M.Sc. HUMAN GENETICS
Course Structure - (w.e.f. 2011-2012)

Semester – I

Paper	Title of the paper	Internal	Semester End	Total	Credits
1.1	Basic Human Genetics	15	85	100	4
1.2	Population Genetics and Biostatistics	15	85	100	4
1.3	Human Cytogenetics and Reproductive Genetics	15	85	100	4
1.4	Molecular Genetics and Molecular mechanisms of Human Pathogens	15	85	100	4
1.5	Practical - I	--	100	100	2
1.6	Practical - II	--	100	100	2

Semester – II

Paper	Title of the paper	Internal	Semester End	Total	Credits
2.1	Developmental and Behavioral Genetics	15	85	100	4
2.2	Human Biochemical and Immunogenetics	15	85	100	4
2.3	Medical and Cancer Genetics	15	85	100	4
2.4	Molecular Genetics and Human Genome Project	15	85	100	4
2.5	Practical - III	--	100	100	2
2.6	Practical - IV	--	100	100	2
2.7	Fundamentals of Human Genetics (Non-Core/Optional Paper)	15	85	100	4

Semester – III

Paper	Title of the paper	Internal	Semester End	Total	Credits
3.1	Clinical Genetics and Genetic Toxicology	15	85	100	4
3.2	DNA Technology and Genetic Engineering	15	85	100	4
3.3	Genetic Screening, Counseling and Gene Therapy	15	85	100	4
3.4	Genomics and Proteomics	15	85	100	4
3.5	Practical -V	--	100	100	2
3.6	Practical -VI	--	100	100	2
3.7	Advanced Human Genetics (Non-Core/Optional Paper)	15	85	100	4

Semester – IV

4.1	Dissertation based on Project work	200	8	4.2
Comprehensive Vivo – Voce		100	4	
Grand Total I – IV Semesters		2300 -	80	

SEMESTER – I

PAPER–1.1: BASIC HUMAN GENETICS

Unit -I

Introduction to Genetics; Mendelism- Mendel and his experiments, Law of segregation Law of independent assortment; Chromosomal basis of segregation and independent assortment. Extensions of Mendelism-Allelic variation and gene function- Dominance relationships, basis of dominant and recessive mutations; Multiple allelism, allelic series.

Unit -II

Genotype to phenotype: effect of the environment on phenotype development- Penetrance and expressivity, phenocopy; lethal and sub lethal mutations; Gene interactions and modifying genes; Pleiotropy; Polygenic inheritance; Multifactorial inheritance

Unit -III

History of Human Genetics; Pedigrees- gathering family history, pedigree symbols, construction of pedigrees; Monogenic traits - Autosomal inheritance-dominant and recessive; Sex-linked inheritance- dominant and recessive; Sex-limited and sex-influenced traits; Y-linked ; Mitochondrial inheritance

Unit -IV

Linkage and crossing over – types of crossing over ; Genetic and Physical mapping; heredity and environment (twin studies).

Recommended Books				
1.	Atherly *et al*	The Science of Genetics	Saunders	1999
2.	EJ Mongia and AP Mongia	Basic Human Genetics		
3.	Fairbanks *et al*	Genetics	Wadsworth	1999
4.	Gardner *et al*	Principles of Genetics	John Wiley	1991
5.	Snustad *et al*	Principles of Genetics	Wiley and sons	1998
6.	Griffiths *et al*	An Introduction to Genetic Analysis	Freeman	2004
7.	Curt stern	Principles of Genetics		
8.	Snustad *et al*	Principles of Genetics	Wiley and sons	1998
9.	Strickberger	Genetics	Mcmillan	1985
10.	Thomson and Thomson	Genetics in Medicine		

PAPER-1.2: POPULATION GENETICS AND BIOSTATISTICS

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 for stable polymorphism. DNA markers and populations differences. Application of population genetics. Role of population genetics in genetic counseling. Genetics of 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. Rich New Delhi
4. Introduction to Biostatistics by Robert R. Sokel and James F. Kohlf

PAPER-1.3: HUMAN CYTOGENETICS AND REPRODUCTIVE GENETICS

Unit – I

History and development of Human cytogenetics–;Morphological variability of the human chromosome and karyotyping; Banded chromosomes and individual characterization of the human chromosomes; Standardization in Human Cytogenetics;

Unit – II

The origin and transmission of chromosomal abnormalities; numerical chromosomal abnormalities(five classical syndromes);structural chromosomal abnormalities (5p-); Fragile X – chromosome, heterochromatin and Lyon's hypothesis; somatic cell hybridization and use of somatic cell hybrids in gene mapping; Fluorescence in situ hybridization

Unit – III

Male and female reproductive systems; gonads and differentiation of sexual characters; hormonal regulation of sexual differentiation; Reproductive disorders- pseudohermaphroditism; true hermaphroditism; gonadal disgenesis, testicular feminization;

Unit – IV

Infertility - genetic basis of male infertility**,** genetic basis of female infertility; recurrent pregnancy loss;

SUGGESTED BOOKS

1. Human Cytogenetics (vol. I & II) – J.L. Hamerton
2. Human chromosomes : E.H. FORD
3. Human Genetics – F. Vogel and A.G. Motulsky.
4. genetics and Medicine – M.W Thompson, R.R. Meinees and H.F Willard
5. Basic human genetics – E.J. Mange and A.P. Mange.
6. Medical Genetics – Jorde et al
7. New chromosomal syndromes : J.J. Yunis
8. Comprehensive clinical endocrinology : Besser and Thorner
9. Principles and practice of Medical genetics : Rimoin etal

PAPER- 1.4: MOLECULAR GENETICS AND MOLECULAR MECHANISMS OF HUMAN PATHOGENS

Unit-I

Structure and types of nucleic acids(DNA and RNA); mitochondrial genome; transposons; pseudogenes; mechanisms of replication in prokaryotes and eukaryotes; regulation of DNA synthesis; DNA repair systems.

Unit-II

Transcription mechanisms in prokaryotes and eukaryotes; post transcriptional processing-5' capping, 3' polyadenylation, splicing; translation mechanisms in prokaryotes and eukaryotes; post translation modifications; regulation of transcription- operon and regulon ; regulation of translation.

Unit-III

Host-pathogen interaction, evolution of pathogenecity and regulation of virulence; Mechanism of drug resistance in pathogens; Molecular mechanisms for origin of new pathogens

Unit-IV

Molecular biology of pathogens: HIV, Hepatitis viruses, Mycobacterium tuberculosis, Vibrio cholerae, Plasmodium, Leishmania, Trypanosoma, Entamoeba

SUGGESTED BOOKS

1. Molecular Biology: Weaver, RF
2. Genes and Genomes: Singer,M and Berg, P
3. Molecular Cell Biology: Lodish et al.
4. Molecular Biology: Bruce Alberts et al.
5. Microbiology: Principles & Explorations : Black
6. Introductory Microbiology : Heritage et al
7. Biology of Microorganisms : Madigen et al
8. Microbiology-A Human Perspective : Nesler et al

PAPER- 1.5: PRACTICAL – I

Part – A

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

2. **Genetic Traits**
    a) Colour Blindness
    b) Phenyl Thio Carbamide (PTC)
3. **Dermatoglyphics**
    a) Finger ball patterns
    b) Palmar patterns

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- 1.6: PRACTICAL – II

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

1. Isolation of DNA from peripheral Blood
2. Isolation of DNA from Tissue
3. Isolation of RNA from Lymphocytes
4. Quantification of Genomic DNA
5. Quality check for Genomic DNA

SEMESTER – II

PAPER – 2.1: DEVELOPMENTAL AND BEHAVIORAL GENETICS

Unit - I

Genetics of embryonic development; basic concepts in development , animal models in human development. Major processes in embryonic development- pattern formation, axis specification and organogenesis.

Unit – II

Genomic imprinting; transgenic animals; later phases of embryonic development; phenocopies and malformations; the development of structure and birth defects in humans; sex differentiation and its errors

Unit – III

Scope and importance of behavioral genetics; animal models; behavioral genetic experiments in the mouse

Unit – IV

Genetics of human behaviour; chromosomes and mental retardation; chromosomes and antisocial behaviour and aggressiveness; genetics and alcoholism; genetics of psychiatric disorders- Schizophrenia; ethnic differences in IQ tests; twins in behaviour genetic research; behavioural genetics and affective disorders

SUGGESTED BOOKS

1. Human Genetics: Vogel and Motulsky
2. Basic Human Genetics : Mange and Mange
3. Genetic studies in Affective Disorders: DP Papolos and HM Lachman
4. Medical Genetics : Jorde et al

PAPER-2.2: HUMAN BIOCEMICAL AND IMMUNOGENETICS

Unit – I

The concept of Biochemical polymorphism, enzyme, protein and DNA polymorphisms – ACP, ESD, HP and GC systems; Hemoglobinopathies; Pharmacogenetics – Glucose- 6-Phosphate dehydrogenase deficiency ; Ecogenetics – Alpha -1- Antitrypsin.

Unit – II

Inborn errors of metabolism–Disorders of carbohydrate metabolism–Galactosaemia; Disorders of amino acid metabolism – Alkaptonuria, Phenylketonuria, Albinism and Homocystinuria; Disorders of Lysosomal enzymes–Tay- Sachsdisease and Mucopolysaccharidoses; Disorders of Lipoprotein and lipid metabolism – Hyper Lipoproteinemia; Disorders of Purine metabolism-Lesch Nyhan syndrome; Disorders of Pyrmidine metabolism – Orotic Aciduria

Unit – III:

The immune response – Basic concepts; the innate immune system–Phagocytes, the complement system, natural killer cells; The adaptive immune system – Cellular immune system, humoral immune system; Genetic basis of structure and diversity; Immunoglobulin molecules and The genetic basis of antibody diversity.

Unit – IV

The Major Histocompatability complex-Class I (HLA-A,B,C,E,F & G),Class II (HLA-DP,DR,DQ) and Class III (Complement genes); Immunodeficiency diseases- Agamma- globulinemia, Severe combined immuno-deficiency, Ataxia telangiectasia, Wiskott- Aldrich syndrome.
Autoimmunity – Altering immune function (vaccines and transplants); immuno-therapy (monoclonal antibodies and cytokines), immunity breakdown (AIDS)

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. Jorde et al

5. Basic immunogenetics - Fudenberg et al

PAPER- 2.3: MEDICAL AND CANCER GENETICS

Unit – I

Scope of Medical Genetics. Skin- Ichthyosis, baldness, psoriasis, hereditary hemorrhagic telenglectsia, epiloia, multiple neurofibromatosis, the porphyrias, blooms syndrome. The skeletal system – Marfan's syndrome, nail patella syndrome, brachydactyly, syndactyly, polydachyly, spina bifida and anencephaly, Ankylosing spondylitis, Rheumatoid arthritis, Osteogenesis imperfecta. Muscle – muscular dystrophies, Myotonia.

Unit – II

Eye – Glaucoma, ptosis, squint, nystagmus, cataract, retinoblastoma. Jaws – Hare lip and palate. Ears - Deafness. Alimentary system – Hypertrophic pyloric stenosis, gastic 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.

Unit – III

Cancer Genetics: What is cancer? 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 – IV

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;. Cancer prevention, diagnosis and treatment

SUGGESTED BOOKS

1. Medical Genetics – Jorde et al
2. genetics and Medicine – M.W Thompson et al
3. Clinical Genetics – A. Sorsby
4. Genetic Disorders of Man – R. M. Goodman
5. Emery's Elements of Medical Genetics – R. F. Mueller and I.D Yound
6. Human Genetics – F. Vogel and A.G. Motulsky.

PAPER- 2.4: MOLECULAR GENETICS AND HUMAN GENOME PROJECT

Unit – I

Cloning of disease genes: functional cloning-Hemophilia, positional cloning- Cysticfibrosis, Duchenmuscular dystrophy, Huntington disease; molecular pathology- gene deletions, codon deletions, duplications and insertions; mutations- point mutations, RNA splice mutations, transcriptional mutations; Mitochondrial mutations and diseases

Unit – II

Molecular diagnosis- infectious diseases; genetic diseases and direct testing-Sickle cell anemia, Beta thalassemia, Cystic fibrosis, Duchenmuscular dystrophy, Huntington disease; gene tracking- Cystic fibrosis and Huntington disease

Unit – III

The genome project- history, organization and goals of human genome project; mapping strategies, current status of various maps; human genome diversity; Organization of human genome- Mitochondrial genome, gross base composition of nuclear genome,gene density.

Unit – IV

Gene families-Multigene families, Classical gene families, families with large conserved domains, families with small conserved domains,Gene superfamilies,Gene families in clusters,origin of gene families

SUGGESTED BOOKS

1. Human Molecular Genetics: Strachan and Read
2. Principles of Genetics : Snustad and Simmons
3. Human genetics-The Molecular Revolution: Mc Kokey
4. Molecular Diagnosis: Jeffery et al.
5. The Human Genome : Hawley and Mori
6. From Genes to Genomes :Dale & Schartz
7. Genomes :Brown

PAPER- 2.5: PRACTICAL – III

Part – A

(Assignment)

1. Structure of Egg
2. Structure of Sperm
3. Cell cycle and Mitosis
4. Meiosis
5. Spermatogenesis
6. Oogenesis
7. Cleavage and Gastrulation in Mammals
8. Non-disjunction
9. Human Embryo Development
10. Transgenic Animals
11. Klinefelters syndrome
12. Downs syndrome
13. Fragile X syndrome

Part – B

1. Sickling Test
2. Separation of abnormal heamoglobins
3. Estimation of Hb
4. Estimation of Hb A2
5. Red Cell Enzymes – ACP, ESD
6. Plasma proteins – HP, CP
7. ELISA Test
8. Radial Immunodiffusion (RID)
9. Quantitative Precipitin Assay

PAPER- 2.6: PRACTICAL – IV

Part – A

(Assignment)

1. Introduction to medical genetics
2. Skin disorders- Ichthyosis, Multipile neurofibromatosis, Blooms syndrome
3. Skeletal disorders- Rheumatoid Arthritis, Osteogenesis imperfeta
4. Muscle disorders- Muscular distrophys
5. Eye disorders- Glacoma, Retinoblastoma
6. Cardiovascular disorders- Congenital heart disease, coronary heart disease, hypertension
7. Endocrinal disorders- Critinism, Goiter

Part – B

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

Paper: 2.7: FUNDAMENTALS OF HUMAN GENETICS

(*Optional Paper /Non Core Subject*)

UNIT- I:

Mendals laws of inheritance; Simple single factor inheritance (autosomal dominant, autosomal recessive, x-linked dominant, x-linked recessive and Y-linked characters); Multifactorial inheritance; Sex influenced and sex limited characters ; Polygenic inheritance.

UNIT- II:

Multiple alleles; Lethal and sub lethal genes; Penetrance and expressivity; Mutations; Linkage and Crossing over; Heredity and environment (Twin study).

UNIT- III:

Population studies and their importance; Family studies and pedigree analysis; Estimation of gene, genotype and phenotype frequencies; Randam mating and its consequences; Hardy- Weinberg law; Genetic polymorphism (balanced and transient); Inbreeding – types of consanguineous marriages.

UNIT- IV:

Scope of genetic councelling- Methods of genetic councelling
Scope of genetic screening - Prenatal and postnatal genetic screening methods.

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. Fuhramann and F. Vogel
5. Genetics of Human Populations – Cavallis S. forza and Bodmer

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 Haemopoitic 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 *et al*	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 vectorsCloning 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 *et al*	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 *et al*	Manipulation & Expression of Recombinant DNA	AP	1997
	Twyman	Advanced Molecular Biology	Viva	1999
	Watson *et al*	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 Jordee et al
5. Genetic Counseling by W. Fuhramann 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

PAPER- 3.5: PRACTICAL – V

Part – A

(Assignment)

1. Introduction to clinical genetics
2. Monogenic diseases- Cystic fibrosis, Tay-Sachs syndrome
3. Polygenic diseases- Hyperlipidemia, Diabetes mellitus
4. Inborn errors of metabolism and their genetic basis- Phenylketonuria, Galactosemia.
5. Neurogenetic disorders- Major regions of human brain and nerve conduction- Alzheimer's disease, Charcot-Marie-Tooth syndrome
6. Genetic disorders of Haemopoitic systems- Sickle cell anemia, Thalassemias,

Part – B

1. Preparation of Plasmids
2. Restriction digestion
3. Ligation of DNA fragments
4. Extraction of DNA fragments from agarose gel
5. Restriction Mapping
6. Transformation
7. Blotting Techniques:

   a) Southern
   b) Western
   c) Northern

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

Practicals based on paper-3.4: Genomics and Proteomics

1. Genome Mapviewer from NCBI
2. Gene Structure Prediction
3. Modelling of a Protein
4. Molecular Docking by Molegro Virtual Docker
5. Detecting Open Reading Frames
6. Obtaining, viewing and analyzing Structural data of proteins.
7. Identifying the protein through database by Using Gene Sequence
8. A Brief Visit to Ensemble Database
9. Microarray data analysis.
10. Secondary structure prediction of protein.

Paper 3.7: ADVANCED HUMAN GENETICS

(*Optional Paper /Non Core Subject*)

UNIT- I :

History and development of human cytogenetics; Stndardization in human cytogenetics- different conferences; Morphological variability of human chromosomes; Karyotyping.

UNIT- II :

Origin and transmission of numerical chromosomal abnormalities (classical syndromes); Structural chromosomal abnormalities (4p- and 5p-); Human leucocyte blood culture; Human chromosome banding techniques.

UNIT- III :

The concept of bio chemical polymorphism- enzyme and protein polymorphisms- ACP, ESD, HP and GC; Pharmacogenetics- G6PD; Hemoglobinopathies; In born errors of metabolism- Carbohydrate and aminoacid metabolisms.

UNIT- IV :

The immune response – Basic concept; Innate immune system (phagocytes, complement system, natural killer cells); Adaptive immune system (cellular and humoral); Genetic basis of structure and diversity; Antibody diversity ; Major histocompatability complex (class I, II and III)..

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. Principles of Human Biochemical Genetics- H. Harris
5. Basic Immunogenetics – Fuden Berg et al.,