

Paper 3.5: PRACTICAL – V

Part – A

1. Sickling Test
2. Separation of abnormal heamoglobins
3. Estimation of Hb F
4. Estimation of Hb A2
5. Red Cell Enzymes – ACP, ESD, and GLO1
6. Plasma proteins – HP, CP, ALB and TF
7. ELISA Test
8. Radial immunodiffusion
9. Quantitative precipitin Assay
10. Immunoelectrophoresis

Part – B

1. Literature search from NCBI.
2. Working with Genome Databases
3. Working with Protein Databases
4. Similarity search using BLAST
5. Working with Bioinformatics tools
    a. Rasmol
    b. SwissPDB
    c. Hex
    d. Clustal X\W
6. Construction of Phylogenic Trees

Paper 3.6: PRACTICAL – VI

Part – A

(Assignment)

1. Genetic Councelling methods
2. Prenatal screening methods
3. Postnatal screening methods
4. Patterns of inheritance
5. Councelling for single gene disorders (Case studies)
6. Councelling for multifactorial disorders (Case studies)

Part – B

Tissue culture
Procedure
Types

Paper 3.7: Advanced Human Genetics

(*Optional Paper / Non Core Subject)*

UNIT- I :

History and development of cytogenetics; Morphological variability of human chromosomes; Different conferences and chromosomal nomenclature; Nondisjunction; Numerical abnormalities leading to syndromes; Chromosomal structural abnormalities; Setting up of cultures, harvesting and making chromosomes; Karyotyping; Human chromosome banding techniques.

UNIT- II :

Immune responses-Innate immune system and Adaptive immune system; Complement system; Genetic basis of Ig structure and diversity; Antibody diversity; Major histocompatability complex; Immunodeficiency diseases.

UNIT- III :

Double helical structure of DNA; Alternative forms of DNA; Types and structure of RNA; Mechanism of DNA replication in prokaryotes and eukaryotes; Regulation of eukaryotic replication; DNA modification systems; DNA repair systems; Transcription, Translation and Regulation.

UNIT- IV :

Enzymes used in DNA technology; Restriction and modification enzymes, nucleases, polymerases, ligases, kinases and phosphatases; Nucleic acids isolation and purification; Gel electrophoresis of nucleic acids (denaturing and native); Polymerase chain reaction; Blotting techniques(Southern and Northern); DNA sequencing.

Suggested Books:

1. Human Cytogentics - J. L. Hamerton
2. New Chromosomal Syndromes – J.J. Yunis
3. Molecular Structure of Human Chromosome – J.J. Yunis
4. Molecular Biology and Genetic Engineering – P. K. Gupta (Rastogi Publ)
5. Basic Immunogenetics – Fuden Berg et al.,
6. Molecular Biology - M.P. Arora (Himalaya Publishing House)
7. Fundamentals of Molecular Biology – Avinash & K Upadhaya