M.Sc., HUMAN GENETICS

<u>Course Structure - (</u>w.e.f. 2009-2010) Semester – I

Paper	Title of the paper	Internal	Semester End	Total	Credits
1.1	Principles of Human Genetics	15	85	100	4
1.2	Human Anatomy and Physiology	15	85	100	4
1.3	Human Cytogenetics	15	85	100	4
1.4	Population Genetics and Biostatistic	s 15	85	100	4
1.5	Practical - I		100	100	4
1.6	Practical - II		100	100	4
	Semeste	r – II			
Paper	Title of the paper	Internal	Semester End	Total	Credits
4					
2.1	Medical and Cancer Genetics	15	85	100	4
2.2	Human Biochemical Genetics	15	85	100	4
2.3	Human Molecular Genetics	15	85	100	4
2.4	Bioinformatics	15	85	100	4
2.5	Practical - III		100	100	4
2.6	Practical - IV		100	100	4
2.7	Fundamentals of Human Genetics	15	85	100	4
	(Optional Paper/Non-core subject - f	or other De	partments)		
	Semeste	r – III			
Paper	Title of the paper	Internal	Semester End	Total	Credits
3.1	Immunogenetics	15	85	100	4
3.2	Genetic Screening and Counseling	15	85	100	4
3.3	Genetic Engineering	15	85	100	4
3.4	Genomics and Proteomics	15	85	100	4
3.5	Practical -V		100	100	4
3.6	Practical -VI		100	100	4
3.7	Advanced Human Genetics	15	85	100	4
	(Optional Paper/Non-core subject - fo	or other Dej	partments)		
	Semeste	r - IV			
4.1	Dissertation based on Project work			100	5
4.2	Comprehensive Viva – Voce			100	5
	Grand Total: I –	IV Ser	nesters	2200	-90

Semester - I

Paper – 1. 1: Principles of Human Genetics

Unit – I

Cell – cell growth – cell cycle and cell death Chromosomal basis of Heredity – distribution of chromosomes during mitosis, meiosis and Gametogenesis. Human Chromosomes – types.

Unit – II

Mendel's Laws of inheritance, Gene action, Simple single factor inheritance (autosomal dominant ; autosomal recessive, X - linked dominant, X - linked recessive and Y - linked characters) Sex influenced and sex – limited characters.

Unit – III

Multiple alleles – Polygenic inheritance Linkage and Crossing over – types of crossing over – Genetic and Physical mapping.

Unit – IV

Mutations – Lethal and sub - lethal genes aspects of phenotype expression (Penetrance and Expressivity) – Heredity and Environment (Twin studies).

SUGGESTED BOOKS:

- 1. Principles of Human Genetics Curt Stern
- 2. Human Genetics Victor A, Mckusick
- 3. Human Genetics F. Vogel and A.G. Motulsky
- 4. Genetics in Medicine M.W. Thompson, R.R Meinees and H.F. Willard.
- 5. Basic Human Genetics E.J Mande and A.P. Mange.
- 6. Human Genetics Niyogi and Srivasthava.
- 7. Genetics M. P. Arora and G. S. Sandhu ((Himalaya Publishing House)
- 8. Genetics P. K. Gupta(Rastogi Publications)

Paper - 1.2: Human Anatomy and Physiology

Unit – I

Introduction to Human Anatomy and Physiology. Types of tissues – Epithelial, Connective, Muscular and Nervous. Glands.

Bones – structure, development, function and classification

Skeletal system- Axial skeleton, Appendicular skeleton and Articulations

Unit – II

The Integument system – structure, function, derivatives of the skin. Muscular system- types of muscles, muscle physiology. Digestive system – organization of the digestive system, regional digestion, metabolism of carbohydrates, proteins, lipids.

Unit – III

Respiratory system – organs of respiratory system, physiology of respiration Urinary system – organs of urinary system, urine formation Endocrine system – hormones, mechanism of hormone action, different ductless glands – thyroid, parathyroid, adrenal and pituitary glands.

Unit – IV

Circulatory system – Heart, blood vessels, cardiac cycle, blood pressure. Body fluids – blood, lymph and tissue fluids. Nervous system – Neurons, nerve impulse Reproductive system – male and female reproductive systems, basic embryology.

SUGGESTED BOOKS:

- 1. Human Anatomy and Physiology by JE Crouch and JR Mac Clintick
- 2. Human Anatomy and Physiology by EB Steen and A. Montagua (Vol. I & II)
- 3. Human Anatomy and Physiology by DC Kimber et al
- 4. Human Anatomy and Physiology by EJ Reith et al

Paper – 1.3: Human Cytogenetics

Unit – I

History and development of Human Cytogenetics

The Human Chromosomes: Introduction – Morphological variability of the human chromosome; Banded chromosome and individual characterization of the human chromosomes; Standardization in Human Cytogenetics; General remarks;

Unit – II

The origin and transmission of chromosomal abnormalities–Introduction, numerical changes resulting from disturbances of chromosome distribution; structural chromosomal abnormalities, Fragile X – chromosome, Genomic imprinting.

Unit – III

Heterochromatin and genetic inactivation – Nature of the X - chromatin, X-inactivation as the mechanism of gene dosage compensation; Lyon's hypothesis;

Unit – IV

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. **Molecular Cyogenetics** – Fluorescence in situ hybridization.

SUGGESTED BOOKS:

- 1. Human Cytogenetics (vol. I & II) J.L. Hamerton
- 2. Human chromosomes : E.H. FORD
- 3. Human chromosomes : S. Makina
- 4. chromosome in mitosis and interphase H.G. schwarzacher
- 5. Molecular structure of human chromosomes : J.J. yunis
- 6. Human Genetics Victor A, Mckusick
- 7. Human Genetics F. Vogel and A.G. Motulsky.
- 8. genetics and Medicine M.W Thompson, R.R. Meinees and H.F Willard
- 9. Basic human genetics E.J. Mange and A.P. Mange.
- 10. Human Genetics Niyogi and Srivasthava.
- 11. Medical Genetics Jorde et al
- 12. Clinical atlas of human chromosomes : Jean Degrouchy and C. Turleau.
- 13. New chromosomal syndromes : J.J. Yunis

Paper- 1.4: 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. Genetic 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 combinationsconditional probability. Binomial distribution. Tests of significance – chi square test and 't' test.

SUGGESTED BOOKS :

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. Qunatitative Genetics Douglas S. Falconer (Pearsons Publications)

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

<u>Part – B</u>

- 1. Landmarks of Human skeleton
- 2. Identification of the sex based on the Pelvis, skull and other bones
- 3. Anthropometrical measurements on living subjects
- 4. Anthroposcopic observations

Paper 1.6: PRACTICAL – II

$\underline{Part - A}$

- 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

SEMESTER – II

Paper- 2.1 : Medical and Cancer Genetics

Unit – I

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 – II

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.

Unit – III

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 – 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, Diet – cancer associations, 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.
- 7. Molecular Genetics of Cancer by Cowell
- 8. Molecular Genetics of Cancer by Stillman
- 9. A Text Book of Molecular Biotechnology S. Ram Reddy, K. Venkateswarlu & V. K. Reddy (Himalaya Publications)

Paper -2.2: Human Biochemical Genetics

Unit – I

The concept of biochemical polymorphism – Blood groups, enzymes, protein and DNA polymorphisms – SNPs

The haemoglobin – Structure, synthesis and functions of haemoglobin; Structural variants and synthesis of globin chains.

Unit – II

Enzyme and protein polymorphism – Acid phosphatase, Glyoxalase, Esterase- D, Superoxide dismutase, Haptoglobin, Albumin and group specific component

Unit – III

Pharmacogenetics – Glucose- 6- Phosphate dehydrogenase deficiency and serum cholinesterase deficiency. Ecogenetics – Alcohol dehydrogenase and Alpha -1- Antitrypsin Pharmacogenomics

Unit – IV

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

SUGGESTED BOOKS

- 1. The principles of Human biochemical genetics H. Harris
- 2. The Biochemical Genetics of man D.J.H Brock and O. Mayo
- 3. Genetic markers in human blood E.R. Giblett
- 4. Medical Biotechnology S. N. Jogdand (Himalaya Publications)

Paper – 2.3: Human Molecular Genetics

Unit – I

DNA structure ; chromatin organization ; internal organization of gene ; nuclear genome ; mitochondrial genome organization ; unique sequences and reiterated sequences, transposons, Pseudogenes; gene families.

Unit – II

DNA replication – Meselson and stahl experiment, enzymes involved in replication, Mechanism of replication.

DNA damage and repair, Central dogma.

Transcription - Types of RNA, mechanism of transcription, processing of m RNA

Unit – III

Translation – Gene code, protein synthesis, post translational modification. Regulation of gene expression-

Transcriptional- promoters, transcription factors, inducible gene expression, Alternate promoters.

Post transcriptional- alternative splicing, alternate polyadenylation, RNA editing; Epigenetic mechanisms- DNA mathylation.

Unit – IV

The antisense oligonucleotides;molecular explanation of dominance and recessive ness; unipareatal disomy; mosaicism; X – chromosome inactivation.

SUGGESTED BOOKS

- 1. Human Molecular genetics T. Strachan and A.P Read
- 2. Genetics in Medicine M.W. Thompson et al
- 3. Principles of Genetics D.P. Snuslad & M.J Simmons
- 4. Human Genetics. The Molecular Revolution E.H Mc Konkey
- 5. Introduction to Molecular Medicine D.W. Ross.
- 6. Molecular Biology M.P. Arora (Himalaya Publications)
- 7. Human Molecular Genetics-2nd Edition-Peter Sudvery (Pearson Education)
- 8. Molbio,Fundamentals of Molecular Biology-Avinash&Kakoli Upadhyay (Himalaya Publishing House)

Paper -2.4: 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 locators

Unit – II

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

Unit – III

Acess 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. Pharmacogenomics, 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. Introduction to Bioinformatics by Attwood and Parry Smith
- 2. Bioinformatics for Geneticists by Barnes and Gray (ed)
- 3. Introduction to Bioinformatics by Lesk
- 4. Bioinformatics: Sequence and analysis by Mount
- 5. Bioinformatics Basics by Rashidi and Buchler
- 6. Bioinformatics: Concepts, skills and applications by Rastogi et al
- 7. Bioinformatics Instant Notes by Westhead et al
- 8. Bioethics in India by Azariah et al
- 9. Bioethics for Scientists by Bryant et
- 10. Bioinformatics N.J. Chikhale & V. S. Gomese (Himalaya Publishing House)

Paper 2.5: PRACTICAL – II

$\frac{Part-A}{(Assignment)}$

- 1. Case studies on single gene disorders
 - a) Austosomal Dominant
 - b) Autosomal Recessive
 - c) X-linked Dominant
 - d) X-linked Recessive
- 2. Case studies on multifactorial disorders

<u>Part – B</u>

- 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. Screening for Inborn Errors of Metabolism
- 8. Estimation of hemoglobin

Paper 2.6: PRACTICAL – IV

Part – A

- 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

<u>Part – B</u>

Bioinformatics

- 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 $X \setminus W$
- 6. Construction of Phylogenic Trees

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

SEMESTER – III

Paper- 3.1: Immunogenetics

Unit – I

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

Unit – II

Immune response proteins – Genetic basis of structure and diversity. Immunoglobulin molecules, The genetic basis of antibody diversity, T- cell Receptors

Unit – III

The Major Histocompatability complex Class I (HLA-A,B,C,E,F & G) Class II (HLA-DP,DR,DQ) and Class III (Complement genes) MHC- Disease associations

Unit – IV

Immunodeficiency diseases- Agamma- globulinemia, Severe combined immunodeficiency, Ataxia telangiectasia, Wiskott- Aldrich syndrome. Autoimmunity – Altering immune function (vaccines and transplants); immunotherapy (monoclonal antibodies and cytokines), immunity breakdown (AIDS)

SUGGESTED BOOKS:

- 1. Basic immunogenetics Fudenberg et al
- 2. Medical Genetics Jorde et al
- 3. Human genetics Ricki Lewis
- 4. The Metabolic Basis of inherited Disease Scriver et al
- 5. Genetics in Medicine J.S. Thompson and M.M. Thompson
- 6. Blood Groups in Man R.R Race and R. Sanger
- 7. Human Genetics- F Vogel and A.G. Motulsky

Paper 3.2: Genetic Screening and Counseling

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:

- a. Human Molecular Genetics by T. Strachan and AP Read
- b. Human Genetics by F. Vogel and A.G. Motulsky
- c. Genetic Engineering by Sandhya A. Mitra
- d. Medical Genetics by Jordee et al
- e. Genetic Counseling by W. Fuhramann and F. Vogel
- f. Genetics in Medicine by Thomson and Thomson.

Paper – 3.3: Genetic Engineering

Unit – I:

Isolation of nucleic acids, restriction enzymes, restriction mapping, vectors from Animal viruses, special vectors, DNA sequencing, short gun method, Polymerase Chain Reaction (PCR), primer design, modifications of PCR, automated DNA sequencing.

Unit – II:

Ligation of DNA fragments, DNA delivery methods, introducing genes in prokaryotes and eukaryotes. Identification of recombinants-direct and indirect methods. Expression of cloned genes.

Unit – III:

Olegonucleotide synthesis, isolation of mRNA, cDNA synthesis, screening of DNA and cDNA libraries. Mutagenesis- directed and insertional mutagenesis. Gene targeting, site specific recombination.

Unit – IV:

Applications of gene cloning- biological, clinical, environmental and industrial applications. Protein engineering, biosensers and biochips. Hazards of genetic engineering and impact on society.

SUGGESTED BOOKS:

- 1. Genetic engineering principles and practice by Sandhya Mitra
- 2. Molecular Biology and Biotechnology by JM Walker and R. Rapley
- 3. Gene cloning and DNA analysis by T.A. Brown
- 4. Human Molecular Genetics by T. Strachan and A.P. Read
- 5. From Genes to clones introduction to gene technology by EL Winnacker.
- 6. Molecular Biology and Genetic Engineering-P. K. Gupta (Rastogi Publications)

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 homology- comparative modeling of proteins.

Unit – IV

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

SUGGESTED BOOKS:

1. Genomics, proteomics and Bioinformatics

By Malcolm Campbell and Laurie J. Heyer

2. Encyclopedia of Genetics, Genomics, Proteomics and Bioinformatics

By Lynn B. Jorde et al

3. Proteomics - V. S. Gomase and N. J. Chikhale (Himalaya Publishing House)

Paper 3.5: PRACTICAL - V

<u>Part – A</u>

- 1. ABH Secretor status
- 2. MNSs Blood groups
- 3. ELISA Test
- 4. Radial Immunodiffusion (RID)
- 5. Quantitative Precipitin Assay
- 6. Immunoelectrophoresis

<u>Part – B</u> (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)

Paper 3.6: Practical – VI

Part – A

- 1. Preparation of Plasmids
- 2. Restriction digestion and mapping
- 3. Ligation of DNA fragments
- 4. Transformation
- 5. Bacterial cultures
- 6. Blotting Techniques:
 - a) Southern
 - b) Western
 - c) Northern

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 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.,