





## HUMAN GENETICS

Course Structure (w.e.f. 2023-2024)

(Modifications in BoS meeting held on 28-08-2023)

<b>I Semester:</b>					
Paper	Title of the paper	Internal	Semester end	Total marks	Credits
1.1	Basic Human Genetics	30	70	100	4
1.2	Population Genetics and Biostatistics	30	70	100	4
1.3	Human Cytogenetics	30	70	100	4
1.4	Molecular Genetics	30	70	100	4
1.5	Practical - I	30	70	100	4
1.6	Practical - II	30	70	100	4
<b>Total marks and credits for I semester</b>				<b>600</b>	<b>24</b>
<b>II Semester:</b>					
Paper	Title of the paper	Internal	Semester end	Total marks	Credits
2.1	Cell Biology and System Physiology	30	70	100	4
2.2	Human Biochemical and Immunogenetics	30	70	100	4
2.3	Medical Genetics	30	70	100	4
2.4	Diagnostic Molecular Pathology	30	70	100	4
2.5	Practical - III	30	70	100	4
2.6	Practical - IV	30	70	100	4
<b>Total marks and credits for II semester</b>				<b>600</b>	<b>24</b>
<b>III Semester:</b>					
Paper	Title of the paper	Internal	Semester end	Total marks	Credits
3.1	Genetic Toxicology and Cancer Genetics	30	70	100	4
3.2	Genetic Engineering	30	70	100	4
3.3	Research Methodology	30	70	100	4
3.4	Data Analysis for Biologists	30	70	100	4
3.5	Practical - V	30	70	100	4
3.6	Practical - VI	30	70	100	4
3.7	MOCCs course - I	--	--	50	2
3.8	Intellectual Property Rights	--	--	--	-
<b>Total marks and credits for III semester</b>				<b>650</b>	<b>26</b>
<b>IV Semester:</b>					
Paper	Title of the paper	Internal	Semester end	Total marks	Credits
4.1	Genetic Screening, Counseling and Gene Therapy	30	70	100	4
4.2	Genomics, Proteomics and Bioinformatics	30	70	100	4
4.3	Practical -VII	30	70	100	4
4.4	Dissertation based on Project work	--	--	200	8
4.5	Comprehensive Viva-Voce			100	4
4.6	MOCCs course - II			50	2
4.7	Value added course			--	-
<b>Total marks and credits for IV semester</b>				<b>650</b>	<b>26</b>
<b>Grand total marks and credits for all four semesters (I, II, III and IV)</b>				<b>2500</b>	<b>100</b>

## **POs**

PO1. Students can pursue careers in research, biotechnology, pharmaceuticals, clinical diagnostics, and academia. They can contribute to cutting-edge research in multiple disciplines and can tackle complex questions that require expertise from different areas, such as genomics, bioinformatics, and epidemiology

PO2. Expertise of the students enable them to play vital role in health care and public health initiatives. They will use their knowledge to educate individuals and communities about disease prevention strategies, including lifestyle modifications, vaccinations, and early detection methods.

PO3. By understanding cellular processes, genetic factors and biomarkers associated with various conditions, students will attain skills to accurately diagnose diseases.

PO4. Knowledge and skills acquired through the program lays the foundation for the students to advance our understanding of disease mechanisms and make substantial contributions to the treatment and management of diseases by identifying drug targets and designing therapeutic interventions.

PO5. Graduates can identify genetic markers associated with disease susceptibility and treatment response, leading to personalized healthcare strategies.

PO6. Students with their technical, analytical and interpersonal skills can become good genetic counsellors. They can apply their expertise to provide clients with accurate genetic information, emotional support, and guidance, enabling them to make informed decisions about their genetic health and reproductive choices.

PO7. Students are capable of presenting ideas, arguments, or findings effectively with the improved written and oral communication skills and can work effectively in a team and collaborate with others on course-related projects or assignments.

PO8. Students can develop ethical policies and regulations to protect individuals' genetic information and prevent misuse and design clinical trials that minimize risks and maximize benefits while upholding ethical principles.

## **PSOs**

PSO1: Graduates will possess a comprehensive understanding of Human Genetics, including principles of inheritance, genetic variation, and the molecular mechanisms underlying genetic disorders.

PSO2: Develop a keen awareness of ethical considerations in human genetics research, clinical practice, and genetic counseling, with an understanding of the social implications of genetic advancements.

PSO3: Apply advanced statistical methods to analyze genetic data and interpret population-level genetic variations, considering factors influencing genetic diversity and evolution.

PSO4: Acquire knowledge on advanced laboratory techniques and skills relevant to Human Genetics research, including Cytogenetics, Molecular Genetics, and Bioinformatics.

PSO5: Graduates will be competent in performing and interpreting molecular diagnostic tests, and will have the knowledge and skills to provide genetic counseling.

PSO6: Able to conduct independent and collaborative research in Human Genetics, including the design, implementation, and interpretation of experiments, and contribute to the advancement of genetic knowledge.

**SEMESTER I**  
**PAPER 1.1- BASIC HUMAN GENETICS**

**Course Outcomes:**

CO1: Understand the basic concepts of Human Genetics which helps in figuring out the genetic basis of disease which aids in the design and development of effective treatment strategies.

CO2: Have a comprehensive understanding of Genetics, from Mendel's foundational discoveries to the more complex aspects of genetic inheritance and its applications in various fields of science and medicine.

CO3: Understand linkage analysis which enables the identification and mapping of genes responsible for Mendelian disorders which is vital for diagnostic and counseling purposes.

CO4: Able to apply twin studies data to inform public health policies and interventions, especially in cases where genetic factors play a significant role in disease risk.

CO5: Able to explore ethical and social issues related to Human Genetics.

**Learning outcomes:**

LO1: Acquire knowledge on the importance of classical Mendelian Genetics and extension of Mendelism.

LO2: Obtain an insight into gene interactions and different types of inheritance like polygenic and multifactorial inheritance.

LO3: Able to construct and interpret pedigrees to trace the inheritance patterns of genetic traits in families.

LO4: Capable of performing linkage analysis by which the student can contribute to genetic counseling.

LO5: Understand how genetic factors interact with environmental factors to influence traits or susceptibility to diseases.

**Course Specific Outcomes:**

CSO1. Capable of predicting disease risk in family members of an affected individual by understanding inheritance patterns of the genetic variants distributed in families.

CSO2 Able to detect the chromosomal location of disease genes by using linkage analysis tool.

CSO3. Competent enough to map genes which is vital for diagnostic and therapeutic purposes, as well as for understanding inheritance patterns.

CSO4. Able to explore the interaction of genetic factors with environmental factors in various traits and diseases with the help of Twin studies

### **CO-PO Mapping**

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓	✓	✓	✓	✓	✓	✓
CO2	✓	✓		✓	✓	✓		✓
CO3		✓	✓	✓	✓	✓		✓
CO4		✓		✓	✓			✓
CO5	✓			✓	✓	✓	✓	✓

#### **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- Linkage mapping, Linkage analysis; Crossing over – Types of crossing over, Mechanism of crossing over, Factors affecting crossing over; Genetic mapping; Physical mapping.

#### **Unit -V**

Heredity and environment (twin studies), Eugenics-history, Modern Eugenics, Types of Eugenics; Euthenics and Euphenics.

#### **Books suggested:**

1. Principles of Human Genetics - Curt Stern
2. Human Genetics - McKusick V. A
3. Basic Human Genetics –Mange E.J and Mange A.P
4. Principles of Genetics - Gardner et al.
5. Principles of Genetics - Snustad et al.
6. Genetics in Medicine-Thompson and Thompson
7. Genetics, Molecular Biology, Evolution and Ecology-Verma P.S and Aggarwal V.K

## **SEMESTER I**

### **PAPER-1.2: POPULATION GENETICS AND BIOSTATISTICS**

#### **Course Outcomes:**

CO1: Able to differentiate between random and non-random mating patterns and understand the principles of the Hardy-Weinberg equilibrium.

CO2: Able to synthesize knowledge of Population Genetics and apply it in the study of genetic demography which helps to understand the prevalence and distribution of genetic diseases within different populations.

CO3: Able to classify data and explain the process of data tabulation and frequency distribution.

CO4: Master the analytical skills to interpret and understand the central tendencies of data sets, such as disease incidence rates, mortality rates, or risk factors.

CO5: Proficient in using the Hardy-Weinberg equilibrium to investigate evolutionary processes such as genetic drift, gene flow, mutation, and natural selection.

#### **Learning outcomes:**

LO1: Able to recall the key concepts related to population genetics, including gene frequencies and mating patterns.

LO2: Can explain the factors that can disrupt the Hardy-Weinberg equilibrium and their consequences.

LO3: Able to judge the significance of the index of opportunity for natural selection in the context of genetic variation.

LO4: Calculate and interpret measures of dispersion for datasets which are essential for understanding and managing data variability, making informed decisions, and conducting meaningful statistical analyses.

LO5 (Analyzing): Able to examine the principles of probability and apply them in solving probabilistic problems.

#### **Course Specific Outcomes:**

CSO1: Analyze real-world scenarios to determine whether populations are in Hardy-Weinberg equilibrium or not.

CSO2: Calculate and interpret the index of opportunity for natural selection in different populations.

CSO3: Calculate and interpret measures of dispersion to assess data variability and spread.

CSO4: Conduct and interpret chi-square tests and 't' tests to assess the significance of experimental data in biology and genetics.

### CO-PO Mapping

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓	✓	✓	✓	✓		✓
CO2	✓	✓	✓	✓	✓	✓	✓	
CO3	✓	✓	✓	✓	✓	✓	✓	
CO4	✓	✓		✓		✓		
CO5	✓	✓	✓	✓	✓	✓		

#### **Unit – I**

Mendelian Population and scope of population genetics; Gene and genotype frequencies; Mating patterns- Random and Non-random mating; Hardy-Weinberg principle, Extension of H-W principle to multiple alleles, Sex-linked alleles, Factors that change Hardy Weinberg Equilibrium; Inbreeding and Inbreeding coefficient.

#### **Unit – II**

Genetic polymorphism, Types of Genetic polymorphisms, Examples for Genetic Polymorphism; Application of population genetics and role of Population Genetics in Human Genetics; Genetics of origin and Evolution of Human Races; Genetic Demography- Birth rate, Death rate, Reproductive rate, Index of opportunity for Natural Selection.

#### **Unit – III**

Bio-Statistics- Definition, Basic concepts of Biostatistics; Collection of data; Sampling techniques; Classification of data and Tabulation; Frequency distribution; Graphic presentation of data and Diagrammatic presentation of data.

#### **Unit-IV**

Measures of central tendency- Mean, Median and Mode. Measures of Dispersion - Range, Mean Deviation, Standard Deviation and Variance

#### **Unit – V**

Probability – laws of probability for independent events – permutations and combinations– conditional probability. Binomial distribution. Correlation and regression, Tests of significance –chi square test and ‘t’ test.

#### **Books Suggested:**

1. The Genetics of Human Populations by LL Cavalli-Sforza and WF Bodmer
2. Population Genetics Theory by James F. Crow and W. Kimura Harper and Row,
3. Introduction to Biostatistics by P.S.S. Sundara Rao and J. Rich,
4. Introduction to Biostatistics by Robert R. Sokel and James F. Kohlf



## **SEMESTER I**

### **PAPER 1.3 - HUMAN CYTOGENETICS**

#### **Course Outcomes:**

CO1: Able to know how Human Cytogenetics became closely linked to human health and recognizes its clinical applications in medicine and genetic counseling.

CO2: Develop skills needed to diagnose, counsel, and contribute to research and therapy development in the field of genetics.

CO3: Provide explanations regarding the genetic factors behind male and female infertility and pregnancy loss.

CO4: Able to analyze the structure and number of chromosomes in cells to diagnose genetic disorders and syndromes.

CO5: Able to assess the causes and consequences of both numerical and structural chromosomal abnormalities.

#### **Learning Outcomes:**

LO1: Obtain knowledge on the mechanisms of non-disjunction and their role in numerical chromosomal abnormalities.

LO2: Able to understand case studies and explanations for numerical chromosomal abnormalities like Down syndrome and Edward syndrome.

LO3: Examine the genetic basis and clinical manifestations of structural chromosomal abnormalities.

LO4: Assess the significance of chromosomal instability syndromes and the role of genetic imprinting.

LO5: Acquire deep insight into the genetic factors contributing to male and female infertility and chromosomal abnormalities in recurrent pregnancy loss.

#### **Course Specific Outcomes:**

CSO1: Demonstrate proficiency in karyotyping and characterizing individual human chromosomes.

CSO2: Evaluate the advantages and limitations of the old and new classification systems in Human Cytogenetics.

CSO3: Develop skills in diagnosing and explaining numerical chromosomal abnormalities in real-life scenarios.

CSO4: Investigate and interpret the genetic and clinical features of conditions such as Cri-du-chat syndrome and Fragile X syndrome.

### **CO-PO Mapping**

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓	✓	✓	✓	✓		✓
CO2	✓	✓	✓	✓	✓	✓		✓
CO3	✓	✓	✓	✓	✓	✓	✓	✓
CO4	✓	✓	✓	✓		✓	✓	✓
CO5	✓	✓	✓	✓	✓	✓	✓	✓

#### **Unit – I**

History and growth of Human Cytogenetics; Morphological variability of the human chromosomes; Karyotyping; Banded chromosomes and individual characterization of the human chromosomes; Standardization in Human Cytogenetics - Old classification and New classification.

#### **Unit – II**

The origin and transmission of chromosomal abnormalities- Non- disjunction; Types of numerical chromosomal abnormalities – Down syndrome, Edward syndrome, Patau syndrome, Turner syndrome, Klinefelter syndrome, XXX- syndrome and XYY- syndrome.

#### **Unit – III**

Structural chromosomal abnormalities - Cri-du-chat syndrome, Wolf- Hirschhorn syndrome, Fragile X – syndrome; Chromosomal instability syndromes; Heterochromatin and Lyon's Hypothesis; Uniparental disomy and Genomic imprinting

#### **Unit – IV**

Gene mapping- Somatic Cell Hybridization ; Molecular Cytogenetics - Fluorescence In Situ Hybridization; Comparative Genomic Hybridization; Array Comparative Genomic Hybridization; Marker chromosomes; Human artificial chromosomes.

#### **Unit – V**

Disorders of sex development – Primary Amenorrhea, Gonadal dysgenesis, Testicular feminization, Pseudohermaphroditism, True hermaphroditism; Infertility - Genetic basis of male infertility, Genetic basis of female infertility; Chromosomal abnormalities in recurrent pregnancy loss.

#### **Books suggested:**

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. Basic Human Genetics – E.J. Mange and A.P. Mange.
5. New Chromosomal Syndromes - J.J. Yunis

**SEMESTER-I**  
**PAPER 1.4 - MOLECULAR GENETICS**

**Course Outcomes:**

CO1: Proficiency in nucleic acid chemistry opens up career opportunities in research, biotechnology and diagnostic industries.

CO2: Gain understanding in DNA replication and repair mechanisms.

CO3: Capable of designing strategies to modulate gene expression for therapeutic purposes.

CO4: Grasp the broader implications of epigenetics in development, health, and disease and appreciate the potential of epigenetic therapies in treating diseases including cancer.

CO5: Gain expertise in translational regulation and post translational modifications.

**Learning Outcomes:**

LO1: Recall the central dogma and the structural features of DNA and RNA.

LO2: Understand how cells maintain genomic integrity and repair damaged DNA.

LO3: Develop a deep understanding of epigenetic modifications and their role in regulating gene expression.

LO4: Examine the role of RNA polymerases and regulatory elements in transcription which helps in development of nucleic acids-based drugs/vaccines.

LO5: Acquire insights into the dynamic and intricate processes that govern protein synthesis and function.

**Course Specific Outcomes:**

CSO1: Understand the concept of genome organization, including genes, chromosomes, and genetic elements.

CSO2: Obtain knowledge of DNA repair mechanisms which has implications for cancer therapy

CSO3: Develop proficiency in techniques such as chromatin immunoprecipitation analysis, mobility shift assay, and DNA foot printing for studying DNA-protein interactions.

CSO4: Investigate the functional significance of post-translational modifications and protein trafficking in cellular processes.

**CO-PO Mapping**

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓		✓	✓	✓	✓		✓
CO2	✓	✓	✓	✓	✓		✓	
CO3	✓	✓	✓	✓	✓			✓
CO4	✓	✓	✓	✓	✓	✓	✓	✓
CO5	✓	✓	✓	✓	✓		✓	✓

### **Unit- I**

Central dogma; Structure of nucleic acids - DNA and RNA and their types; Chromatin organization; Genome organization - Mitochondrial genome, Nuclear genome - Unique sequences and Reiterated sequences, Transposons, Pseudo genes and Gene families; Internal organization of gene.

### **Unit- II**

DNA replication - Enzymes involved in replication, Mechanism of replication in Prokaryotes and Eukaryotes; Regulation of DNA synthesis in Prokaryotes and Eukaryotes; DNA Damage - Endogenous damage and Exogenous damage; DNA Repair - Direct repair, Excision repair, Mismatch repair and Post replication repair.

### **Unit- III**

Transcription - RNA polymerases, Promoters, Transcription factors, Enhancers and Silencers; Mechanism of transcription in Prokaryotes and Eukaryotes; Post transcriptional modifications; DNA Protein interactions-Chromatin immunoprecipitation analysis, Mobility shift assay, DNA footprinting.

### **Unit- IV**

Regulation of transcription- Prokaryotes: Operon and Regulon, Eukaryotes: Inducible gene expression, Alternative promoters, Alternative splicing, Alternative polyadenylation, RNA editing, Post transcriptional gene silencing, Epigenetic mechanism.

### **Unit – V**

Translation- Genetic code; Protein synthesis in Prokaryotes and Eukaryotes; Translation regulation in Prokaryotes and Eukaryotes; Post translational modifications; Protein trafficking; Prions.

### **Books suggested:**

1. Lewin's GENES XII- J. E. Krebs et al.
2. Molecular Biology of the Gene- Watson et al.
3. Human Molecular Genetics- Strachan and Read
4. Principles of Genetics- Snustad and Simmons
5. Molecular Cell Biology-Lodish et al.
6. Molecular Biology- Weaver, R.F
7. Molecular Biology- Bruce Alberts et al.

## **SEMESTER-I**

### **PAPER- 1.5: PRACTICAL – I**

#### **Course Outcomes:**

CO1: Student will be able to demonstrate proficiency in performing ABO-typing and Rh (D) typing, and interpret the results accurately.

CO2: Graduate can analyze and identify the ABH Secretor status of individuals and explain its significance in genetics and forensic applications.

CO3: Student can evaluate and diagnose genetic traits such as color blindness and Phenyl Thio Carbamide (PTC) sensitivity through practical experiments.

CO4: Graduate can compute and interpret measures of central tendency and measures of dispersion, enabling data analysis and summarization.

CO5: Student will be able to calculate and interpret probabilities in genetic contexts, such as Mendelian genetics and genetic counseling.

#### **Learning Outcomes:**

LO1: Able to identify patterns in dermatoglyphics, specifically finger ball patterns and palmar patterns.

LO2: Can explain the genetic basis of blood group inheritance and its relevance in medical genetics.

LO3: Can apply Mendelian genetics principles to predict the inheritance of genetic traits.

LO4: Able to perform hypothesis tests and interpret their outcomes in a genetic context.

LO5: Can estimate gene frequencies and aware of the evolutionary forces acting on them.

#### **Course specific Outcomes:**

CSO1: Enhance critical thinking and problem-solving skills by applying genetic principles to real-world situations.

CSO2: Promote teamwork and collaboration among students when conducting experiments and analyzing genetic data.

CSO3: Encourage the application of statistical analysis to genetics research and hypothesis testing.

### **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

## **SEMESTER-I**

### **PAPER- 1.6: PRACTICAL – II**

#### **Course Outcomes:**

CO1: Student can perform karyotyping, including the identification of chromosomal abnormalities and disorders

CO2: Able to identify Drumstick and Barr bodies in cells and understand their significance in genetic diagnosis

CO3: Student will be able to isolate DNA and RNA from lymphocytes and understand the differences between DNA and RNA extraction techniques.

CO4: Can quantify genomic DNA accurately using various methods and instruments.

CO5: Able to extract DNA fragments from agarose gel, demonstrating proficiency in gel electrophoresis and purification techniques.

#### **Learning Outcomes:**

LO1: Analyze karyotype images to identify chromosomal abnormalities and genetic disorders.

LO2: Generate accurate metaphase drawings depicting chromosome arrangements.

LO3: Interpret G-banded chromosomes and recognize genetic anomalies.

LO4: Successfully isolate genomic DNA from blood samples, applying appropriate techniques and precautions.

LO5: Evaluate the quality of genomic DNA samples and troubleshoot issues related to sample purity and integrity.

#### **Course specific Outcomes:**

CSO1: Enhance observational and analytical skills in cytogenetics, enabling students to identify and analyze chromosomal aberrations and anomalies.

CSO2: Foster an understanding of the practical applications of chromosome analysis in genetics research, diagnostics, and genetic counseling.

CSO3: Develop proficiency in DNA isolation and quantification methods, preparing students for molecular biology research and clinical genetics applications.

### **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 RNA from Lymphocytes
3. Quantification of Genomic DNA
4. Quality check for Genomic DNA
5. Extraction of DNA from agarose gel



**SEMESTER II**  
**PAPER 2.1- CELL BIOLOGY AND SYSTEM PHYSIOLOGY**

**Course Outcomes:**

CO1: Able to identify different biomolecules and understand their roles in cellular processes and appreciate the complexity of cellular biology and its relevance to human health.

CO2: Can apply knowledge of nuclear ultra structure to inform strategies for disease prevention and intervention.

CO3: Thorough understanding of cell death processes, cell aging and cell signaling and communication. equips the student with the knowledge and skills to investigate disease mechanism and identify drug targets.

CO4: With the knowledge of cardiovascular physiology and pathology able to understand various disorders which has significant implications for healthcare.

CO5: Understand the mechanisms of hormone action, neuroendocrine regulation, and reproductive processes.

**Learning Outcomes:**

LO1: Understand the role of chemical constituents in cellular structure and function.

LO2: Apply the concepts of nuclear ultra structure to explain nuclear functions.

LO3: Assess the significance of cell death mechanisms and understand cell signaling pathways.

LO4: Can analyze the relationships between structure and function of different biological systems.

LO5: Able to understand how the biological systems are interconnected and work in harmony to maintain homeostasis, support growth and development, and ensure the survival and functioning of the organism.

**Course Specific Outcomes:**

CSO1: Obtain an insight into the significance of chemical constituents in cellular processes.

CSO2: Evaluate the implications of altered cellular interactions in the context of cancer research.

CSO3: Develop proficiency in explaining the differences between mitosis and meiosis and their relevance.

CSO4: Evaluate the impact of structural variations in the biological systems on physiological processes.

**CO-PO Mapping**

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓	✓	✓				✓
CO2	✓	✓	✓	✓	✓	✓		✓
CO3	✓	✓	✓	✓	✓	✓		✓
CO4	✓	✓	✓	✓	✓		✓	
CO5	✓	✓	✓	✓	✓		✓	

## **Unit- I**

Cell structure; Organelles of the cell- Golgi apparatus, Mitochondria, Ribosomes, Nucleus, Lysosomes, Peroxisomes, Endoplasmic reticulum, Vesicles, Cytoskeleton, Plasma membrane, Cytoplasmic matrix; Chemical constituents of cell- Carbohydrates, Lipids, Proteins, Nucleic acids, Vitamins, Minerals.

## **Unit- II**

Ultra structure of nucleus-Nuclear membrane, Nucleolus, Chromatin composition and organization; Cellular interactions- Differentiations of the Cell membrane, Intercellular communication and gap junctions, Cell coat and Cell recognition; The cell surface of cancer Cells.

## **Unit- III**

Cell division- Mitosis and Meiosis; Cell cycle and its regulation; Cell death (apoptosis, cell aging); Cell signaling and communication- Signal transduction: Cell signaling hormones and receptors, Cell surface receptors, Signaling through G-protein coupled receptors, Signal transduction pathways.

## **Unit-IV**

Circulatory system – Blood, Anatomy of the heart, Cardiac cycle, Blood pressure, Neural and chemical regulation; Respiratory system – Structure and function of lungs; Neural and chemical regulation; Nervous system – Brain, Spinal cord and Neurons, Central and Peripheral nervous system, Action potential and regulation; Skeletal and Muscular system.

## **Unit- V**

Digestive system – Digestion, Absorption; Excretory system – Structure and function of kidney, Regulation; Endocrine system – Endocrine glands, Mechanism of hormone action, Neuroendocrine regulation; Reproductive system – Female and Male reproductive systems, Reproductive processes.

### **Books suggested:**

1. The Cell, A Molecular Approach- G. M. Cooper and R. E. Hausman
2. Molecular Biology of the Cell - Bruce Alberts et al.
3. Cell and Molecular Biology- E.D.P. De Robertis and E.M.F. De Robertis, JR.
4. Molecular Cell Biology-Harvey Lodish et al.
5. Fundamentals of Anatomy and Physiology-F.H. Martini et al.
6. Gray's Anatomy for Students –Raveendranath Veeramani
7. Guyton and Hall Textbook of Medical Physiology –J.E. Hall et al.

## SEMESTER II

### PAPER-2.2: HUMAN BIOCHEMICAL AND IMMUNOGENETICS

#### Course Outcomes:

CO1: Apply knowledge of genetic variation to understand hemoglobinopathies, pharmacogenetics and ecogenetics which leads to a deeper understanding of the genetic basis of various health conditions and drug responses

CO2: Able to analyze inborn errors in various metabolic pathways .

CO3: Gain valuable insights into the intricacies of the immune system.

CO4: Possesses a solid foundation in immunology and genetics.

CO5: Able to evaluate the Major Histocompatibility Complex (MHC) and its classes, immunodeficiency diseases, autoimmunity, and immunotherapy.

#### Learning Outcomes:

LO1: Understand the types and significance of enzyme and protein polymorphisms.

LO2: Able to explain the genetic mechanisms underlying specific metabolic disorders and the clinical manifestations

LO3: Assess the role of phagocytes, complement system, natural killer cells, and immune cells in immune responses.

LO4: Able to examine the genetic factors involved in immunological processes and immune regulation.

LO5: Acquire knowledge of the genetic and immunological factors contributing to immune system dysfunction and potential therapeutic interventions.

#### Course Specific Outcomes:

CSO1. Acquire knowledge about the main concepts of human biochemical genetics and immunogenetics

CSO2. Familiar with the importance of pharmacogenetics and ecogenetics.

CSO3. Able to define the genetic systems that encode molecules with integral roles in immune regulation

CSO4. Gain knowledge about the importance of the immune system, and its role in health and disease.

#### CO-PO Mapping

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓	✓	✓	✓	✓		✓
CO2	✓	✓	✓	✓	✓	✓		✓
CO3	✓	✓	✓	✓	✓	✓		
CO4	✓	✓	✓	✓	✓	✓		✓
CO5	✓	✓	✓	✓	✓	✓		✓

### **Unit – I**

The concept of Biochemical polymorphism; Enzyme and protein 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- Sachs disease and Mucopolysaccharidoses; Disorders of Lipoprotein and lipid metabolism – Hyper Lipoproteinemia; Disorders of Purine metabolism- Lesch Nyhan syndrome; Disorders of Pyrimidine 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; Organization and structure of lymphoid organs – Bone marrow, Thymus, Spleen and Lymph nodes, Cells of the immune system – B Lymphocytes, T-Lymphocytes. T-cell receptor – Structure and function

### **Unit-IV**

Genetic basis of structure and diversity; Immunoglobulin molecules and the genetic basis of antibody diversity; Immunological memory; Immuno regulation; Adjuvants and immunological tolerance.

Nature of antigens and antibodies; Structure and function of antibodies; Isotypes, Allotypes and Idiotypes; Antigen – antibody interactions

### **Unit-V**

The Major Histocompatibility Complex-Class I (HLA-A,B,C,E,F & G), Class II (HLA-DP,DR,DQ) and Class III ( Complement genes ); Immunodeficiency diseases- Agammaglobulinemia, 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)

### **Books Suggested:**

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
6. Text book of Immunology - S.T, Barrot

**SEMESTER II**  
**PAPER 2.3 - MEDICAL GENETICS**

**Course Outcomes:**

CO1: Able to understand the scope of Medical Genetics and its relevance in healthcare.

CO2: Able to apply knowledge of genetic aspects to explain muscle disorders, ocular, auditory, and orofacial disorders.

CO3: Capable of identifying genetic disorders affecting multiple organ systems.

CO4: Synthesize knowledge of lifestyle-related disorders and bring a multidimensional perspective to healthcare.

CO5: Competent to evaluate neurodegenerative disorders, neuromuscular disorders and psychiatric disorders from a genetic perspective.

**Learning Outcomes:**

LO1: Able to recall the significance of Medical Genetics in identifying and managing genetic disorders.

LO2: Can apply genetic principles to understand the etiology and manifestations of muscular, ocular, auditory, and orofacial disorders.

LO3: Able to examine the genetic basis and clinical presentations of disorders in various organ systems.

LO4: Formulate explanations for the genetic and environmental factors contributing to lifestyle disorders.

LO5: Can assess the genetic and molecular basis of various neurological and psychiatric disorders.

**Course Specific Outcomes:**

CSO1: Analyze real-world case studies of individuals affected by genetic skin and skeletal disorders.

CSO2: Develop skills in assessing the genetic predisposition and lifestyle-related risk factors for common disorders.

CSO3: Able to explain the genetics of respiratory disorders, with a focus on Cystic fibrosis, and its implications for patient management.

CSO4: Can explain the genetic aspects of psychiatric disorders, with a focus on Schizophrenia and Bipolar disorder.

**CO-PO Mapping**

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓	✓	✓	✓	✓		
CO2	✓	✓	✓	✓	✓	✓		
CO3	✓	✓	✓	✓	✓	✓		
CO4	✓	✓	✓	✓	✓	✓		
CO5	✓	✓	✓	✓	✓	✓		✓

## **Unit – I**

Scope of Medical Genetics; Skin- Ichthyosis, Psoriasis, Multiple neurofibromatosis, Porphyrrias, Blooms syndrome; Skeletal system – Ankylosing spondylitis, Osteogenesis imperfecta, Rheumatoid arthritis; Osteoporosis; Marfans syndrome.

## **Unit – II**

Muscle – Muscular dystrophies, Myotonia; Eye – Glaucoma, Cataract, Retinoblastoma ; Jaws – Hare lip and Cleft palate; Ears - Deafness.

## **Unit – III**

Digestive system – Hypertrophic pyloric stenosis, , Cirrhosis of liver ; Respiratory system – Cystic fibrosis ; Cardiovascular system – Congenital heart disease; Central nervous system – Spina bifida, Anencephaly; Kidney and urogenital system – Cystinosis, Polycystic kidney ; Reproductive system- Polycystic ovary disease, Endocrine system – Congenital hypothyroidism or Cretinism, Goiter.

## **Unit – IV**

Life style disorders- Diabetes, Hypertension, Hyperlipidemia, Coronary heart disease, Stroke, Obesity, Anxiety, Chronic obstructive pulmonary disease.

## **Unit – V**

Neurodegenerative disorders- Parkinson's disease, Alzheimer's disease, Spinocerebellar ataxia, Prion disease; Neuromuscular disorders- Multiple sclerosis, Amyotrophic lateral sclerosis, Myasthenia gravis; Psychiatric disorders- Schizophrenia, Bipolar disorder.

### **Books Suggested:**

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 Young
6. Human Genetics – F. Vogel and A.G. Motulsky

## SEMESTER II

### PAPER 2.4 - DIAGNOSTIC MOLECULAR PATHOLOGY

#### Course Outcomes:

CO1: Students will have a strong foundation in Molecular Biology, and Genomics and able to explain various types of genetic mutations.

CO2: Can apply knowledge of various molecular techniques for detecting genetic mutations and can contribute to the early diagnosis of genetic diseases.

CO3: Proficient in various cloning techniques which are essential for isolating and characterizing disease-related genes.

CO4: Competent to diagnose infectious and genetic diseases by utilizing molecular diagnostic techniques.

CO5: Can evaluate the role of biomarkers in disease diagnosis, monitor disease progression, treatment response, and recurrence, preclinical testing, and clinical trial design.

#### Learning Outcomes:

LO1: Understand the mechanisms and consequences of various genetic mutations.

LO2: Apply molecular techniques to detect genetic mutations in different contexts.

LO3: Examine the approaches used for cloning disease genes, with a focus on specific examples like Phenylketonuria, Hemophilia etc.

LO4: Can analyze how biomarkers can predict disease risk, helping individuals make informed lifestyle choices and healthcare decisions

LO5: Have knowledge of advanced techniques like Imaging Mass Spectrometry, Whole Genome Sequencing, and Exome Sequencing techniques that have revolutionized medical diagnosis.

#### Course Specific Outcomes:

CSO1: Have a deep understanding of the different types of genetic mutations that can occur within an individual's DNA.

CSO2: Gain knowledge of genetic disorders and molecular diagnosis and with their expertise can contribute to public health efforts, such as newborn screening programs.

CSO3: Learn molecular diagnostic techniques which are crucial in clinical laboratories to perform genetic tests that aid in patient diagnosis and treatment.

CSO4: Understand how biomarkers can be used to predict disease risk and their role in design and execution of clinical trials

#### CO-PO Mapping

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓	✓	✓	✓	✓		✓
CO2	✓	✓	✓	✓	✓	✓	✓	✓
CO3	✓	✓	✓	✓	✓	✓		
CO4	✓	✓	✓	✓	✓	✓		
CO5	✓	✓	✓	✓	✓	✓		✓

## **Unit- I**

Molecular Pathology – Gene deletions, Codon deletions, Duplications and Insertions, Point mutations, RNA Splice mutations; Transcriptional mutations, Dynamic mutations and Mitochondrial mutations; Mutation nomenclature; Molecular explanation of dominance and recessiveness.

## **Unit- II**

DNA fingerprinting; Molecular techniques for detection of genetic mutations – PCR, PCR - RFLP, ARMS - PCR, Multiplex PCR, Nested PCR, RT PCR, Real time PCR, LAMP PCR, SSCP, DGGE, Hetero duplex analysis, PTT.

## **Unit- III**

Strategies for cloning of disease genes; Functional cloning - Phenylketonuria, Hemophilia; Positional cloning - Cystic fibrosis, Huntington's disease, Duchene muscular dystrophy.

## **Unit- IV**

Molecular diagnosis of Infectious diseases and Genetic diseases: Direct testing - Sickle cell anemia, Beta thalassemia, Cystic fibrosis, Huntington's disease, Duchene muscular dystrophy; Gene tracking - Cystic fibrosis and Huntington's disease.

## **Unit- V**

Biomarkers; Molecular Testing in Personalized Medicine – Breast Cancer, Long QT Syndrome, Hereditary Diabetes, Neurological disorders; Imaging Mass Spectrometry in Clinical Pathology; Whole Genome Sequencing; Exome Sequencing.

## **Books suggested:**

1. Human Genetics-The Molecular Revolution-E. H. McConkey, Jones and Bartlett
2. Molecular Genetics for the Clinician- D.J.H. Brock
3. Principles of Genetics- Snustad and Simmons
4. Human Molecular Genetics- Strachan and Read
5. Polymerase Chain Reaction- S. Shafique
6. PCR, The Polymerase Chain Reaction- Kary B. Mullis
7. Diagnostic Molecular Pathology - William Coleman, Gregory Tsongalis



## **SEMESTER-II**

### **PAPER- 2.5: PRACTICAL – III**

#### **Course Outcomes:**

CO1: Student will be able to identify and describe the structure and function of key eukaryotic cell organelles.

CO2: Can differentiate mitotic chromosomes in onion root tips and meiotic chromosomes in onion flowers.

CO3: Able to conduct the sickling test and accurately diagnose the presence of abnormal hemoglobins.

CO4: Can estimate hemoglobin (Hb) levels in blood samples using standard methods.

CO5: Able to perform Disc-PAGE to identify polymorphisms of haptoglobin and ceruloplasmin.

#### **Learning Outcomes:**

LO1: Able to recognize and identify cell organelles accurately using microscopic observations.

LO2: Can explain the differences between mitosis and meiosis and describe chromosome behavior during these processes.

LO3: Able to conduct electrophoresis to separate and identify abnormal hemoglobins in blood

LO4: Able to explain the clinical relevance of hemoglobin A2 and its significance in diagnosing thalassemias.

LO5: Can analyze red cell enzyme activity and erythrocyte sedimentation rate to assess health conditions.

#### **Course specific Outcomes:**

CSO1: Cultivate expertise in laboratory techniques for cell biology and hematology, including sample preparation and analysis.

CSO2: Enhance knowledge of hematological parameters and their use in diagnosing and monitoring various blood disorders.

CSO3: Promote critical thinking and problem-solving skills in interpreting laboratory results and making clinical correlations.

### **Part – A**

1. Spotters- Eukaryotic Cell Organelles- Membrane, Nucleus, Nucleolus, Mitochondria, Endoplasmic Reticulum, Golgi Apparatus, Chloroplasts, Ribosomes.
2. Mitotic Chromosomes in Onion Root Tips
3. Meiotic Chromosomes in Onion Flowers

### **Part – B**

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

## **SEMESTER-II**

### **PAPER- 2.6: PRACTICAL – IV**

#### **Course Outcomes :**

CO1: Student will gain a comprehensive understanding of medical genetics, its principles, and its relevance in clinical practice.

CO2: Student will be able to recognize skeletal disorders such as Rheumatoid Arthritis and Osteogenesis Imperfecta and explain their genetic underpinnings.

CO3: Develop the ability to design primers for PCR (Polymerase Chain Reaction) experiments targeting specific genetic sequences.

CO4: Can perform PCR-RFLP (Restriction Fragment Length Polymorphism) experiments for genetic variant detection and analysis, which has great application in molecular diagnostics.

CO5: Graduate can execute multiplex PCR (Polymerase Chain Reaction) to simultaneously amplify multiple DNA targets in a single reaction, which is useful in diagnosing genetic diseases like DMD in the laboratory.

#### **Learning Outcomes:**

LO1: Able to explain the fundamental concepts of medical genetics and its applications in diagnosing and managing genetic disorders.

LO2: Can evaluate the genetic factors contributing to cardiovascular and endocrine disorders and their implications.

LO3: Able to explain the principles of insertion-deletion polymorphism and its significance in genetic diversity studies.

LO4: Execute ARMS-PCR for the detection of specific genetic mutations or alleles

LO5: Interpret the results of molecular techniques accurately and apply them to genetic research and diagnostics.

#### **Course specific Outcomes:**

CSO1: Cultivate expertise in explaining the genetic basis of various disorders and their clinical implications.

CSO2: Foster critical thinking skills in analyzing the genetic aspects of cardiovascular and endocrine disorders.

CSO3: Student will get hands-on experience in genetic variant analysis using molecular techniques for research and clinical applications

**Part – A**  
( Assignment )

1. Introduction to Medical Genetics
2. Skin disorders- Ichthyosis, Multiple Neurofibromatosis, Blooms syndrome
3. Skeletal disorders- Rheumatoid Arthritis, Osteogenesis Imperfecta
4. Muscle disorders- Muscular Dystrophies
5. Eye disorders- Glaucoma, Retinoblastoma
6. Cardiovascular disorders- Congenital heart disease, coronary heart disease, hypertension
7. Endocrinal disorders- Cretinism, Goiter

**Part – B**

1. Primer designing
2. Insertion deletion polymorphism
3. PCR- RFLP
4. ARMS-PCR
5. Multiplex PCR

### SEMESTER III

#### PAPER 3.1 - GENETIC TOXICOLOGY AND CANCER GENETICS

##### Course Outcomes:

CO1: Students will be equipped with the knowledge and skills needed to understand and address the complex issues associated with genetic toxicology.

CO2: Able to assess and manage the potential genotoxic effects of toxicants in various settings.

CO3: Possess strong foundation in the genetics and biology of cancer, enabling them to critically analyze the role of genetic factors in cancer development.

CO4: Get deep insight into the genetic basis of cancer, the role of carcinogens in cancer development, diagnosis, prevention and treatment of cancer.

CO5: Proficient in the biology of cancer stem cells, the pathways involved in their regulation, and the various therapeutic strategies aimed at targeting these cells.

##### Learning Outcomes:

LO1: Able to understand different types of mutagens and the chromosomal changes caused by them.

LO2: Get a hold on the basic sites of toxic exposure, their distribution, metabolism and excretion.

LO3: Get deep insight into the characteristics of cancer cells and chromosomal changes in neoplasia

LO4: Gain knowledge on Knudson's hypothesis and different types of cancers.

LO5: Acquire knowledge on cancer prevention, diagnosis and treatment.

##### Course Specific Outcomes:

CSO1: Acquire knowledge on genotoxicity and its consequences in humans.

CSO2: Able to distinguish different types of cancers.

CSO3: Obtain an insight in to the genes involved in the development of cancer.

CSO4: Understand the importance of cancer stem cells in the initiation and progression of cancer

##### CO-PO Mapping

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓	✓	✓	✓			
CO2	✓	✓	✓	✓	✓			
CO3	✓	✓	✓	✓	✓	✓		
CO4	✓	✓	✓	✓	✓	✓		
CO5	✓	✓	✓	✓	✓	✓		

## **Unit – I**

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 – II**

Routes and sites of exposure; Absorption; Distribution and excretion of toxicants; Xenobiotic metabolism; Consequences of genotoxic effects in humans; Techniques to detect genotoxicity; Treatment

## **Unit – III**

Introduction to cancer; Characteristics of cancer cells; Chromosomes in neoplasias and relation of oncogenes to chromosomal defects; Chromosomal instability syndromes; Cancer as a genetic disorder and Cancer in families; Inherited versus Sporadic cancers; The role of Epigenetics in cancer

## **Unit – IV**

Molecular changes in proto-oncogenes; Tumor suppressor genes; Knudson's hypothesis; Types of cancers – Retinoblastoma, Skin cancer, Lung cancer, Esophageal and Colon cancer, Brain cancer, Breast cancer, Cervical cancer, Prostate cancer; Cancer and environment - Physical, Chemical and Biological carcinogens; Cancer prevention; Diagnosis; Treatment

## **Unit – V**

Cancer stem cells: Introduction to stem cells, Angiogenesis, Stochastic versus Cancer stem cell model for cancer formation, Cancer stem cells in cancer initiation and progression, Cancer stem cell pathways; Regulation by microRNAs; Therapeutic strategies

### **Books suggested:**

1. Molecular Biology in Medicine - Cox and Sinclair
2. Principles of Genetic Toxicology- David Brusick.
3. Genetic Toxicology- Albert P, Li, R.H, Heflich.
4. Principles and Practice of Medical Genetics- Rimoin et al.
5. Clinical Genetics- Robinson and Linden
6. Clinical Genetics: A Short Course – Wilson
7. Cancer Genetics E-book-Cubocube (Wasserman Laboratory, University of British Columbia (UBC))

## SEMESTER III

### PAPER 3.2 - GENETIC ENGINEERING

#### Course Outcomes:

CO1: Familiarize with the tools of recombinant DNA technology enabling them to work in research, biotechnology, or related fields.

CO2: Equipped with knowledge and skills necessary to apply various gene transfer and hybridization techniques in research, biotechnology, agriculture, and medicine.

CO3: Good grasp of the principles and practical skills involved in molecular cloning and gene expression.

CO4: Acquaint with the process of mutagenesis and applications of Genetic Engineering.

CO5: Get an insight in to genome editing technology, role of stem cells in regenerative medicine and significance of cord blood banking.

#### Learning Outcomes:

LO1: Gain knowledge on the various enzymes and vectors used in rDNA technology.

LO2: Understand the different steps involved in cloning the gene of interest and expression of the cloned gene in different expression systems.

LO3: Attain knowledge regarding the creation of mutation to examine the function of a gene or to produce protein with a novel function.

LO4: Appreciate genome editing tools like CRISPR-Cas9 and applications of genome editing technology.

LO5: Have knowledge regarding the types, differentiation and use of stem cells.

#### Course Specific Outcomes:

CSO1: Well versed with the tools used in Genetic Engineering.

CSO2: Thorough knowledge of the process of cloning a gene of interest and expressing it.

CSO3: Skill development in handling restriction digestion, ligation etc. in laboratory.

CSO4: Have an understanding of genome editing

#### CO-PO Mapping

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓	✓	✓	✓			
CO2	✓	✓	✓	✓	✓			
CO3	✓	✓	✓	✓	✓			
CO4	✓	✓	✓	✓	✓			
CO5	✓	✓	✓	✓	✓			

## **Unit-I**

Enzymes used in rDNA technology; Vectors for Gene Cloning; Isolation and purification of genomic and plasmid DNA; Electrophoresis - Agarose Gel electrophoresis of nucleic acids, Pulsed-field gel electrophoresis of DNA; Denaturing SDS PAGE, Native Non-denaturing PAGE, 2D gel electrophoresis

## **Unit – II**

Gene Transfer Techniques - Electroporation, Microinjection, Lipofection; PEG mediated, Calcium chloride mediated, DEAE dextran mediated, Transformation, Transfection; Oligonucleotide synthesis; Preparation of probes; Hybridization Methods - Southern, Northern and Western

## **Unit- III**

Concept of Gene cloning; Ligation of DNA fragments; Recombinant selection; Construction of genomic and cDNA libraries; Screening of cDNA libraries; Selection of rDNA clones; Expression of the recombinant DNA in the host; Purification of recombinant protein

## **Unit- IV**

Mutagenesis - Random mutagenesis, Site specific mutagenesis, Transposon mutagenesis; Construction of knock-out mutants; Applications of Genetic Engineering - Crop and live-stock improvement, Transgenic plants and animals, DNA drugs and vaccines, Identification of human disease genes; Biosafety, Hazards and ethical considerations of Genetic Engineering

## **Unit- V**

Genome editing technology: Structure and mechanism of genome editing tools - CRISPR/Cas9; Genome editing for disease modeling and gene therapy; Applications of genome editing technology; Genetic engineering of stem cells for enhanced therapy - Cord blood banking, Types of stem cells, Differentiation, Stem cell therapy in treatment of different diseases

## **Books suggested:**

1. Gene Cloning and DNA Analysis, An Introduction - T.A. Brown
2. Principles of Gene Manipulation - R.W. Old and S.B. Primrose
3. Recombinant DNA - J.D. Watson et al.
4. Genetic Engineering - Sandhya Mitra
5. Recombinant DNA Technology - Keya Chaudhuri
6. Molecular Biology and Genetic Engineering - P. K. Gupta
7. Genome editing - Toni Cathomen, Matthew Hirsch, Matthew Porteus



## SEMESTER III

### 3.3 RESEARCH METHODOLOGY

#### Course Outcomes:

CO1: Apprehend the meaning, significance, and objectives of research and able to select and formulate a research problem.

CO2: Can justify the need for a research design in the context of scientific inquiry.

CO3: Develop strong skills in statistical interpretation, hypothesis testing, and data analysis.

CO4: Competent to develop a research project protocol that aligns with ethical and scientific standards.

CO5: Able to prepare a manuscript for publication adhering to standard guidelines.

#### Learning Outcomes:

LO1: Create hypotheses and effectively articulate research statements.

LO2: Able to determine appropriate sample sizes considering general considerations and power analysis.

LO3: Distinguish between Type I and Type II errors and explain their implications.

LO4: Understand the procedure for obtaining a p-value and its interpretation.

LO5: Recognize and address ethical considerations, including conflicts of interest and plagiarism.

#### Course specific outcomes:

CSO1: Competent to perform literature reviews using resources like Medline, Entrez, PubMed, Scopus etc.

CSO2: Choose an appropriate research design based on research goals and constraints.

CSO3: Develop a well-structured research proposal for a study in the domain of Human Genetics.

CSO4: Present research findings effectively through scientific lectures, posters, and visual displays.

#### CO-PO Mapping

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓					✓	✓
CO2	✓	✓					✓	✓
CO3	✓	✓					✓	✓
CO4	✓	✓					✓	✓
CO5	✓	✓					✓	✓

## **UNIT - I**

Meaning, significance and objectives of research; Criteria of good research; Merits and demerits; Different types of research; Current research areas in Human Genetics

## **UNIT – II**

Selection of research problem, Statement formulation and types of hypothesis; Review of literature from sources like Index Medicus, Medline, Entrez and PubMed based on SCI, Scopus and ICI; Research variables- Meaning, Types and their significance in research; Writing a research proposal

## **UNIT – III**

Need for a research design; Features of a good design; Types of research designs - Case Study, Cohort study, Case - control study and Cross-sectional studies with merits and demerits; Sample size determination - General consideration and power analysis; Sampling techniques

## **UNIT – IV**

Statistical significance; Hypothetical testing; Type -I and Type - II errors; General procedure to obtain P value; Use of SPSS software; Test of methods through sensitivity, specificity and likelihood ratio; Positive and negative predictivity; Odds ratio for independent and matched samples

## **UNIT - V**

Preparing a manuscript for publication; Editing and gallery proof correction of manuscript; Publication ethics; Conflicts of interest; Plagiarism; Protocol for research project; Preparation and presentation of scientific lecture; Guidelines for putting up for poster display

### **Books suggested:**

1. Research Methodology Methods and Techniques, New Age International (P) Ltd. Publishers, C.R.Kothari, (2004)
2. Microsoft Excel Data Analysis and Business Modeling, Microsoft Press, Wayne L. Winston, (2004)
3. Research Methodology: a step-by-step guide for beginners, 4<sup>th</sup> edition, Pearson Education, Kumar, (2014)
4. Practical Research Methods, UBSPD Pvt. Ltd. Dawson, C, (2002)
5. Practical Statistics for Data Scientists: 50 Essential Concepts, Peter Bruce and Andrew Bruce, (2020)
6. How to Write a Lot: A Practical Guide to Productive Academic Writing, Paul J. Silvia, (2018)
7. The Handbook of Scholarly Writing and Publishing, Tonette S. Rocco and Tim Hatcher (2011)

## SEMESTER III

### 3.4 DATA ANALYSIS FOR BIOLOGISTS

#### Course Outcomes:

CO1: Able to differentiate between various types of data encountered in biology and recognize their significance in analysis.

CO2: Create diverse graphical representations, such as scatter plots, bar graphs, pie charts, box plots, and histograms, to visually interpret biological data.

CO3: Utilize basic principles of linear algebra to comprehend and analyze biological data structures.

CO4: Develop competence in using tools and software like Microsoft Excel and GraphPad for data entry, analysis, and graphical representation.

CO5: Acquire a foundational understanding of R and RStudio for data analysis, visualization, and manipulation and generate customized graphics and visualizations using R's ggplot2 package.

#### Learning Outcomes:

LO1: Use Microsoft Excel proficiently for data entry, simple calculations, and generating basic graphs

LO2: Employ GraphPad to produce various graphical representations, enhancing data visualization.

LO3: Navigate and utilize R and RStudio environments effectively for data analysis and visualization.

LO4: Prepare and format data files for analysis and presentation

LO5: Ability to apply acquired skills to analyze and interpret biological data in research

#### Course specific outcomes:

CSO1: Demonstrate the ability to retrieve and interpret relevant data from biological databases

CSO2: Utilize basic linear algebra concepts to analyze biological data structures, such as gene expression matrices

CSO3: Generate graphical representations and interpret and communicate findings from them

CSO4: Create a variety of plots using the ggplot2 package, customize and save them in different formats for presentation and publication.

#### CO-PO Mapping

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓					✓	
CO2	✓	✓					✓	
CO3	✓	✓					✓	
CO4	✓	✓					✓	
CO5	✓	✓					✓	

## **Unit - I**

Introduction to data; Levels of biological study; Different types of data in biology; Importance of data analysis of biological data; Biological databases – types, storage and retrieval.

## **Unit - II**

Descriptive statistics; Different types of plots - Scatter plot, Bar graph, Line graph, Pie chart, Box plot, Frequency histogram, Heat maps; Understanding error bars; Basic concepts of probability; Bayes theorem; Binomial, Multinomial, Poisson, Exponential, and Gaussian distribution; Basic concepts of linear algebra

## **Unit - III**

Introduction to Microsoft Excel; Using Microsoft Excel for data entry; Features and formula in Excel; Generation of graphical representations using Microsoft Excel; Introduction to GraphPad; Generation of graphical representation using GraphPad – Bar plots, Scattered plots and heat maps.

## **Unit - IV:**

Introduction to R and RStudio; Rscript vs console; Working Directory; R basic syntax; Introduction to variable types and functions; Manipulate vectors, Matrices and data frames; Read and load data into R

## **Unit - V:**

Getting started with data –Data files; Formatting the data; Generation of plots – plotting with ggplot2; Customizing the plots and saving the plots.

## **Books suggested:**

1. R. C. Elston and W. D. Johnson, Basic Biostatistics for Geneticists and Epidemiologists: A Practical Approach, 1<sup>st</sup> Edition, Wiley, 2008.
2. Robert S. Witte and John S. Witte, (2018), Statistics, 9<sup>th</sup> edition, John Wiley & Sons. Inc
3. M. C. Whitlock, and D. Schluter, The Analysis of Biological Data, 2<sup>nd</sup> Edition, W. H. Freeman & Company, 2014.
4. G.P. Quinn, and M. J. Keough, Experimental Design and Data Analysis for Biologists, Cambridge University Press, 2002
5. M.D. Ugarte, A. F. Militino, and A. T. Arnholt, Probability and Statistics with R, 2<sup>nd</sup> Edition, CRC Press, 2016
6. James Gareth, Witten Daniela, Hastie Trevor and Tibshirani Robert. An introduction to statistical learning with application in R. Vol. 112. New York: Springer, 2013
7. Hadley Wickham, (2016), ggplot2: Elegant Graphics for Data Analysis, Springer

## **SEMESTER-III**

### **PAPER- 3.5: PRACTICAL – V**

#### **Course Outcomes:**

CO1: Graduate will acquire a foundational understanding of genetic toxicity, including its principles, causes, and consequences.

CO2: Student will be able to identify the various routes and sites of exposure to genotoxic agents and assess their implications.

CO3: Student will explore molecular changes in proto-oncogenes and their contributions to tumorigenesis.

CO4: Graduate will get proficiency in the preparation of plasmids for molecular cloning experiments.

CO5: Student can extract DNA fragments from agarose gel matrices, ensuring their purity and suitability for downstream applications.

#### **Learning Outcomes:**

LO1: Apply Knudson's hypothesis to understand the genetic basis of cancer susceptibility.

LO2: Able to explain the molecular mechanisms underlying chromosomal alterations and sister chromatid exchanges.

LO3: Can analyze the health consequences of genotoxic effects, including the development of cancer.

LO4: Able to construct restriction maps and interpret them for molecular analysis and genetic mapping.

LO5: Perform DNA ligation to create recombinant DNA molecules for cloning.

#### **Course specific Outcomes:**

CSO1: Cultivate a deep understanding of genetic toxicology principles, equipping students to assess and mitigate genotoxic risks.

CSO2: Foster critical thinking skills in analyzing the genetic basis of cancer and its implications for human health.

CSO3: Foster the ability to interpret and communicate molecular and genetic results effectively in a laboratory or research context.

**Part – A**  
(Assignment)

1. Fundamentals of genetic toxicity
2. Mechanism of induction of chromosomal alterations and sister chromatid exchanges
3. Routes and sites of exposure
4. Consequences of genotoxic effects in humans
5. Inherited versus sporadic cancers
6. The role of epigenetics in cancer
7. Molecular changes in proto- oncogenes
8. Knudson's hypothesis
9. Cancer stem cells
10. Cancer stem cell pathways

**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

## **SEMESTER-III**

### **PAPER 3.6: PRACTICAL – VI**

#### **Course Outcomes:**

CO1: Student can identify and access recent research papers in the field of biology and explain the significance of the studies.

CO2: Graduate can perform effective literature searches using databases like PubMed and present findings coherently.

CO3: Student will be able to practice various sampling methods, such as random sampling, stratified sampling, and cluster sampling.

CO4: Able to explore diverse biological databases and retrieve specific information relevant to research questions.

CO5: Graduate will be able to install R and RStudio, set up working directories, and write and execute basic R scripts.

#### **Learning Outcomes:**

LO1: Generate research hypotheses and justify their selection based on scientific reasoning.

LO2: Construct well-structured research proposals with clear problem statements, objectives, methods, and expected outcomes.

LO3: Calculate and interpret P-values to make informed statistical inferences.

LO4: Evaluate the suitability of different probability distributions for modeling biological datasets.

LO5: Customize and enhance the visual appeal of plots in GraphPad.

#### **Course specific Outcomes:**

CSO1: Develop a strong foundation in research methods and data analysis techniques, preparing students for scientific inquiry.

CSO2: Develop proficiency in data visualization and interpretation using graphical plots.

CSO3: Encourage adaptability in data analysis tools and methods for future research endeavors.

### **Part - A**

1. Searching for recent research papers and explaining the importance of the studies.
2. Evaluate published research articles to identify and discuss the criteria that make them "good" research. Analyze aspects like study design, methodology, data analysis, and conclusions.
3. Search and review literature on a given topic using databases like PubMed, and present findings.
4. Formulate hypotheses for the given problem and explain their rationale.
5. Draft a concise research proposal on a hypothetical research problem, including problem statement, objectives, methods, and expected outcomes (team work).
6. Analyze and compare different research designs (case study, cohort study, case-control study, cross-sectional study) using real research examples.
7. Determining sample size for given research scenario
8. Practice various sampling methods, such as random sampling, stratified sampling, and cluster sampling.
9. Calculation of P-values for different statistical tests for the provided dataset related to genetics
10. Calculate and interpret Type I and Type II errors based on hypothesis testing outcomes for given research scenario
11. Calculation of odds ratios for different sample groups. Odds Ratio Calculation: Provide data from a genetic study and guide students through calculating odds ratios for different sample groups.
12. Preparation of a draft manuscript for a hypothetical research project including introduction, methods, results, and discussion sections.
13. Discussion on case studies involving ethical dilemmas, including plagiarism issues
14. Presentation of hypothetical research findings through oral presentations and poster displays

### **Part-B**

1. Explore different biological databases and retrieve specific information relevant to a given research question.
2. Generate a frequency histogram and a box plot to depict the distribution of a quantitative biological variable.
3. Fit different probability distributions (binomial, Gaussian, etc.) to biological datasets and compare their suitability.
4. Calculate descriptive statistics for selected data columns using Excel functions and prepare graphical plots in Excel and by using GraphPad
5. Customize the appearance of plots in GraphPad, including labels, colors, and legends.
6. Install R and RStudio, set up a working directory, and write and execute basic R scripts
5. Create and manipulate vectors and matrices in R, performing arithmetic operations
6. Load a biological dataset from a CSV file, explore its structure, and perform summary statistics.
7. Subset and filter data in R to extract specific subsets for analysis
8. Practice writing R code to generate basic plots using base R graphics
9. Prepare a biological dataset by cleaning and formatting it for analysis in R.
10. Use the ggplot2 package to create scatter plots, bar plots, and histograms from the dataset and customize them including labels, titles, colors, and themes in ggplot2.
11. Create side-by-side bar plots and compare distributions of different groups within the dataset.



12. Save plots as image files (PNG, JPEG) and incorporate them into a report or presentation.

**Books Suggested:**

1. Research Methodology Methods and Techniques, New Age International (P) Ltd. Publishers, C.R.Kothari, (2004),
2. Microsoft Excel Data Analysis and Business Modeling, Microsoft Press, Wayne L. Winston, (2004),
3. Research Methodology: a step-by-step guide for beginners, 4 th edition, Pearson Education, Ranjit Kumar, (2014),
4. Practical Research Methods, UBSPD Pvt. Ltd. Dawson, C, (2002),
5. Research Methodology, KSK Publishers, New Delhi. Sharma, N. K
6. Research Methods: The Basics, 2 nd edition Nicholas G. Colomb, and Joseph M. Williams, (2016)
9. How to Write a Lot: A Practical Guide to Productive Academic Writing, Paul J. Silvia, (2018)
10. Case Study Research and Applications: Design and Methods, Robert K. Yin, (2017)
11. Statistics for People Who (Think They) Hate Statistics, 5 th edition, SAGE publications Neil J. Salkind, (2021)
12. Practical Statistics for Data Scientists: 50 Essential Concepts, Peter Bruce and Andrew Bruce, (2020)
13. Publication Manual of the American Psychological Association, 7 th edition American Psychological Association (APA), (2020)
14. Writing for Publication in Nursing and Healthcare: Getting it Right, Karen Holland and Roger Watson, (2018)
15. The Handbook of Scholarly Writing and Publishing, Tonette S. Rocco and Tim Hatcher (2011)

## **SEMESTER-IV**

### **PAPER 4.1-GENETIC SCREENING, COUNSELING AND GENE THERAPY**

#### **Course Outcomes:**

CO1: Gain a comprehensive understanding of the scope and medical importance of genetic screening, its various methods, and its role in prenatal and postnatal healthcare, population health and family planning.

CO2: Equipped with knowledge and understanding of prenatal and postnatal screening methods and also population screening for genetic diseases.

CO3: Able to conduct effective genetic counseling, addressing educational, emotional, ethical, and genetic aspects, and providing guidance to individuals and families dealing with genetic conditions.

CO4: Gain a comprehensive understanding of the classifications and types of gene therapy, enabling them to appreciate the progression of gene therapy and its potential in treating genetic disorders and diseases.

CO5: Able to apply appropriate techniques effectively to identify carriers of autosomal recessive genetic disorders, aiding in genetic counseling.

#### **Learning Outcomes:**

LO1. Get deep insight into population screening for genetic diseases.

LO2. Enhanced knowledge on the Prenatal and postnatal screening methods

LO3. Acquire knowledge on scope of genetic counseling and methods of counseling, patterns of inheritance and risk assessment.

LO4. Obtain knowledge on gene therapy, its classification and its clinical applications.

LO5. Get a hold on the detection of autosomal recessive carriers.

#### **Course Specific Outcomes:**

CSO1. Acquire knowledge on the screening tests used for genetic testing before and after birth of a child to identify certain birth defects.

CSO2. Able to identify the genetic risk factors based on an expert review of personal and family health histories.

CSO3. Able to manage the health care and future perspectives of gene mapping and gene therapy

CSO4: Well-equipped with the knowledge, skills, and ethical understanding necessary to contribute effectively to the field of genetic analysis and counseling.

### **CO-PO Mapping**

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓	✓	✓	✓	✓	✓	✓
CO2	✓	✓	✓	✓	✓	✓	✓	✓
CO3	✓	✓	✓	✓	✓	✓	✓	✓
CO4	✓			✓	✓			✓
CO5	✓	✓				✓		

#### **Unit – I**

Scope of Genetic Screening-Invasive and Noninvasive testing methods - Prenatal and Postnatal screening. Population screening for genetic diseases, Newborn screening, Carrier screening, Family screening and their medical importance

#### **Unit - II**

Prenatal screening methods- Amniocentesis, Chronic Villous sampling, Ultrasonography, Fetoscopy, Maternal blood sampling.

Postnatal screening - Chromosomal abnormalities both structural and numerical 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**

History of Gene Therapy - Classification of gene therapy- Class-I, Class II and Class

#### **Unit - V**

Detection of autosomal recessive carriers - Huntington's Disease - Restriction length polymorphisms (RFLPs) and DNA probes-Cystic fibrosis - Sickle cell Disease.

#### **Books suggested:**

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 Jorde et al
5. Genetic Counseling by W. Fuhrmann and F. Vogel

## SEMESTER-IV

### PAPER 4.2- GENOMICS, PROTEOMICS AND BIOINFORMATICS

#### Course Outcomes:

CO1: Develop critical thinking skills necessary to make informed decisions regarding mapping techniques and DNA markers, aligning them with research goals.

CO2: Gain a comprehensive understanding of DNA sequencing techniques, advanced genomic technologies, Bioinformatics tools, and the role of Genomics in studying both the structure and function of genes and genomes.

CO3: Gain a strong foundation in understanding protein structure and various approaches for predicting protein structures.

CO4: Acquire a comprehensive understanding of protein chip arrays, functional Proteomics, docking, drug design strategies, and Pharmacogenomics.

CO5: Equipped with a fundamental understanding of Bioinformatics, online and offline tools, biological databases, and sequence searching techniques.

#### Learning Outcomes:

LO1: Gain knowledge on concepts of genetic mapping and physical mapping techniques.

LO2: Understand the principles and applications of Biochips and Biosensors and DNA microarrays

LO3: Obtain knowledge on the protein structure and its determination methods

LO4: Get deep insight into docking, drug design methods and Pharmacogenomics.

LO5: Get knowledge on types of databases and database searching.

#### Course Specific Outcomes:

CSO1. Acquire fundamental knowledge in Genomics, Proteomics and Bioinformatics.

CSO2. Understand the concepts and approaches involved in different methods of drug designing

CSO3. Gain working knowledge of the Biological databases, tools and methods and appreciate their relevance for investigating specific contemporary biological questions

CSO4. Critically analyze and interpret results of the study

#### CO-PO Mapping

CO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PO8
CO1	✓	✓	✓	✓	✓			
CO2	✓	✓	✓	✓	✓			
CO3	✓	✓	✓	✓	✓			
CO4	✓	✓	✓	✓	✓			
CO5	✓	✓	✓	✓	✓		✓	

## **Unit – I**

Introduction to Genomics; Genetic mapping and Physical mapping techniques-Restriction mapping (fingerprint mapping and optical mapping); Fluorescent in situ hybridization mapping; DNA Markers (STS, RFLP, CAPS RAPDs, SNPs, AFLP) for genetic mapping; Human Genome Project.

## **Unit – II**

DNA sequencing – Maxam - Gilbert and Dideoxy methods; Shot gun method; Cycle sequencing; Automated DNA sequencing; Next Generation Sequencing; Biochips; Biosensors; 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 modelling of proteins

## **Unit – IV**

Protein chip arrays; Functional Proteomics; Docking; Drug design- Computer Aided Drug Design; Ligand-Based Drug Design; and Structure-Based Drug Design; Pharmacogenomics

## **Unit V**

Bioinformatics – Online and offline tools; Biological databases; Types of data bases – GenBank, Swiss- prot, EMBL, NCBI, and PDB; Database searching using BLAST and FASTA

### **Books suggested:**

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
3. “Bioinformatics: A practical Guide to the analysis of genes and proteins” by Andreas D. Baxevanis, B.F Francis Ouellette, 3<sup>rd</sup> edition.
4. “Bioinformatics and functional Genomics” by Jonathan Pevsner, 2<sup>nd</sup> edition.
5. “Bioinformatics: Sequence and Genome Analysis” by David W. Mount, 2<sup>nd</sup> edition.

## **SEMESTER-IV**

### **Paper 4.3: Practical –VII**

#### **Course Outcomes:**

CO1: Students will understand the principles and methodologies of genetic counseling and its importance in healthcare.

CO2: Familiarize with prenatal and postnatal screening methods for identifying genetic disorders.

CO3: Can evaluate the ethical considerations involved in genetic counseling and adhere to professional guidelines.

CO4: Able to navigate and utilize Genome Mapviewer from NCBI for genomic data exploration.

CO5: Can access, visualize, and analyze structural data of proteins from databases.

#### **Learning Outcomes:**

LO1: Analyze inheritance patterns and predict the risk of genetic disorders in families.

LO2: Demonstrate proficiency in genetic counseling techniques for single gene disorders through case studies.

LO3: Predict gene structures and annotate genomic features accurately.

LO4: Identify Open Reading Frames and understand their functional significance.

LO5: Predict protein secondary structures using computational methods for structural biology research.

#### **Course specific Outcomes:**

CSO1: Cultivate a deep understanding of genetic counseling principles and methodologies, equipping students for clinical practice.

CSO2: Develop computational biology skills for genomic data analysis and structural biology research.

CSO3: Gain hands-on experience in computational biology techniques and tools for genomic research and analysis.

**Part – A**  
(Assignment)

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

**Part – B**

1. Gene structure prediction
2. Modelling of a protein
3. Molecular docking by Molegro Virtual Docker
4. Detecting Open Reading Frames
5. Obtaining, viewing and analyzing structural data of proteins.
6. Identifying the protein through database by using gene sequence
7. A brief visit to Ensembl database
8. Microarray data analysis.
9. Secondary structure prediction of protein.
10. Genome Mapviewer from NCBI