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*ANY ONE PAIR OF ELECTIVE PAPER A OR B OR C
Unit 1: Mendelian Genetics and Extensions
1.1 Physical basis of Heredity. Cell division – Mitosis & Meiosis
1.3 Incomplete dominance and codominance; Multiple alleles; Lethal alleles; Pleiotropy, Penetrance and Expressivity – human examples
1.4 Two gene interactions – Epistatic, nonepistatic interactions; Polygenic inheritance in man and other animal organisms.
1.5 Genes and environment – norm of reaction, phenocopies, developmental noise

Unit 2 Sex Linked Inheritance and Sex Determination
2.1 Sex Linked Inheritance - Sex linked inheritance in Drosophila and human; Sex limited and Sex influenced inheritance
2.2 Sex Determination - Sex determination in Drosophila – Genic balance theory
2.3 Sex determination in eukaryotes – heterogametic, homogametic, haplodiploidy, role of environmental factors, mosaics
2.4 Sex determination in mammals- and role of human Y chromosome

Unit 3 Extrachromosomal Inheritance
3.1 Mitochondrial inheritance (petite mutations); Mitochondrial inheritance in man
3.2 Maternal inheritance-shell coiling in snail, Ephestia pigmentation
3.3 Infective heredity - symbionts in Drosophila, Kappa particles in Paramecium.
3.4 Epigenetics and genome imprinting in humans

Unit 4: Linkage, crossing over and chromosome mapping
4.1 Linkage and chromosome mapping in eukaryotes – cytological basis of crossingover; recombination frequency, two factor and three factor crosses; interference and coincidence; Mitotic recombination
4.2 Linkage and chromosome mapping in prokaryotes – bacteria and bacteriophages – transformation, transduction, conjugation; gene mapping in bacteria.
4.3 Genetic definition of gene - Complementation test, intragenic complementation, rII locus of phage T4

Unit 5: Variation in Chromosome number and structure

5.1 Specialized chromosomes - Lampbrush chromosomes. Polytene chromosomes: Supernumerary chromosomes.

5.2 Variation in chromosome structure - Deletion, Duplication, Inversion, Translocation, Position effect

5.3 Variation in chromosome number - Euploidy and Aneuploidy in man

HGP-I (PRACTICLS)

1. Mendel’s laws through seed ratios & Drosophila mutants.
2. Statistical tests in genetic analysis - application of laws of probability (product rule, sum rule, binomial probability); chi square test and its application in the analysis of genetic data.
3. Study of linkage, recombination, chromosome mapping using test cross data.
4. Pedigree analysis for dominant and recessive autosomal and sex linked traits.
6. Incomplete dominance and gene interaction through seed ratios
8. Study of aneuploidy: Down’s, Klinefelter’s and Turner’s syndromes.
10. Smear technique to demonstrate sex chromatin in buccal epithelial cells.

Suggested Readings

Unit 1 Basic Human Genetics – Monogenic traits
1.1 History of Human Genetics.
1.2 Pedigrees – family history, symbols, construction of pedigree
1.3 Monogenic traits - autosomal inheritance, sex-linked inheritance, sex-limited and sex influenced inheritance, mitochondrial inheritance
1.4 Complications in pedigree patterns – non-penetrance, expressivity, pleiotropy, genetic heterogeneity, genomic imprinting, uniparental disomy, male lethality, X inactivation, consanguinity

Unit 2 Basic Human Genetics – Complex traits
2.1 Approaches to analysis of complex traits - Nature vs nurture, monozygotic and dizygotic twins
2.2 Polygenic inheritance of continuous traits – normal growth charts, dysmorphology
2.3 Polygenic inheritance of discontinuous traits – threshold model, liability and recurrence risk
2.4 Genetic susceptibility in multifactorial disorders – diabetes

Unit 3 Genetic Mapping of Mendelian and Complex characters
3.1 Identifying recombinants and non-recombinants in pedigrees
3.2 Genetic and physical map distances – genetic markers, mapping of genetic traits
3.3 Two point mapping – LODscore analysis, multipoint mapping, homozygosimapping
3.4 Genetic mapping of complex traits – difficulties in mapping, allele sharing methods, sib pair analysis, allelic association, linkage disequilibrium mapping

Unit 4 Human Chromosomes
4.1 History of human cytogenetics
4.2 Cell division cycle – mitotic process, meiotic process
4.3 Human karyotype – banding, nomenclature of banding
4.4 Nomenclature of aberrant karyotypes

Unit 5 Chromosome anomalies
5.1 Common syndromes due to numerical chromosome changes
5.2 Common syndromes due to structural alterations (translocations, duplications, deletions, microdeletions, fragile sites)
5.3 Common chromosome abnormalities in cancer

**HGP-II (PRACTICALS)**

1. Preparation of pedigree charts for blood group, tongue rolling, ear lobes and colorblindness
2. Genetics of codominant genes – blood groups.
4. Dermatoglyphics
5. Polygenic inheritance – finger print ridge count
6. Preparation of metaphase chromosome spread using peripheral blood sample.
7. Sterilization techniques for leukocyte culture
8. Inoculation and Culture of human leucocytes
9. Preparation of metaphase plates and their staining and analysis
12. Micrometric analysis of chromosomes.
13. Study of various abnormal karyotypes observed in humans.
14. G-band of metaphase plates and their analysis
15. Sister Chromatid exchange analysis from peripheral blood

**SUGGESTED READINGS:**

1. Human Genetics: Concept and Application by Ricki Lewis 10th Edition
2. Vogel and Motulsky’s Human Genetics: Problems and Approaches
Unit 1 DNA, RNA and Protein Structure
1.1 Building blocks and chemical bonds in DNA – structure of DNA, A-B-Z and triplex DNA,
1.2 Building blocks and chemical bonds in RNA – Structure of RNA
1.3 Building blocks and chemical bonds in peptides- primary, secondary, tertiary and quaternary structure of proteins

Unit 2 Gene expression
2.1 Central dogma of molecular biology
2.2 RNA transcription
2.3 RNA processing
2.4 Translation, post-translation processing

Unit 3 DNA replication, recombination, Mutagenesis and DNA repair
3.1 DNA replication – semiconservative, semi-discontinuous, DNA replication machinery
3.2 DNA recombination
3.3 DNA mutagenesis
3.4 DNA repair

Unit 4 Human Chromosome Organization
4.1 Packaging of DNA – multiple hierarchies of DNA folding
4.2 Chromosomes as functional organelles – origins of replication, telomeres, centromeres
4.3 Heterochromatin and euchromatin

Unit 5 Human Genome organization
5.1 Mitochondrial genome – replication, genes, genetic code
5.2 Nuclear genome – protein coding genes, RNA genes
5.3 Nuclear genome – highly repetitive DNA, heterochromatin and transposon repeats
HGP-III(PRACTICALS)

1. Extraction of DNA from human lymphocytes
2. Paper chromatography of amino acids
3. Electrophoresis: agarose gel electrophoresis, PAGE
4. Study of isozymes by PAGE
5. Comet assay to measure DNA damage
6. Problem based on homologous and site-specific recombination
7. Effects of mutagens on wt and repair deficient E.coli strains.
8. Preparation of Human chromosome spread and banding

Suggested Readings:

1. Human Molecular Genetics by T. Strachan
2. Human Molecular Genetics by Gerard Meurant
3. Human Molecular Genetics by Christopher G Mathew.
4. Human Molecular Genetics by Sudbery
ANDHRA PRADESH STATE COUNCIL OF HIGHER EDUCATION  
CBCS PATTERN FOR HUMAN GENETICS  

HUMAN GENETICS  
BSc SECOND YEAR - SEMESTER-IV  
HGT-IV: RECOMBINANT DNA AND STEM CELL TECHNOLOGY  
(Revised Syllabus-2020)  
THEORY  

Unit 1 Cell Based Cloning  
1.1 Restriction endonucleases and other enzymes used in manipulating DNA molecules  
1.2 Cloning vectors – plasmid vectors, lambda and cosmid vectors, P1 phage vectors, YAC, BAC, M13 or phagemid vectors, expression vectors  
1.3 Introducing recombinant DNA into recipient cells  
1.4 DNA libraries -generation of genomic and cDNA libraries; chromosomal DNA libraries  

Unit 2 Cloning Human disease genes  
2.1 Cloning human disease genes- functional candidate gene cloning, positional candidate gene cloning  
2.2 Detection of mutations in human genes –SSCP analysis, DGGE, chemical mismatch cleavage  
2.3 Detection of mutation in human gene – DNA sequencing, heteroduplex analysis, protein truncation  

Unit 3 Applications of rDNA technology  
3.1 DNA fingerprinting – use of mini-satellites for DNA fingerprinting, single locus probes, STRs  
3.2 Genetic testing – prenatal testing, neonatal screening, diagnosis of genetic disease in children after birth, pre-symptomatic testing.  
3.3 In vivo, in vitro gene therapy; vehicles for gene therapy; gene therapy for heritable and non-heritable genetic diseases.  

Unit 4 Biology of stem cells  
4.1 Historical perspectives, concept of stem cells  
4.2 Cellular and molecular features of stem cells  
4.3 Embryonic stem cells and germ stem cells  
4.4 Fetal adult stem cells and cancer stem cells  

Unit 5 Applications  
5.1 Medical need for stem cells and preservation of stem cells  
5.2 Genetically engineered stem cells for gene therapy
5.3 Stem cell therapy – neurodegenerative disorders, cardiovascular disorders, metabolic disorders, hematopoietic disorders, organ disorders, autoimmune disorders, reproductive failures

**HGP-IV (PRACTICALS)**

1. Isolation of plasmid DNA from *E. coli* cells.
2. Digestion of plasmid DNA with restriction enzymes.
3. Estimation of size of a DNA fragment after electrophoresis using DNA markers.
4. Construction of restriction digestion maps from data provided.
5. Recovery of DNA from low-melting temperature agarose gel.
6. Preparation of competent cells of *E. coli*.
7. Transformation of competent *E. coli* cells with plasmid DNA.
8. Amplification of a DNA fragment by PCR.
10. Southern blotting.
11. Western blotting.
12. Culturing cells – aseptic techniques, media.
13. Subculturing and cell lines.

**Suggested Readings**

4. Human Molecular Genetics by Sudbery.
Unit 1 Descriptive Statistics
1.1 Methods of presentation and interpretation of data – frequency distribution, graphical representation of data, histogram, frequency polygon, frequency curve.
1.2 Measures of Central tendency – mean, median, mode
1.3 Measures of Dispersion - standard deviation, variance, coefficient of variation.

Unit 2 Elementary Probability
2.1 Mathematical definition of probability of an event, Use of permutations and combinations in calculations of Probability
2.2 Conditional probability, Additive and Multiplication law of Probability, Random Variables, Mathematical expectation and variances
2.3 Probability Distributions: Binomial, Poisson and normal distributions.
2.4 Bayes theorem

Unit 3 Correlation analysis, test of significance and ANOVA
3.1 Correlation and regression analysis—Relationship between variables
3.2 Test of significance – statistical and scientific hypothesis, null and alternative hypothesis, procedure of hypothesis testing,
3.3 Test of significance – student’s t test, chi-square test, F test
3.4 ANOVA – general idea of one way and two way analysis

Unit 4 Computers, operating systems and Internet
4.1 Principles of computer operations – basic computer architecture, hardware architecture
4.2 Principles of computer operations – software architecture, operating systems, Programming languages – traditional and scripting languages, Java, markup languages, application programs
4.3 Communication and Networks – network architecture, standards for exchange of information, internet services - email, WWW search engines
Unit 5 Bioinformatics

5.1 History of Bioinformatics
5.2 Databases and search tools – NCBI, EBI, GenomeNet; Databasemining tools – BLAST
5.3 Database archives – nucleic acid sequence databases, genome databases and genome browsers, protein sequence databases, databases of protein families, databases of structures, expression and proteomic databases, bibliographic databases
5.4 Gateways to archives – ENTREZ, PIR, ExPAsy

HGP-V(PRACTICALS)

1. Frequency distribution
2. Various types of graphs
3. Mean, Median, Mode
4. Standard deviation, variance and coefficient of variation
5. Testing of hypotheses regarding population mean
6. Testing of hypotheses about the difference between population means
7. Chi-square test
8. Testing of Correlation Coefficient
9. Fitting of simple linear regression
10. One-way ANOVA&Two-way ANOVA
11. Internet basics
12. Sequence retrieval (protein and gene) from NCBI, Structure download (protein and DNA) from PDB
13. Molecular file formats - FASTA, GenBank, Genpept, GCG, CLUSTAL, SwissProt,FIR

Suggested Readings

THERE WILL BE THREE PAIRS OF EACH DOMAIN OF CORE COURSE.
STUDENT HAS TO CHOOSE ONE PAIR FROM EACH DOMAIN.

ELECTIVE THEORY
A - PAIR
HGT A1: CLINICAL GENETICS & GENETIC COUNCILING

UNIT-1 GENETIC DISORDERS I
- 1.1 Monogenic diseases – Cystic fibrosis, Tay-Sachs syndrome, Marfan syndrome
- 1.2 Inborn errors of metabolism – Phenylketonuria, Maple syrup urine syndrome, galactosemia
- 1.3 Genome imprinting syndromes – Prader Willi and Angelman syndrome

UNIT-2 GENETIC DISORDERS II
- 2.1 Genomic syndromes – Neurofibromatosis I
- 2.2 Neurogenetic disorders – Charcot Marie Tooth syndrome, spinal muscular atrophy, alzhemiers diseases, syndromes due to triplet nucleotide expansion
- 2.3 Muscle genetic disorders – dystrophies, myotonia, myopathies

UNIT-3 GENETIC DISORDERS III
- 3.1 Genetic Disorders of Haemopoiteic systems- sickle cell anaemia, thalassemias, hemophilia
- 3.2 Genetic disorders of eye – colorblindness, retinitis pigmentosa, glaucoma
- 3.3. Complex polygenic syndromes – atherosclerosis, diabetes mellitus
- 3.4 Mitochondrial syndromes

UNIT-4 GENETIC COUNCELLING
- 4.1 Role of genetic counseling
- 4.2 Causes and factors for seeking counselling
- 4.3 Dysmorphology
- 4.4 Prenatal and preimplantation diagnosis

UNIT-5 PRACTICAL GENETIC COUNCELING
5.1 Process of genetic counselling - Constructing a family tree, diagnostic information, risks and odds, estimation of risks
5.2 Genetic counselling in Mendelian disorders
5.3 Genetic counselling in Non-Mendelian disorders
5.4 Ethical and legal issues in genetic counselling

HGP A1: CLINICAL GENETICS & GENETIC COUNSELING

1. Metaphase chromosome preparations from bone marrow of mouse, rat, human
2. Chromosome preparation from lymphocyte culture
3. G-banding, C-banding, R-banding
4. Karyotyping
5. Meiosis in mouse testis
6. Sex chromatin (buccal mucosa, hair bud)
7. Micronuclei assay
8. Chromosome preparation from chorionic villi, stem cells, cell line
9. Sister Chromatid Exchange (SCE)
10. Molecular markers for tumor detection
11. Genetic counselling (pedigree analysis in disease conditions, risk calculation)
12. Y-chromosome microdeletion
13. Biochemical tests for sugar, albumin, Creatine phosphokinase-CPK, glucose 6 phosphate dehydrogenase-G6PD

SUGGESTED READINGS

2. Thompson and Thompson & Thompson Genetics in Medicine, Robert L. Nussbaum, Roderick R. McInnes, Huntington F. Willard (eds)
UNIT-1 BASICS OF CELL CULTURE & INSTRUMENTATION
1.1 Techniques of cell cultures (short term lymphocyte, primary and secondary cell cultures, maintenance of cell lines)
1.2 Spectrophotometer, centrifugation
1.3 ELISA, radioactivity detection
1.4 Mass spectrometry
1.5 High performance liquid chromatography

UNIT-2 TECHNIQUES IN CHROMOSOME ANALYSIS
2.1 Techniques of chromosome analysis - (a) Chromosome preparation from cultured lymphocytes, cell lines and solid tumors (b) Karyotyping, C-, G-banding and fluorescence banding, nomenclatures of bandings (c) In-situ hybridization techniques (d) Meiotic chromosomes in mouse testis
2.2 Chromosomal anomalies and disorders - Numerical (polyploidy, aneuploidy, autosomal, sex- chromosomal) - Structural (deletion, duplication, translocation, inversion, isochromosome, ring chromosome) - Chromosomal abnormalities in cancer
2.3 Microscopy - Metaphase chromosome preparations from bone marrow of mouse, rat, human, Sex chromatin (buccal mucosa, hair bud), Comet assay, Micronuclei assay, Chromosome preparation from chorionic villi, Sister Chromatid Exchange (SCE)

UNIT-3 GENETIC DISORDERS & MOLECULAR TECHNIQUES FOR DISEASE IDENTIFICATION
3.1 Genetic Disorders - Classification of genetic disorders, Single gene Disorders (Cystic Fibrosis, Marfan’s syndrome), Multifactorial disorders (Diabetes, Atherosclerosis, Schizophrenia)
3.2 Molecular Techniques PCR-RFLP, Multiplex-PCR, SSCP, MALDI-TOF
3.3 Disease identification and Genetic tests for following disorders: (a) Thalassemia, Fanconi, Sickle Cell anaemia, Fragile-X syndrome, Alzheimer’s disease (b) Duchenne Muscular Dystrophy/Becker’s Muscular Dystrophy, Huntington’s disease (c) Allelic susceptibility test for multifactorial disorders (Neural Tube Defect, Cleft Lip and Palate, Cardio Vascular Disorder, Male infertility)
UNIT-4 BIOCHEMICAL GENETIC DIAGNOSIS

4.1 Biochemical tests: sugar, albumin, urea, protein, globulin, vitamin;
4.2 Biochemistry and diagnostic tests of following diseases - Duchenne Muscular Dystrophy (DMD) (Creatine phosphokinase-CPK), Phenylketonuria-PKU (phenylketone), G6PD deficiency syndrome (G6PD), Endocrine disorders related to thyroid and reproduction (TSH, T3, T4, Estradiol, Testosterone, LH, FSH)

UNIT-5 GENETIC COUNCELLING AND PRENATAL DIAGNOSIS

5.1 Causes and factors for seeking counselling
5.2 Dysmorphology
5.3 Ethical and legal issues in genetic counselling
5.4 Prenatal and preimplantation diagnosis

HGP A 2 : LABORATORY DIAGNOSIS IN GENETICS

1. Metaphase chromosome preparations from bone marrow of mouse, rat, human
2. Chromosome preparation from lymphocyte culture
3. G-banding, C-banding, R-banding
4. Karyotyping
5. Fluorescence in-situ Hybridization (FISH)
6. Meiosis in mouse testis
7. Sex chromatin (buccal mucosa, hair bud)
8. Comet assay
9. Micronuclei assay
10. Chromosome preparation from chorionic villi, stem cells, cell line
11. Sister Chromatid Exchange (SCE)
12. Molecular markers for tumor detection
13. Bcr-abl (RT-PCR)
14. Genetic counseling (pedigree analysis in disease conditions, risk calculation)
15. Prenatal diagnosis of Thalassemia
16. Y-chromosome micro deletion
17. Biochemical tests for sugar, albumin, Creatine phosphokinase-CPK, glucose 6 phosphate dehydrogenase-G6PD
Suggested Reading

3. Fundamentals of Molecular Diagnostics by David E. Bruns, Edward R. Ashwood, Carl A Burti
4. Human Genetics: From Molecules to Medicine by Christian Patrick Schaaf, Johannes Zschocke, Lorraine Potocki

ANDHRA UNIVERSITY
HUMAN GENETICS
BSc THIRD YEAR – SEMESTER-V
B-PAIR
HGT- B 1 : HUMAN GENOME PROJECT AND GENOMES

Unit 1 Genome Organization and Study

1.1 Genome – general features, features of eukaryotic nuclear genomes
1.2 Genomes, transcriptomes and proteomes

Unit 2 Mapping Genomes

2.1 Genetic mapping – pedigree analysis, DNA markers – RFLPs, SSLPs, SNPs
2.2 Physical mapping – restriction mapping, FISH, radiation hybrid mapping, STS mapping
2.3 Sequencing genome- assembly of contiguous DNA sequence, shotgun method, clonecontig method, whole-genome shotgun sequencing

Unit 3 Genome Projects

3.1 Human genome project, HapMap Project, 1000 genome project, ENCODE project
3.2 Other genome projects.
3.3 Applications and proposed benefits of HGP – ELSI.
Unit 4 Understanding Genome sequence
   4.1 Locating the genes in a genome sequence
   4.2 Determining the functions of individual genes
   4.3 Transcriptome – microarrays
   4.4 Proteome – protein profiling

Unit 5 Molecular phylogenetics
   5.1 Phenetics and cladistics
   5.2 Reconstruction of DNA based phylogenetic tree
   5.3 Applications of molecular phylogenetics – evolutionary relationship between humans and primates; origin of AIDS; human pre history.

HGP- B 1 : HUMAN GENOME PROJECT AND GENOMES

1. Isolation and purification of genomic DNA.
2. Detection of SNPs using SNP specific primers and PCR.
3. Study of VNTR’s in human genome as the polymorphic loci.
4. Design primers for PCR based detection of the gene and mapping primers on the genome
5. Introduction to NCBI websites
7. Web based analysis to retrieve a nucleotide sequence from NCBI.
8. Sequence alignment using BLASTn, BLASTp, CLUSTALW.
9. Gene finding tools – GenScan, GLIMMER
10. Introduction to proteomics – Protparam, GOR, unPredict, SWISSMODEL.
11. Visualization software – Rasmol
12. Generating phylogenetic tree using PHYLIP

Suggested Readings
   1. Human Genome Project by James Torrillo.
   2. Understanding the Human Genome Project by Michael A Palladino.
   5. Genomes 3 by Terence A Brown.
UNIT-1 NUCLEIC ACID ISOLATION AND AGAROSE GEL ELECTROPHORESIS

Conventional and kit method for isolation of nucleic acids-Plasmid DNA-Genomic DNA from Bacterial cells, Plant cells, animal cells-RNA isolation and m-RNA purification –Agarose purification-Agarose gel electrophoresis-Staining techniques –Pulse field gel electrophoresis

UNIT-2 PCR TECHNIQUES

Principle of Polymerase Chain Reaction (PCR)-Components of PCR reaction and optimization of PCR –Gene specific primer- Inverse PCR, Hot-start PCR ,Loop mediated PCR – Reverse transcription PCR and Real time PCR.Chemistry of primer synthesis

UNIT-3 HYBRIDIZATION METHODS

Probes –Labelling of probes-Radio active and non-radio active probes-Detection techniques, Southern hybridization, Northern hybridization, Western blotting

UNIT-4 DNA SEQUENCING AND GENE SYNTHESIS

Sangers’s method of DNA sequencing – Manual and automated methods. Pyrosequencing-massive parallel 454-sequencing, illumina sequencing, SOLID sequencing, single molecule sequencing

UNIT-5 PROTEIN TECHNIQUES

Electrophoresis of protein –native and denaturing conditions, capillary and gel electrophoresis, 3D gel electrophoresis, ELISA, yeast hybrid system-one hybrid system, phage display
HGP-B 2: MOLECULAR TECHNIQUES IN GENETIC ENGINEERING

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
9. SDS-Gel electrophoresis
10. Southern blotting
11. Northern blotting
12. Western blotting

REFERENCES


UNIT-1: Germ Cells and Fertilization

Germ Cells
Spermatogenesis
Oogenesis
Fertilization and Gastrulation

UNIT-2: Molecular Aspects of Development

Maternal effect gene
Gap gene
Pair rule gene
Segment polarity genes
Homeotic genes

UNIT-3: Genetics of Embryonic Development in Drosiphila

Overview of Drosophila development
Zygotic genes and segment formation

UNIT-4: Flower Development in Arabidopsis

Development, Role of Homeotic Selector Gene

UNIT-5: Genetic Control of Behaviour

Introduction, Behaviour in Invertebrates, Honeybee, Drosophila – Genetic basis of alcoholism, genetic basis for sexual orientation, Courtship behaviour in various animals.
HGP C 1: DEVELOPMENTAL AND BEHAVIORAL GENETICS

1. Study of development in chick embryo
2. Dissection of imaginal disc in Drosophila larvae
3. Life cycle of drosophila, husbandary and handling.
4. Role of SHH signaling in chick development
5. Observation of living and plastic embedded chick embryos
6. The maternal effect gene in drosophila

REFERENCES

The cell – Bruce Alberts
Principles of Development - Wolpert
Principles of Genetics – Snustad, Simmons, Jenkins.
Unit 1 Human diseases I
1.1 Etiology, pathology and symptoms of genetically inherited diseases – PKU, alkaptonuria, galactosemia, Von Gierke disease, LeschNyhan syndrome, Gout, sickle cell anaemia, beta thalassemia, diabetes
1.2 Mode of infection, symptoms and epidemiology of disease causes by viruses (HIV, Hepatitis B, Rabies, HSV-1)
1.3 Mode of infection, symptoms and epidemiology of disease caused by bacteria – typhoid, syphilis, TB

Unit 2 Human diseases II
2.1 Mode of infection, symptoms and epidemiology of disease caused by fungi – aspergillosis, histoplasmosis.
2.2 Mode of infection, symptoms and epidemiology caused by protozoa – malaria, amoebiasis.
2.3 Cancer genetics - tumor suppressor genes, oncogenes, Molecular basis of oncogenesis

Unit 3 Basic Instrumentation principles and techniques
3.1 Principles of electrophoresis and immunoblotting
3.2 Principles of DNA sequencing and methods of genotyping and mutation analysis
3.3 Principles and applications of PCR
3.4 In situ hybridization techniques – ISH, FISH

Unit 4 Genetic testing for hereditary disorders
4.1 Genetic testing for thalassemia
4.2 Genetic testing for familial colorectal cancer
4.3 Genetic testing for familial breast and ovarian cancer
4.4 EGFR mutation in lung cancer, HER2 amplification in breast cancer, FISH test for early bladder cancer detection, KRAS mutation detection for colorectal cancer

Unit 5 Molecular diagnosis of infectious diseases
5.1 Principles of HPV testing and methods of genotyping
5.2 Hepatitis B virus infection – testing for viral load and HBV DNA mutants detection
5.3 Molecular techniques - NestedPCR, Real Time PCR for different clinical applications

**HGP VIII-C 2: Molecular Pathology in Human Diseases**

1. Preventing contamination.
2. Extract and assess the purity of DNA.
3. Agarose gel electrophoresis
4. Set up PCR.
5. Evaluate Southern blot data
6. Analyze PCR product using agarose gel electrophoresis and interpret results
7. Demonstration of karyotyping
8. Isolate cellular RNA, purify mRNA
9. Set up RT-PCR using commercial kit
10. Analyze RT-PCR results by agarose gel.

**SUGGESTED READING**

5. DNA from A to Z & Back Again; Carol A. Holland and Daniel H. Farkas; AACC Press 2008