# M.Sc. Molecular Genetics

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

### Semester – I

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(Optional Paper/Non-core subject - for other Departments)

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(Optional Paper/Non-core subject - for other Departments)

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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

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

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<th>Publisher</th>
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<tr>
<td>1.</td>
<td>Atherly et al</td>
<td>The Science of Genetics</td>
<td>Saunders</td>
<td>1999</td>
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<td>Brooker</td>
<td>Genetics – Analysis and Principles</td>
<td>Benjamin/Cumings</td>
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<td>Fairbanks et al</td>
<td>Genetics</td>
<td>Wadsworth</td>
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<td>Griffiths et al</td>
<td>Modern genetic Analysis</td>
<td>Freeman</td>
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<td>Griffiths et al</td>
<td>An Introduction to Genetic Analysis</td>
<td>Freeman</td>
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<td>Strickberger</td>
<td>Genetics</td>
<td>Mcmillan</td>
<td>1985</td>
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<td>Tamarin</td>
<td>Principles of Genetics</td>
<td>WCB</td>
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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


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

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<td>5. Jeremy <em>et al</em></td>
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<td>7. Lewin</td>
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<td>8. Lodish <em>et al</em></td>
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Paper –1.3: Medical Biochemistry

Unit - I

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


Unit - II

Amino acids and peptides

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


Unit - III


Unit - IV

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

Recommended Books

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<td>Bhagwan</td>
<td>Medical Biochemistry</td>
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<td>Devlin</td>
<td>Biochemistry: with clinical correlations</td>
<td>Wiley</td>
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<td>S. N. Jogdand</td>
<td>Medical Biotechnology</td>
<td>Himalaya Publ.</td>
<td>2008</td>
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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

Unit-III

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:

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<td>2</td>
<td>Brown, T.A</td>
<td>Genomes:</td>
<td>Wiley</td>
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<td>Lewin, B.</td>
<td>Genes VII</td>
<td>Oxford</td>
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<td>6</td>
<td>Weaver, R.F.</td>
<td>Molecular Biology</td>
<td>McGraw Hill</td>
<td>2002</td>
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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**


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:

Unit – II:

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:

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 Biostatistics

Unit – I:

Unit – II:

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:

Books suggested:

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

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<td>1</td>
<td>Malcolm Campbell and Laurie J. Heyer</td>
<td>Genomics, proteomics and Bioinformatics</td>
<td>BenjaminCummings</td>
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<td>Lynn B. Jorde et al</td>
<td>Encyclopedia of Genetics, Genomics, Proteomics and Bioinformatics</td>
<td>Wiley</td>
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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

Page No. MG.10
# Suggested Readings

## Recommended Books

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<td>Short Protocols in Molecular Biology</td>
<td>Wiley</td>
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<td>Brown</td>
<td>Essential Molecular Biology vol. II</td>
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<td>Glick &amp; Pasternak</td>
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<td>Kracher</td>
<td>Molecular Biology - A Practical Approach</td>
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<td>Krenzer &amp; Massey</td>
<td>Recombinant DNA and Biotechnology</td>
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<td>Micklos &amp; Freyer</td>
<td>DNA Science</td>
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<td>Primrose</td>
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<td>Reischel</td>
<td>Molecular Diagnosis of Infectious Diseases</td>
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<td>Robertson <em>et al</em></td>
<td>Manipulation &amp; Expression of Recombinant DNA</td>
<td>AP</td>
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<td>Sambrook <em>et al</em></td>
<td>Molecular Cloning Vol. 1, 2, 3</td>
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<td>Twyman</td>
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<td>Watson <em>et al</em></td>
<td>Recombinant DNA</td>
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<td>P.K. Gupta</td>
<td>Molecular Biology and Genetic Engineering</td>
<td>Rastogi Publ.</td>
<td>2008</td>
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<td>K.Venkateswarlu</td>
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<td>V. Krishna Reddy</td>
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<td>17</td>
<td>P.K. Gupta</td>
<td>Genetics</td>
<td>Rastogi Publ.</td>
<td>2009</td>
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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
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 alleleism; 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 counselling; 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 responses-G6PD.

Suggested 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. Fuhrmann 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


Unit – III:


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 websites, 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:

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<td>1</td>
<td>Attwood and Parry Smith</td>
<td>Introduction to Bioinformatics</td>
<td>pearson</td>
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<td>Barnes and Gray (ed)</td>
<td>Bioinformatics for Geneticists</td>
<td>Wiley</td>
<td>2003</td>
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<td>Lesk</td>
<td>Introduction to Bioinformatics</td>
<td>Oxford</td>
<td>2003</td>
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<td>Mount</td>
<td>Bioinformatics: Sequence and analysis</td>
<td>CBC</td>
<td>2003</td>
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<td>Rashidi and Buchler</td>
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<td>6</td>
<td>Rastogi et al</td>
<td>Bioinformatics: Concepts, skills and applications</td>
<td>CBC</td>
<td>2003</td>
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<td>7</td>
<td>Westhead et al</td>
<td>Bioinformatics Instant Notes</td>
<td>Viva Books</td>
<td>2003</td>
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<td>8</td>
<td>Azariah et al</td>
<td>Bioethics in India</td>
<td>Eubios Ethics</td>
<td>1998</td>
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<td>9</td>
<td>Bryant et al</td>
<td>Bioethics for Scientists</td>
<td>Wiley</td>
<td>2002</td>
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Paper 3.3: Prenatal Diagnosis and Gene Therapy

Unit – I

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

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. Fuhrmann and F. Vogel
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


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<th>Recommended Books</th>
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<tr>
<td>2. Cowell</td>
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<td>3. Ehrlich</td>
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<td>4. Gersen &amp; Keagle</td>
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<td>5. Lewin</td>
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<td>7. Stillman</td>
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<td>8. Thompson and</td>
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<td>Thompson</td>
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<td>10 S.N. Jogdand</td>
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</tbody>
</table>
Paper 3.5: PRACTICAL – V

Part – A

1. Sickling Test
2. Separation of abnormal haemoglobins
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. Immuno-electrophoresis

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

Part – B

Tissue culture
Procedure
Types
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 histocompatibility 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 Cytogenetics - J. L. Hamerton
2. New Chromosomal Syndromes – J. J. Yunis
4. Molecular Biology and Genetic Engineering – P. K. Gupta (Rastogi Publ)
5. Basic Immunogenetics – Fuden Berg et al.,
7. Fundamentals of Molecular Biology – Avinash & K Upadhaya